PREDICTIVE TESTING AND CLINICAL GENETIC COUNSELLING SERVICES FOR HUNTINGTON DISEASE IN THE WESTERN CAPE: AN EVALUATION OVER ELEVEN YEARS

Thesis submitted in fulfilment of the requirement for the degree of Doctor of Philosophy in the Division of Human Genetics, Faculty of Health Sciences,
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DECLARATION

I, Merle Futter, hereby declare that the work on which this dissertation is based is my original work (except where acknowledgements indicate otherwise) and that neither the whole work nor any part of it has been, is being, or is to be submitted for another degree in this or any other university

Signature: Signed by candidate ....

Date: ....

Map of the Huntington disease research project in the Western Cape, South Africa

TABLE OF CONTENTS

Page

Chapter One: Introduction and orientation to the study

Chapter Two: Literature review

Chapter Three: Methodology

Page

Chapter Four: Findings and Discussion

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ABSTRACT

Predictive testing for Huntington disease, by means of direct mutation analysis, has been offered as part of a genetic counselling programme by the Division of Human Genetics from 1995. The neurogenetic clinic was started in the early 1980s specifically for patients with neurogenetic disorders, rather than them having to attend the general neurology outpatients clinics. It was thus opportune for a formal evaluation of these services to be conducted from the users' perspective and that it should include a broad social profile of the three user-groups, namely, the individuals of the predictive testing programme, patients from the neurogenetic clinic and their carers.

AIMS

The aims of this research were to:

- Evaluate the effectiveness of the genetic counselling processes of the predictive testing programme and neurogenetic clinic from the users' perspectives;
- Identify the components and determinants which ensure that the services are evaluated positively;
- Where possible, to implement changes and additions to these services as recommended by the users;
- Explore the nature and extent of the social barriers experienced by the users of the predictive testing programme and neurogenetic clinic; and
- Improve the quality of life of the families of patients by assisting them with the management of their affected relatives and providing them with psychological support.

METHODOLOGY

The research was a phenomenological cross-sectional design using a 'multi-method' approach of both qualitative and quantitative methods. As it entailed an

evaluation of the predictive testing programme and neurogenetic clinic genetic counselling services from the users' perspectives, a phenomenological design was the most appropriate method to study these aspects.

The three groups of participants comprised:

- all individuals (36) who had undergone the predictive testing programme from 1995 to 2005;
- all patients (41) who had attended the Groote Schuur Hospital Neurogenetic Clinic over the same period; and
- all main caregivers involved in the care of their affected relatives (38).

The study was conducted in the homes of the participants as the real meaning of their quality of life is experienced in their home setting and is, therefore, best investigated within this social structure frame of reference.

A semi-structured interview schedule comprising closed- and open-ended questions was developed as the measuring tool to gather quantitative and qualitative data. It included scales measuring the knowledge level; Shiloh et al's satisfaction with the genetic counselling services; the Stanford Health Assessment Questionnaire Scale; the Hospital Anxiety and Depression Scale; level of social problems; Social Support profile; and caregiver burden scale. The scales had all been tested for reliability and validity. Audio recordings were done of all interviews. They were transcribed and the data were captured on Excel spreadsheets. Analysis of the quantitative data was done by various interdependence and scaling techniques using SPSS, a software package for data management and statistical analysis. Open-ended questions were analysed by means of a process of constant comparative analysis. Simple frequency tables were used to categorise the responses into descriptive statistics such as percentages, means and modes to summarise various aspects of the interviews.

FINDINGS

The majority of participants rated most of the predictive testing genetic counselling processes as being 'satisfactory'. Their level of knowledge of Huntington disease was excellent. They were largely from a superior educational and socio-economic background similar to those of international studies and thus experienced few social barriers. The determinants of satisfaction were 'the quality of information' provided and the 'amount of empathy' displayed by the genetic counselling team members. The patients' level of satisfaction with all of the neurogenetic counselling processes was rated as 'very satisfactory' or 'highly satisfactory'. The determinants of satisfaction with this counselling service were 'amount of caring', 'empathy and concern displayed by the staff'. The majority of the participants came from socially disadvantaged backgrounds with a low level of education and low socio-economic background causing them to suffer from many of the social barriers associated with poverty of which lack of money, transport and housing were the most distressing. Their level of knowledge of Huntington disease and its pattern of inheritance was poor and frequently confused with other medical conditions such as tuberculosis and HIV/AIDS. The medical model is ineffective for improving the quality of life of patients with Huntington disease or their families as there is no cure or treatment. What is needed is a social model of health care where these families become the responsibility of the departments of social services, housing, transport, roads and sewerage, labour and education as it is in these domains that they experience the great difficulties rather than their symptoms.

Recommendations for improvements to both genetic counselling services were made by the participants, patients and the researcher. The majority has already been implemented and will require further evaluation after the improved services have been in operation for a year.

LIST OF TABLES

Page

LIST OF FIGURES

Page

LIST OF PHOTOGRAPHS

LIST OF APPENDICES

GLOSSARY AND ABBREVIATIONS

Akinesia: delayed initiation of movement or an inability to move.

ANC: African National Congress.

AOO: age of onset.

BIG: basic income grant.

Bradykinesia: increased time taken to execute movement.

Bruxism: grinding of teeth.

CAG: cytosine, adenine and guanine.

Carer group: the 38 carers who cared for their relatives with HD in the study.

CBS: Caregiver burden scale.

CNS: central nervous system.

CT: computed tomography.

DG: disability grant.

DPSA: Disabled People of South Africa.

DSSPA: Department of Social Services and Poverty Alleviation.

DTSS: Data Theory Scaling System Group, Psychology Department, Faculty of Social and Behavioural Sciences, Leiden University, The Netherlands.

Dyspnoea: shortness of breath.

EEG: electroencephalogram.

Freezing: a sudden cessation of a planned movement sequence, usually occurring during well learnt, automatic tasks such as walking or speaking (Morris, 2000).

GABA: gamma-aminobutyric acid which is a neurotransmitter.

Gliosis: excessive proliferation of the supporting structure of nervous tissue leading to scarring in the central nervous system.

GSH: Groote Schuur Hospital.

HAD: The hospital anxiety and depression scale.

HAQ: Health assessment questionnaire.

HD: Huntington disease.

HD gene: Huntington disease gene.

HDCRG: Huntington's Disease Collaborative Research Group.

HDQoL-C: Huntington disease quality of life battery for carers (Aubeeluck and Buchanan 2007).

ICIDH-2: International classification of functioning, disability, and health (short version).

INDS: Integrated national disability strategy.

Incidence: the number of new patients diagnosed with HD in South Africa within a year.

MOS: Social support profile.

MRC: Medical Research Council.

MRI: magnetic resonance imaging.

P: participant. In order to maintain confidentiality and anonymity only the coding numeral of the participants is provided. This code was known only to the researcher.

Patient group: the cohort of 41 patients from the neurogenetic clinic who formed part of the research.

PET: positron emission tomography.

PPC: perceived personal control.

Prevalence: the number of patients with HD in South Africa within a particular year.

Prosody: the variation in stress, pitch and rhythm of speech.

PT: predictive testing.

PT group: the cohort of 27 individuals who underwent predictive testing in the study.

RDP: Reconciliation and Development Plan.
SA: South Africa.

Saccadic eye movements: "an impaired ability to flick the eyes from side to side without undue blinks, delay or head movements" (Quarrel and Cook, 2004:224).

SPECT: single photon emission computed tomography.

SPSS: statistical package for the social sciences. A software package for data management and statistical analysis.

Statistics SA: Statistics South Africa Central Statistical Services.

UCT: University of Cape Town.

UK: United Kingdom.

UHDRS: Unified Huntington's disease rating scale.

USA: United States of America.

WC: Western Cape.

Wendy house: colloquial term for self-constructed informal house.

PREFACE

The terms Black, Coloured, Indian/Asian and White are used in the context which the official population census (Statistica SA), universities, provincial hospitals and corporations categorise students, patients and employees respectively to measure outcomes of affirmative action and Employment Equity put into place to address the political and socio-economic inequalities of the past in South Africa. 'Black' is used to refer to black Bantu-speaking Africans, 'Coloured' to people of mixed ancestry, 'Indian/Asian' to people of Asian descent and 'White' to descendants of European settlers. In this study individuals are classified according to self-perception and self-classification not on legal definition. Capitalisation of the words is used in recognition of a growing grass-roots sentiment that as distinct ethnic groups they deserve a capital letter (Crwys-Williams 1999).

For brevity 'patients' refer to individuals with symptoms of Huntington disease. It is used interchangeably with 'affected individuals' and 'people with Huntington disease'. Likewise 'carers' and 'caregivers' are used interchangeably.

For convenience and simplicity 'disabled' is used in general terms and includes handicap and impairment. It is used interchangeably with 'people with physical/mental disabilities'. These terms are accepted by the disability rights movement of South Africa (DPSA 2001). The Afrikaans-speaking participants refer to Huntington disease as 'sickness', hence this word is found in their quotations when they refer to their symptoms.

The terms gene-positive, mutation-positive, positive are used interchangeably to denote that the individual has CAG repeat lengths of ≥40. Likewise gene-negative, mutation-negative, negative are used interchangeably to denote that the individual does not have the mutated huntingtin gene.

Where descriptions of the living conditions of the patients would not suffice, photographs are provided to show the abysmal circumstances under which they live.

CHAPTER ONE

INTRODUCTION

1.1 INTRODUCTION

Huntington disease (HD) is a highly penetrant autosomal dominant neurodegenerative disorder characterised by a triad of symptoms. These are extrapyramidal motor dysfunction involving both involuntary and voluntary movements (Myers 2004); cognitive impairment; and affective disturbances and/or changes in personality (Shannon 2004; Sobel and Cowan 2003). Lund (1860) and Lyon (1863) were the first to publish cases of adult onset hereditary chorea. However, George Huntington is credited as being the first person to describe and document the salient clinical symptoms and pattern of vertical transmission of HD in 1872, after observing the disease in individuals in a large kindred in New York State (Shannon 2004; Quarrell and Cook; 2004; Okun 2003).

HD is an incurable disease and there are no preventive or effective treatments to delay the onset or reduce the rate of the degeneration of this usually late-onset disorder which leads to death 15 to 20 years after the manifestation of symptoms (Bonelli et al 2004; Myers 2004; Shannon 2004; Taylor 2004; SuttonBrown and Suchowersky 2003) regardless of the age of onset (Roos 1993). Although the prevalence is only approximately 10 per 100 000 persons in most populations, it is considered to be one of the most grave genetic disorders of adulthood because of its devastating effects on the individual as well as the family (Anderson and Marshall 2005; Ramos-Arroyo et al 2005; Quarrell and Cook 2004; Andrew et al; 1993; Harper 1996; Harper 1992; Hayden 1981).

In the Western Cape (WC) patients with HD attend the Groote Schuur Hospital (GSH) Neurogenetic Clinic for their management. This clinic is conducted once per month throughout the year and patients are given ongoing appointments as frequently as required to monitor their progress of symptoms, medication needs and to receive genetic counselling.

With the discovery of the HD gene in 1993 it became possible to predict, with a high degree of accuracy, which individuals are at-risk of having inherited the mutated gene by means of direct mutation analysis (Creighton et al 2003; The Huntington's Disease Collaborative Research Group 1993) (HDCRG). This has made it possible for them to reduce their uncertainty and anxiety about their future and to be able to plan their lives with regards to careers, relationships, procreation and different types of insurance (Evers-Kiebooms and Decruyenaere 1998; Decruyenaere et al 1996).

Once predictive test (PT) results are known to the individual, this knowledge is irreversible and can have significant adverse emotional and psychological consequences for the individual and family members, whether they are mutation-positive or mutation-negative. Research shows that 10% to 15% of individuals tested develop severe adverse reactions such as major depression and hospitalisation for psychiatric illness (Almqvist et al 1999; Taylor and Myers 1997; Lawson et al 1996; Huggins et al 1992). In order to ensure that individuals are optimally prepared to make an informed decision about the test options, understand the gravity of knowing their test result and are ready to undergo testing, most testing centres offer PT genetic counselling programmes in an attempt to reduce the possibility of adverse emotional events from developing (Blain and Brooks 2007; Myers 2004; Sarangi et al 2004; Shannon 2004; Bundey 1997).

A genetic counselling service for PT for HD has been offered by the Division of Human Genetics, University of Cape Town (UCT) since February 1995 (Appendix i). It consists of four counselling sessions provided by the genetic counselling team comprising a molecular geneticist, neurologist, psychologist and genetic nursing sister. The sessions are spaced at four-weekly intervals to allow the individuals ample time to make informed decisions regarding their options for testing and to reflect on their possible reactions to the outcome of their testing.

1.2 SIGNIFICANCE OF THE STUDY

Genetic counselling services have frequently been evaluated by measures of knowledge acquisition, risk comprehension, recall of genetic information, psychological outcomes and reproductive decisions (Wang et al 2004; Pilnick and Dingwall 2001). However, the most appropriate way of ensuring that the service does not have shortcomings with regard to the way in which the counselling is provided, is to evaluate the counselling process itself to ensure that it is meeting client needs (Aktan-Collan et al 2000; Bernhardt et al 2000). This is particularly so for conditions such as HD where there is valid PT available, but no effective treatment.

The UCT PT programme has been in operation for ten years and the genetic team has made incremental improvements to the programme periodically as it became aware of aspects that needed upgrading to improve the genetic counselling service. The same has taken place in the neurogenetic clinic which was started in the early 1980s specifically for patients with neurogenetic disorders rather than them having to attend the general neurology outpatient clinics. It was decided that it was opportune for a formal evaluation of these genetic counselling services to be conducted from the users' perspective and that this should include a broad social profile of the three user-groups, namely, the individuals of the PT programme (PT group), patients from the neurogenetic clinic (patient group) and the carer group.

Extensive research has been done on different single aspects of HD such as psychological consequences of PT and predictors of adverse events after PT; behavioural symptoms, motor manifestations, age of onset, suicide rate, molecular genetics, treatment options, dementia, discrimination; and motor dysfunction associated with HD. There is also a large amount of literature on the psychosocial aspects of disability in general, but no information pertaining to low-income patients with HD and their families living in a developing county.

No research has been published which evaluates the genetic counselling services from the users' perspectives and provides a comprehensive profile of the individuals who have undergone PT and patients with HD within a socio-economic context. Thus the purpose of this study was to investigate whether the PT programme and neurogenetic clinic have been beneficial to the individuals and families involved and to provide an extensive profile of their socio-economic living circumstances. It evaluated the counselling process against the definition of genetic counselling by the National Society of Genetic Counsellors' Task Force (Resta et al 2006); evaluated how supportive and beneficial the users found the services; and recommended and implemented improvements to the counselling services currently being offered. By describing details of the living environment of the patients and their socio-economic problems it provides additional insight into the clinical, social and psychological complexities experienced by the majority of patients affected with HD and their families who reside in historically disadvantaged low-income suburbs in a developing country.

1.3 AIMS OF STUDY

The aims of this research were to:

- Evaluate the effectiveness of the genetic counselling processes of the PT programme and neurogenetic clinic from the users' perspectives;
- Identify the components and determinants which ensure that the genetic counselling services are evaluated positively;
- Where possible, to implement changes and additions to these genetic counselling services as recommended by the users;
- Explore the nature and extent of the social barriers experienced by the users of the PT programme and neurogenetic clinic; and

- Improve the quality of life of the families of patients by assisting them with the management of their affected relatives and providing them with psychological support.

1.4 OBJECTIVES OF STUDY

The objectives of the research were to:

- Compile a profile of the PT participants, patients and caregivers;
- Measure the level of knowledge of HD of the PT participants and patients;
- Measure the level of satisfaction with the PT programme and neurogenetic clinic genetic counselling services;
- Determine what recommendations the users made to improve the services;
- Measure the level of caregiver burden;
- Assess the general needs of the PT participants, patients and caregivers;
- Conduct a community profile of the suburbs in which the research took place;
- Teach the patients and their carers basic management skills to maintain their level of independence and reduce caregiver burden;
- Teach the carers about the nutrition, feeding and exercising needs of their affected relatives;
- Assess the needs of the patients and carers with regards to their entitled social grants they were not receiving;
- Ensure that their applications for social grants were processed;
- Determine the level of knowledge the participants had of HIV/AIDS; and
- Increase their awareness and provide them with information on HIV/AIDS.

1.5 OUTLINE OF RESEARCH DESIGN AND METHODOLOGY

A phenomenological cross-sectional design using a 'multi-method' approach of both qualitative and quantitative methods of research was used for the research. A community profile was compiled of the low-income suburbs in which the majority of patients resided. If research is conducted in a community, in order for it to be successful, it is extremely important for the researcher to be familiar with the community itself as well as its environment (Katzenellenbogen et al 1997; Helander 1993; Bergner 1989). All the PT participants, patients attending the neurogenetic clinic and carers of the patients were included in the study and were interviewed. These interviews were conducted in the PT participants' and patients' homes. Follow-up interviews took place to obtain further data if the contents of the interview schedule had not been completed; as a form of assistance to participants needing psychological support; confirm that the transcriptions reflected the actual meanings the participants intended to portray; ensure that the social grants to which the patients were entitled had been processed; and to provide the carers with management skills to aid in the care of their affected relatives.

1.6 ORGANISATION OF THE STUDY

A literature review is presented in Chapter Two on various aspects of HD; PT; clinical manifestations and management; psychosocial aspects of disability and coping. The social and medical models of analysing disease and disability are described. Problems which affect the disabled in their daily lives are described in terms of employment, poverty, gender, race, discrimination, independence, self-image, abuse and powerlessness. Recent legislation and policies in South Africa, which should have an impact on these aspects when they come into effect, are listed and discussed briefly. A criticism of the inadequacies of disability grants (DG) in South Africa under the umbrella of "social assistance" is provided. The caregiving role is discussed in relation to the cost-saving of government funds that occurs when caregivers are family members. The cost to the caregiver with regard to not being able to work and the stress of decreased income and increased physical work is presented in terms of general strain, isolation, disappointment and emotional involvement.

In view of the HIV/AIDS pandemic in South Africa it would have been indefensible not to make use of the opportunity of increasing the awareness of the participants and their families about HIV/AIDS. This was a crucial aspect of their lives as the Ministry for Welfare and Population Development (1997) projected that the epidemic would grow exponentially to 2010 when between 18% and 27% of the population are likely to be infected.

In Chapter Three the methodology is presented in terms of research design, study population and setting; and measurement scales are described in terms of appropriateness, validity/trustworthiness and reliability. A brief description of the data gathering and statistical analysis is provided. Ethical considerations are discussed in relation to PT and the weaknesses of the study are identified. The process of the entire research study is also described.

The presentation of the analysis, findings and discussion are intermixed in Chapter Four as is customary in qualitative research (McMillan and Schumacher 2001) and subtitles connote the different findings. This facilitates the description and prevents unnecessary repetition of information which separate chapters for each component would require.

The conclusion in Chapter Five summarises the main findings of the research.

Recommendations as a result of the outcome of the study are discussed in Chapter Six. Where the recommendations have already been implemented they are noted.

CHAPTER TWO

LITERATURE REVIEW

2.1 INTRODUCTION

As the subject matter comprised various aspects of HD, the literature overview is extensive and deals with the history of the people of the Western Cape; genetics; neuropathology; clinical manifestations; management of the disease; psychosocial impact of HD on the individual and the family; models of disability; coping strategies; PT; genetic counselling; the evaluation of the effectiveness of genetic and clinical genetic counselling services; and caregiving.

Literature searches were performed using Portal; PubMed; CINAHL; EBSCOhost; Ovid; WebSPIRS; Sabinet and Google Scholar research databases; the African studies library, UCT; and the Faculty of Health Sciences, UCT library to obtain unpublished literature on HD. Search terms included were "Huntington disease", "PT", "genetic counselling", "programme evaluation", "adverse outcomes", "uptake", "clinical manifestations", "age of onset", "psychological affects", "disability", "caregiver", "burden of caring", "HD and substance abuse", "HD and violence" and "HD management". The literature discussed in this chapter mostly refers to research conducted in developed countries, such as the UK, Europe, Canada and USA as there is a dearth of local publications on research conducted in developing countries.

2.2 HISTORY OF THE PEOPLE OF THE WESTERN CAPE (WC)

As it is essential to be familiar with the pedigree of subjects in a genetic research programme a short description is provided of the original people, early settlers and the migration that occurred to and within the WC contributing to the present ethnical distribution of HD in the population. Because the vast majority of the participants affected with HD comprise people of mixed ancestry a brief history is included of the policies and legislation that contributed to their present demographic and socioeconomic problems.

The WC is situated south of the Orange River (Frontispiece) and has been populated for at least a million years (Bank et al 2003). The first people to inhabit the area were the San, previously known as Bushmen, Khoikoi previously called Hottentots and Strandlopers (Barnard 2004; Thompson 2001).

Bartholomew Diaz, a Portuguese explorer, discovered the Cape of Good Hope in 1487 and in 1652 it was settled by Europeans who originated from the Netherlands and Germany and later from France and Britain. They acquired slave labour from Dahomey, Angola, Madagascar, Mozambique, Indonesia, Ceylon, India and the East Indies and later migrant labourers from other parts of southern Africa including Mozambique, the Eastern Cape and Namibia (Banks et al 2003). Throughout the 'company' period there were marriages between European men and freed slave women and a great deal of extramarital sexual activity between white men, visiting sailors and slave and Khoisan women (Thompson 2001).

Thus the first people of mixed ancestry known as 'Coloureds' came into being as a result of the miscegenation between these early European settlers, Khoisan, slaves and African inhabitants previously from north of the Orange River (Adhikari 2005; Press 1990; Lever 1951; MacCrone 1951). According to the government census (Statistics SA 2001), at present Coloured people now comprise 54% of the population of the WC, followed by Blacks (27%), Whites (18%) and Indians (1%).

2.3 SEPARATE DEVELOPMENT AND DISCRIMINATION

Early legislation from 1828 ensured that there was no discrimination between people of different ethnic groups and when the Cape received aRepresentative Government in 1853 all persons of colour received the same rights of franchise as did White citizens. However, in 1857 social pressures caused the synod of the Dutch Reformed Church to authorise the separation of Coloured from White congregations and by 1861 Coloured children were effectively banned from public schools and those who went to school attended mission institutions. The promulgation of the School Board Act of 1905 enforced public schooling for White children only. Thus from the beginning of the twentieth century the White rulers treated the Coloured people as a distinct and inferior community dependent on White employers (Thompson 2001).

Segregation of the South African population into different racial groups and forced removals began in 1901 when African people had to live in *locations* on the outskirts of Cape Town such as Ndabeni, Nyanga and Langa (Bank et al 2003). These restrictions were later enforced by legislation passed which also prevented Black people from the free right to acquire land or live and work in the WC (African National Congress (ANC) 1998).

Although social racial segregation had been practiced since the country was first settled, legislation of discriminatory laws such as the Population Registration Act of 1950, Group Areas Act 41 of 1950, Prohibition of Mixed Marriages Act No 55 of 1949, Immorality Act of 1950 and The Reservation of Separate Amenities Act of 1953 was gradually introduced to ensure Whites did not live or mix socially with Blacks, Coloureds or Indians (Bank et al 2003; Thompson 2001; Lever 1951; Sinclair 1986). The laws, however, failed to stop people from different racial groups from 'mixing' as many people crossed the geographical and social lines and lived and cohabited in a few 'grey' areas which were 'mixed' areas which survived the Group Areas Act (Lever 1951).

2.4 INFERIOR STATUS ASSOCIATED WITH PEOPLE OF COLOUR

From the earliest centuries of the Common Era there were trade contacts between Arabia and East Africa which eventually led to wars and conquests by the victorious empires of Greece, Rome and the Near East. Thus most Blacks in these countries were enslaved. The noticeable different skin colour of the Africans compared to the lighter-skinned indigenous populations created a ready association of Black with "slave" as far back as the seventh century. Consequently from this time onwards the Curse of Ham, that is, the exegetical tie between blackness and servitude was established. Later as the Europeans conquered nations in the New World, Africa and Asia they perpetually dominated the indigenous peoples of these countries causing social status to be linked with physical difference. Consequently the foundation of racism was formed where people of darker complexion were dominated and exploited economically, politically, culturally and socially by people of lighter complexion (Goldenberg 2003).

Thus the original European settlers arrived in South Africa with preconceived notions that the indigenous Khoisan and the imported slaves were of inferior status and thus this racism became the foundation on which the 'apartheid' system was based (Lever 1951).

From the beginning of the 'apartheid' era (1948) cohesive and orderly communities were forcibly moved to new areas without consideration of the factors that had been the essence of their cohesion. Although the housing in the original areas frequently was far from ideal and there was gross overcrowding, the people living there provided a strong support system to each other in times of need. When they were forcibly removed to new areas on the periphery of cities and towns they were separated from each other and lost this support system (Adhikari 2005; Erasmus 2001). They were placed in inferior housing with higher rentals and the new residential areas were inaccessible, had limited facilities and transport was expensive (James 1995; Platsky and Walker 1985). The education, health care, transport, social services and security were all inferior to that of the Whites and affected their family life and community life in general (Erasmus 2001). The people who had owned properties in White areas were not paid out the true market value and this contributed substantially to their financial problems in their new homes (Adhirkari 2005; Van Der Horst and Reid 1981). Many of the Coloured people from leafy attractive suburbs which had all facilities close at hand were moved to unpleasant, flat, windy and sandy environments which were isolated from their social and working areas and which remained for several years without essential facilities such as schools, post offices, libraries, police stations, sports and recreational facilities (Erasmus 2001; James 1995).

All the factors mentioned above caused a vicious circle of poverty from which most Coloureds have been unable to escape even after the 'apartheid' laws were repealed and the new democratic ANC government came into power in 1994. Additional costs of higher rentals; transport to work, school and entertainment; higher food costs; meant that mothers frequently had to go out to work to increase the family income. As there was no support system in the new suburbs children were left unattended and many dropped out of school prematurely or joined gangs and became involved in drug abuse. Teenage pregnancy increased substantially due to the lack of parental supervision, lack of entertainment and sports facilities (Erasmus 2001; James 1996; James 1995).

The Coloured identity was associated with being a product of miscegenation and racial hybridity in a White society obsessed with racial purity. The hybridity led to the Coloured people as a group being assigned attributes of racial and intellectual inferiority and a stigma of illegitimacy as they were perceived to have originated largely from illicit sexual relations and extramarital unions across the colour line, although this was frequently not the case (Adhikari 2005).

The discriminatory laws were repealed in 1990 and 1991. Since then skilled and well-educated Coloured middle classes have profited from the extension of civil liberties as many of them have been able to take advantage of opportunities that have become available to formerly disadvantaged people. They have benefited by having the freedom of living where they wish, sending their children to schools of their choice and marrying whom they wish (Adhikari 2005). However, the Coloured communities living in the historically disadvantaged communities still suffer from the social consequences of poverty, unequal distribution of facilities, inferior housing and educational facilities (Naidu 2006; Spinks 2001; Van Der Berg and Burger 2002). These working classes perceive themselves as having gained little tangible benefits from the new dispensation as they consider themselves to be marginalised by affirmative action and black empowerment initiatives. The advantaged Black employers in the formal sector prefer to hire Blacks in order to have a more racially representative workforce and thus the employment opportunities for many Coloured people have shrunk (Adhikari 2005). Their perception of diminishing opportunities and standard of living since the early 1990s is confirmed by economists reporting on poverty headcount ratios and inability to pay for basic services such as water and electricity (Van Der Berg and Louw 2004; McDonald 2002; Spinks 2001). With the increase of unemployment there has been an increase in drug and alcohol abuse and increasing prevalence of gangsterism and criminal activity in the working-class Coloured residential areas. Crime statistics indicate that there has been a disproportionate increase in the homicide rate among Coloured people since 1990 and that recent studies claim that Coloured people are more than twice as likely to be murdered as people from other racial groups in South Africa (Naidu 2006; Leggett 2004a; Thomson 2004; Spinks 2001). It has become commonplace for the working-class and lower middle class Coloured people to express their disgruntlement with the new democratic environment by complaining that “first we were not white enough and now we are not black enough” (Adhikari 2005:176).

2.5 ORIGINS OF THE HUNTGINTON GENE MUTATION IN SOUTH AFRICA

In South Africa the mutant huntingtin gene is thought to have been introduced 350 years ago by a male of Dutch origin. The progeny of this individual spread the disease throughout the Cape and then the rest of South Africa and was responsible for most of the HD in Afrikaner and families of Mixed Ancestry (Hayden et al 1980b). In the most recent study of HD amongst the Black population it appears that there are many different geographical and ethnically distinct groups of African people with the disease, suggesting that there is more than one origin for the disease for this population group. However, these ten families identified to date, originated and live in provinces other than the WC (Silber et al 1998; Kremer et al 1994; Hayden 1980a).

2.6 GENETICS

2.6.1 PREVALENCE

The occurrence of HD varies between countries, geographical regions within countries and even between racial groups within countries.

The prevalence rate in the USA and most of Western European descent is 4 to 10 cases per 100 000 individuals (Evers-Kiebooms et al 2002; Fletcher 2001; Hayden and Kremer 2001; Morrison et al 1995; Harper and Newcombe 1992; Folstein et al 1987) with small pockets of much higher prevalence (Margolis and Ross 2003). The incidence is much lower in Japan, China, Finland and black South Africans (Shannon 2004; Evers-Kiebooms et al 2002; Hayden and Kremer 2001).

In South Africa the prevalence is approximately 26.5 per million in the White and 22.9 per million in the Mixed Ancestry population subgroups in the WC. Although there have been no data compiled on the prevalence it is thought that the Black African population subgroup may be as much as 40 times less prevalent or only 0.01 per million (Hayden 1981; Hayden et al 1980a; Hayden et al 1980b). However, this may be due to an underestimation because of the limited number of rural communities in the country which have accessibility to genetic services (Christianson and Modell 2004; Silber et al 1998). There is a high incidence of juvenile onset HD in some families of Mixed Ancestry (Hayden and Beighton 1977) where the father is the affected parent 3-4 times more frequently than the mother (Hayden 1981). A South African survey showed that 15.7% of patients from Mixed Ancestry had juvenile-onset disease compared to 4% in White South Africans (Hayden et al 1982).

Since direct assessment of the CAG repeat length in the huntingtin gene has become established, numerous new families with HD have been identified as the disease can now easily be accurately confirmed when clinical diagnosis is difficult (Hayden and Kremer 2001). Thus this has affected the prevalence rates and they are generally higher than what previous studies have found (Hayden and Kremer 2001; Falush et al 2000; Kremer et al 1994; Goldberg et al 1993). Although it was thought that PT may reduce the prevalence of the disease it cannot eliminate it altogether due to the substantial new mutation rate of ≥ 10% (Ramos-Arroyo 2005; Myers 2004; Margolis and Ross 2003; Moskowitz and Marder 2001; Falush et al 2000) arising from intermediate alleles, misdiagnosis of an affected parent, adoption and false paternity (Bradley et al 2004; Margolis and Ross 2003; Hayden and Kremer 2001) and the generally low uptake of PT (Langbehn et al 2004; Myers 2004; Taylor 2004). Many new mutations cause mild phenotypes and may escape detection for several generations (Bradley et al 2004; Hayden and Kremer 2001; Falush et al 2000).

2.6.2 INDIVIDUALS AT-RISK

Because HD is an autosomal dominant disorder each child born to an individual with the Huntington gene mutation is at 50% risk of carrying the mutated gene (Quarrell and Cook 2004; Fletcher 2001; Hayden and Kremer 2001; Horowitz et al 2001).

More than 98% of mutation-positive individuals will develop the disease at some stage of their lives (Myers 2004; Brouwer-DudokdeWit et al 2002; Kirkwood et al 2000) unless they die of trauma or an unrelated illness before the onset (Myers 2004; Duisterhof et al 2001; Houlihan 1999).

As the age at onset range is so extensive those at 50% and 25% risks are never clear of the fear of developing HD some time during their lives (Langbehn et al 2004; Myers 2004; Shannon 2004; Margolis and Ross 2003; Tyler 1996). This apprehension frequently leads to "symptom-watching" as individuals become increasingly focused on observing themselves and other members in the family for symptoms of the disease that might be developing (Folstein 1989).

2.6.3 CYTOSINE, ADENINE AND GUANINE REPEAT SEQUENCE (CAG)

HD results from a pathological expansion of a trinucleotide (CAG) repeat sequence (Shannon 2004; Quinn 2003). The abnormal *huntingtin* protein, therefore, contains an expanded polyglutamine tract (Spires and Hannan 2005; Quarrel and Cook 2004; Subramony and Filla 2001). It is is found within the coding region of the tip of chromosome 4p16.3 (Shannon 2004; Decruyenaere et al 1996; HDCRG 1993). The unstable repeat in exon 1 of the huntingtin gene was first isolated in 1993 (Myers 2004; Margolis and Ross 2003). *Huntingtin* is expressed throughout the body with especially high concentrations in the brain, testis, ovaries and lungs (Quarrel and Cook 2004; SuttonBrown and Suchowersky 2003; Hankey and Wardlaw 2002).

The CAG triplet repeat number is highly polymorphic in the population (Hayden and Kremer 2001; Rubensztein et al 1996) and is repeated approximately 10-35 times in the normal range (Spires and Hannan 2005; Harper 2004; Fletcher 2001) and 36 and above in the affected range

(Quarrell et al 2007; Semaka et al 2006; Hayden and Kremer 2001; Falush et al 2000). Normal alleles have repeats of 26 or fewer and are transmitted stably during meiosis (Mueller and Young 2001; Rubinsztein et al 1996) while all individuals with repeats of 40 to 240 will develop the clinical symptoms of HD, some time during their lives, provided they do not die from some other cause before the onset of symptoms (Semaka et al 2006; Spires and Hannan 2005; Myers 2004; Shannon 2004; Duisterhof et al 2001).

Intermediate numbers of repeats, between 27 and 35, are rare (1.5%) and are not associated with phenotypic expression in these individuals (Mueller and Young 2001; Harper et al 2000; Rubinsztein et al 1996). However, they can be meiotically unstable during intergenerational transmission and increase or decrease in size through either the male or female germlines (Falush et al 2000; Margolis and Ross 2003). If there is an expansion of the mutation during vertical transmission in this range there is a small increased risk of disease in the offspring (Creighton et al 2003; Falush et al 2000). Because repeat instability of this mutation is much more common in spermatogeneis than in oogenesis it occurs mostly in paternal transmission (≤10%), where the descendents have substantially greater CAG repeat lengths than their fathers and they develop HD. Expanded intermediate alleles are also more likely to be transmitted by males of advanced paternal age (mean 36.7 years) (Goldberg et al 1993). The risk of maternal expansion is very low and there are no documented cases of maternal intermediate allele expansion leading to an affected offspring (Myers 2004; Potter et al 2004; Shannon 2004; Fletcher 2001; Hayden and Kremer 2001; Chong et al 1997; Kremer et al 1995; Ranen et al 1995).

Alleles with 36 – 39 repeats are in the reduced-penetrance range. They are also rare and are considered expanded (Shannon 2004; Fletcher 2001; Rubinsztein et al 1996). As the penetrance is reduced it results in some individuals being affected and others who do not develop symptoms (Quarrell et al 2007; Langbehn et al 2004; Myers 2004). However, the risk of expansion into the disease range (≥36 CAG repeats) within one generation to offspring remains (Semaka et al 2006; Tassicker et al 2006). At present there are no reliable methods for estimating what proportion of these individuals will actually develop the disease, but those who do become symptomatic will have a milder phenotype than individuals with 42 repeats and they are also less likely to have affected relatives with larger expansions (Mueller and Young 2001; Falush et al 2000). Quarrell et al (2007) suggest that, in PT, if individuals are found to have a CAG repeat size of 36 to 39 there is at least a 40% probability of them being asymptomatic at age of 65 years and at least a 30% probability of them being asymptomatic at age 75 years. The disease is 100% penetrant in individuals with ≥40 repeats (Falush et al 2000).

2.6.4 Anticipation

"Anticipation is the occurrence of a genetic disorder at earlier age of onset and/or at greater severity in successive generations" (Harper 2004:381) and is caused by meiotic instability (Cleary and Pearson 2005; Zűlke et al 1993).

The tendency towards larger repeat sizes over generations explains why the age of onset tends to become progressively earlier and the symptoms more severe in successive generations (Wichman and Delong 2004; SuttonBrown and Suchowersky 2003; Fletcher 2001; Falush et al 2000; Kehoe et al 1999; McInnes 1996; Rubensztein et al 1996; Ranen et al 1995; Duyao et al 1993; Snell et al 1993).

2.6.5 **Age at onset** (AOO)

There is a great variation in the AOO of HD (Harper 2004; Myers 2004) with a range of 2 to > 90 years (Langbehn et al 2004; Myers 2004; Li et al 2003; Margolis and Ross 2003; Evers-Kiebooms et al 2002; Decruyenaere et al 1996; Roos et al 1991). The most significant factor to affect the AOO is the length of the expanded CAG repeat (Margolis and Ross 2003; Soliveri et al 2002; Kehoe et al 1999; Duyao et al 1993; Stine et al 1993). It contributes up to ≈73% of the variation in AOO of HD (Spires and Hannan 2005; Shannon 2004;Li et al 2003; Brinkman et al 1997; Andrew et al 1993; Duyao et al 1993; Snell et al 1993). The size of the triplet repeat is inversely associated with variation in AOO (Quarrell et al 2007; Langbehn et al 2004; Shannon 2004; Li et al 2003; Kirkwood et al 2000; Brinkman et al 1997; Andrew et al 1993; Duyao et al 1993; Snell et al 1993) and occurs regardless of the sex of the affected parent (Kehoe et al 1999; Andrew et al 1993; Duyao et al 1993; Snell et al 1993).

The size of the CAG repeat on the normal chromosome from the normal parent also influences the AOO. There is a significant association with OA and repeat length on the normal allele when it is paternally transmitted (Kehoe et al 1999; Harper 1996; Rubensztein et al 1996; Andrew et al 1993; Duyao et al 1993; Snell et al 1993). The negative association in maternal transmission is present, but not of a significant level (Kehoe et al 1999). The ratio of pathological to normal CAG repeat lengths also influences AOO. The greater the difference between the two repeat lengths, the earlier AOO is in paternally, but not maternally transmitted HD genes and in female offspring, but not in males (Kehoe et al 1999).

Heritability is an important contributory factor in AOO and parental AOO has been found to have a strong influence (Ranen et al 1995; Trottier et al 1994; Duyao et al 1993; Snell et al 1993). In general, the AOO for immediate family members tends to be similar, but no specific predictions can be made for a given individual (SuttonBrown and Suchowersky 2003). There may be substantial differences in AOO between parents and children with similar repeat lengths and there may be similar AOO between relatives who have a wide range of repeat lengths (Moskowitz and Marder 2001).

Siblingship accounts for 11 – 19% of the variance in AOO in addition to similarity of the CAG repeat length (Rosenblatt et al 2001; Squitieri et al 2000). In general, there is similarity in AOO among siblings and less correlation of the AOO between the parent and offspring (Hayden and Kremer 2001). In general, affected siblings have significantly similar AOO independent of the length of CAG repeat (Djoussé et al 2003; Rosenblatt et al 2001).

AOO in individuals with identical repeat lengths can often be different, not only because of genetic modifiers, but also due to environmental modifiers such as motor, social and cognitive stimulation (Spires and Hannan 2005; Wexler et al 2004). Modifiers appear to have a greater influence when the CAG repeat length is less than 44 as opposed to longer CAG repeat length (Langbehn et al 2004).

2.7 NEUROPATHOLOGY

The expanded polyglutamine tract in mutant *huntingtin* causes neuronal cell death and concurrent gliosis largely in the caudate and putamen nuclei of the basal ganglia and to a lesser extent in the globus pallidus, cortex, brainstem and cerebellum (Jacobs et al 2003; Margolis and Ross 2003; Hankey and Wardlaw 2002; Fletcher 2001; Hayden and Kremer 2001; Myers et al 1991).

Huntingtin is found in the cytoplasm of neurons (Reddy et al 1999). Wild-type *huntingtin* has a neuroprotective role and is fundamental to cell survival during development (Van Raamsdonk et al 2005; Landles and Bates 2004; Reiner et al 2003; SuttonBrown and Suchowersky 2003) and in adult life (Cattaneo et al 2001). The genetic loss of wild-type *huntingtin* expression in individuals with HD reduces the ability of neurons to survive the toxic effects of mutant *huntingtin* and thereby significantly contributes to the pathogenesis of HD (Van Raamsdonk et al 2005).

The role and function that the expression of mutant *huntingtin* plays in brain pathology of HD is not known (Mueller and Young 2001) although there are several different postulations about the effects that the HD mutation causes. It is possible that the inclusions are directly related to the basic pathology as a defense mechanism by the cell (Quarrell and Cook 2004; SuttonBrown and Suchowersky 2003). Another postulation is that there is a dominant-negative effect where the mutant *huntingtin* interacts with the subunits of normal protein to inactivate their normal function (Hankey and Wardlaw 2002) by interfering with gene transcription leading to an alteration in cell phenotype (Quarrell and Cook 2004). This blocking of the normal function of *huntingtin* to upregulate brain-derived neurotrophic factor which is involved in neurotransmission, triggers apoptotic cell death (Shannon 2004; SuttonBrown and Suchowersky 2003; Young 2003; Mueller and Young 2001). The third theory claims that the CAG repeat mutation is translated into an abnormally long poyglutamine tract, which confers a toxic novel gain of function on the mutant protein (Ravikumar et al 2006; Squitieri et al 2003) together with the elimination of some of the functions of wild-type *huntingtin* (SuttonBrown and Suchowersky 2003; Hankey and Wardlaw 2002). According to the authors, whatever role *huntingtin* plays in the pathology of HD it causes a biochemical imbalance between gamma-aminobutyric acid (GABA), acetylcholine and dopamine which results in chorea (Hankey and Wardlaw 2002; Lindvall and Bjorkland 2000).

Recent research indicates that there is evidence of neurodevelopmental and extrastriatal brain abnormalities which contribute to the pathology of the disorder (Paulsen et al 2006; Rosas et al 2003) indicating an alteration in the morphogenesis of the brain (Gomez-Tortosa et al 2001). Paulsen et al (2006) have found that in presymptomatic individuals who were far-from-onset the volume of gray matter in the cerebral cortex is significantly increased and that of the white matter in the cerebral cortex, caudate, putamen and thalamus is significantly decreased. They postulate that the increased volume of cerebral cortex may be due to a predegenenerative process indicating apoptosis or due to abnormal brain development or maturation and the small volume of striatal tissue might be accounted for by maldevelopment or lack of growth and not degeneration of normal tissue. Individuals in their study who were close-to-onset showed the cerebral white matter was further reduced in volume and the caudate, putamen and thalamus were markedly reduced in volume. Thus presymptomatic individuals are vulnerable to maldevelopment of their brains and, later in their lives, to environmental stressors which trigger apoptosis (Paulsen et al 2006).

Macroscopic

Macroscopically early on there is atrophy of and regional loss of normal neurons in the white matter of the striatum and cerebral cortex (Hankey and Wardlaw 2002; Fletcher 2001; Myers et al 1991) with the presence of intraneuronal aggregates of misfolded *huntingtin* and ubiquitin (Hankey and Wardlaw 2002; Maat-Schieman et al 1999; DiFiglia et al 1997). Later in the disease it affects most of the gray matter of the cerebral hemispheres and cerebellum (Hankey and Wardlaw 2002) and the whole brain can appear atrophic with narrow gyri, widened sulci and a reduction in brain weight (Hayden and Kremer 2001).

Microscopic

The most significant cell loss occurs in the basal ganglia, especially in the caudate and putamen nuclei (Quarrell and Cook 2004). Neuronal loss in the caudate precedes neuronal loss in the putamen and ventral striatum (Vonsattel et al 1985). Striatal medium spiny neurons, which make up 90% of all striatal neurons, are selectively impaired (Quinn 2003; SuttonBrown and Suchowersky 2003; Hankey and Wardlaw 2002; Albin and Tagle 1995; Graveland et al 1985); whereas the large aspiny interneurons that are adjacent to the medium spiny neurons are preserved until late in the illness (Quarrell and Cook 2004; Hankey and Wardlaw 2002; Ferrante et al 1991). The mechanism for this regional selective neuronal loss is unknown (Fletcher 2001).

Striatal degeneration occurs in both the motor and associative portions of the structure. Some authors report that the earliest pathological changes are in the associative portions and that the first symptoms manifested by the disease are cognitive and psychiatric rather than motor (Askin-Edgar et al

2004; Quinn 2003; Squitieri et al 2003; Paulsen et al 2001b; Amann et al 2000; Kirkwood et al 2000; Middleton and Strick 2000; Di Maio et al 1993). Others claim that the motor portions degenerate first and that the caudate nucleus and striatal-cortical circuits contributing to the psychiatric and cognitive symptoms only degenerate at a late stage (Wichman and Delong 2004; Jacobs 2003; Zappacosta et al 1996). A third group of authors maintain that patients can present with either neurological or psychiatric signs (Langbehn et al 2004). The pattern of neuropsychological impairment which begins in the frontal lobe gradually spreads to involve all cortical regions (Hayden and Kremer 2001).

Neurochemical

The striatum receives excitatory neurons from the cortex and has efferent neurons containing the neurotransmitters GABA and substance P or GABA and metencephalin (Figure 1).

These neurons project to the internal globus pallidus and substantia nigra via direct and indirect pathways (Quarrell and Cook 2004). In the early stages of HD there is striatal degeneration of some of the GABA-ergic medium spiny neurons which predominate the indirect pathways. This lack of excitatory drive causes reduced inhibition of the output neurons of the external pallidus which leads to chorea (Quarrell and Cook 2004; Shannon 2004; Bilney et al 2003). In the later stages of the disease all GAGA-ergic medium spiny neurons, including the output neurons in the direct pathway, begin to degenerate resulting in increased inhibition of the thalamus (Quarrell and Cook 2004). This accounts for the late-stage reduction in hyperkinetic features of chorea and the development of hypokinetic features such as bradykinesia, rigidity, parkinsonism and tremor (Quarrell and Cook 2004; Shannon 2004; Albin et al 1990; Reiner et al 1988).

The nature of motor dysfunction that the individual exhibits depends on the extent of degeneration in each of the pathways (Quarrell and Cook 2004). It seems likely that both pathways degenerate at a similar rate, but that excess dopamine from the substantia nigra produces an inhibitory effect on the indirect pathways giving an apparent appearance of a different rate of degeneration (Hedreen and Folstein 1995). Juvenile-onset HD which is more severe from the beginning resembles late-stage HD with GABA-ergic neurons affected in both direct and indirect pathways (Shannon 2004).

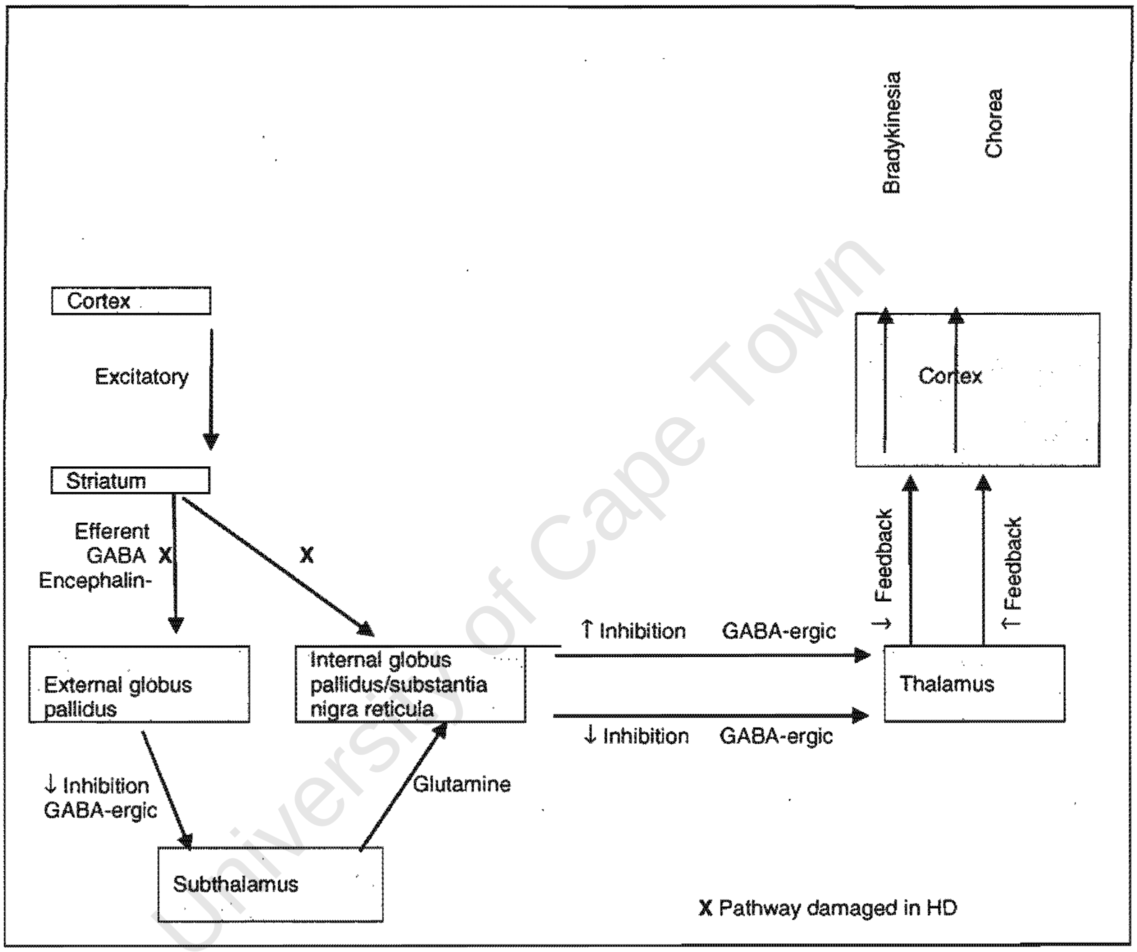

Figure 1: Connections of the basal ganglia, showing the pathways damaged in Huntington disease (HD) (Adapted from Quarrell and Cook 2004)

Delayed initiation (akinesia) and other abnormalities of saccades may result from overinhibition of neurons in the tectum mesencephali. This overinhibition is caused by the action of GABA-ergic afferent neurons from the substantia nigra that are normally inhibited by striatal GABA-substance P-containing projections. This class of neurons seems to degenerate earlier than the GABA-substance P neurons to the internal globus pallidus, and may be affected as early as the GABA-encephalin (Enk) projections to the external palladium, explaining the occurrence of saccadic abnormalities already being evident in the transitional period (Hayden and Kremer 2001).

Dysfunction of the basal ganglia interferes with the normal neural transmission to the supplementary motor area. These two sites are thought to play a role in well-learnt movement sequences (Cunnington et al 1995) and when the neural transmission is disrupted it interferes with the preparatory activity within the supplementary motor area to facilitate the next movement sequence of the task to be completed (Georgiou et al 1993). Disruption of these neural pathways also interferes with voluntary movements (Bilney et al 2003).

2.8 CLINICAL MANIFESTATIONS

Whatever the presenting symptoms, individuals often have difficulty in being able to be specific about the date of the onset and the type of initial symptoms because of its slow and insidious onset. They are frequently unaware or unconcerned about them (Jankovic and Lang 2004; Quarrell and Cook 2004; Shannon 2004; Hankey and Wardlaw 2002; Fletcher 2001; Hayden and Kremer 2001). The affective disturbances and deterioration of cognitive function are often first noticed by their families and are the most debilitating features of the disease and the most difficult and stressful aspects for the family to manage (Bonelli et al 2004; Paulsen and Hoth 2004; Quarrell and Cook 2004; Bilney et al 2003; Quinn 2003; Soliveri et al 2002; Hayden and Kremer 2001).

The CAG repeat length is inversely associated with severity of the clinical symptoms and with the extent of the neuropathology (Quarrell and Cook 2004; Creighton et al 2003; Hankey and Wardlaw 2002; Hayden and Kremer 2001; Kirkwood et al 2000). Elderly-onset (≥60 years) disease presentations are often associated with alleles between 36 and 41 (Shannon 2004; Mueller and Young 2001; Albin and Tagle 1995), have less severe symptoms and deteriorate slower with regards to overall neurological signs, motor impairment, activities of daily living and cognitive function than those with CAG repeats of 47 or more (Rosenblatt et al 2006; Myers 2004; Paulsen and Hoth 2004; Quarrell and Cook 2004; Hankey and Wardlaw 2002; Fletcher 2001; Hayden and Kremer 2001; Mueller and Young 2001; Nance and Myers 2001). Rate of progress is not related to the major clinical feature at onset or the sex of the affected parent (Mahant et al 2003).

The symptoms develop heterogeneously in each family. One member may exhibit more signs of movement dysfunction whereas another may have lack of emotional control. Moreover, the extent of the different symptoms of HD vary in the same individual over time (Quarrell et al 2007; Harper 2004; Myers 2004; Bachoud-Lévi et al 2000; Collins 1999). The disease progresses without any periods of remission (Li et al 2003) and most individuals are significantly affected after five years from the onset of symptoms to the extent that they are unable to continue working (Harper; 2004).

Individuals with HD can be classified into three broad phenotypes: 'classical' late-onset HD; juvenile or early-onset HD; and elderly-onset form from 60 years and older (Fletcher 2001). There is a period between the normal asymptomatic phase to the time at which a definitive diagnosis can be made on a neurological examination which is known as the transitional stage (Hayden and Kremer 2001; Penney et al 1990). The earliest signs are frequently a change in the accuracy or velocity of saccades, nystagmus, dysdiadochokinesia and mild dysarthria (Shannon 2004; Hayden and Kremer 2001; Kirkwood et al 2000). Individuals and their families describe clumsiness in common daily activities such as dropping and bumping into objects (Jankovic and Lang 2004; Hayden and Kremer 2001); general restlessness; and excessive and inappropriate movements of the fingers, hands or toes which become more evident and persistent during periods of emotional stress (Penney et al 1990; Folstein et al 1986; Young et al 1986); cognitive deficits of executive dysfunction and slower speed of movement and reaction times (Paulsen and Hoth 2004; Kirkwood et al 2000) The transitional phase lasts for approximately three years before the overt extrapyramidal dysfunction signs of chorea become evident (Hayden and Kremer 2001; Kirkwood et al 2000; Penney et al 1990; Folstein et al 1986).

2.9 MOTOR MANIFESTATIONS

Chorea: Chorea is the major motor symptom that is most often associated with HD and occurs in more than 90% of individuals with late-onset or elderly-onset HD (Quarrell and Cook 2004; Evers-Kiebooms et al 2002; Fletcher 2001; Hayden and Kremer 2001). It consists of purposeless, irregular, unpredictable, brief, jerky, involuntary movements which flit randomly from one part of the body to another and become more evident during activities such as gait (Jankovic and Lang 2004; Ovsiew 2004). Chorea may affect the muscles of the face, neck, trunk, pelvis and limbs and this causes difficulty with visual tracking, dysarthria, dysphagia and breathing (Hankey and Wardlaw 2002). It causes fine motor movements to be slow, incoordinated and arrhythmic (Fletcher 2001; Ross et al 1997; Folstein 1989). Choreic movements are continuously present during waking hours, only worsening during periods of anxiety or stress. The individual is unable to suppress these movements voluntarily (SuttonBrown and Suchowersky 2003; Hayden and Kremer 2001), yet they may be the least disabling symptoms of the disease (Moskowitz and Marder 2001).

Individuals with chorea demonstrate relatively more hyperactive behaviours such as agitation, irritability, and anxiety (Litvan et al 1998).

Bradykinesia and Rigidity: The time taken to initiate movements (akinesia) and the overall time to execute movements (bradykinesia) is increased (Van Vugt et al 2004; Georgiou et al 1993). Frequently there is a coarse resting tremor associated with the bradykinesia (Folstein 1989; Bittenbender and Quadfasel 1962).

These symptoms gradually appear later in the illness until they often dominate the choreic movements in the final stages when the individual becomes severely rigid and grossly akinetic (Quarrel and Cook 2004; Quinn 2003; SuttonBrown and Suchowersky 2003; Young et al 1986; Bruyn 1968).

Dystonia: These are slow abnormal movements and abnormal posturing which are found sometimes in the early symptomatic period, but worsen and become a prominent feature in the later stages of the illness (Quarrell and Cook 2004; Shannon 2004; Quinn 2003; SuttonBrown and Suchowersky 2003; Fletcher 2001; Young et al 1986; Bittenbender and Quadfasel 1962).

Of particular significance is respiratory dyskinesia which affects the fluency of speech and may cause dyspnoea when walking or, eventually, at rest (Jankovic and Lang 2004).

Dysarthria: Most individuals with HD have dysarthria (Paulson and Hoth 2004; Hayden 1981) from early in the illness (Coleman et al 1990; Podoll et al 1988; Young et al 1986). Speech may be slurred, halting and periodically interrupted as there is insufficient breath support due to the choreic movements of the trunk. There is fluctuating prosody, an increased time delay in responses and mild misarticulations, but the syntactic structure and content of their speech is usually appropriate (Paulson and Hoth 2004).

There is frequently a reduction in spontaneity and quantity of speech output (Jankovic and Lang 2004). The inability to communicate is frustrating for the individuals as they are able to fully comprehend throughout most of the course of the disease, but are unable to respond (Quarrell and Cook 2004).

Dysphagia: This life-threatening complication generally occurs late in the progression of the disease. It affects both the chewing and the movement of the bolus of food to the back of the throat (Moskowitz and Mader 2001). Initially it affects the intake of fluids, but later also affects the intake of solids (Quarrel and Cook 2004; SuttonBrown and Suchowersky 2003; Hayden and Kremer 2001; Myers et al 1985). Swallowing difficulties are a common cause of choking with aspiration leading to episodes of pneumonia and/or death

(Jankovic and Lang 2004; Quarrel and Cook 2004; Shannon 2004; Fletcher 2001).

Gait: Gait disorders in HD are complex due to the varying influences of chorea, parkinsonism, fluctuations in tone of antigravity muscles and ataxia (Shannon 2004). Stance and gait disturbances can occur early in the disease. The gait is unsteady, uneven with a wide base to assist with stability, giving the impression of a drunken stagger (Hankey and Wardlaw 2002; Hayden and Kremer 2001). They have difficulty in initiating the first steps and 'freeze' during walking due to the effects of bradykinesia, akinesia (Nance and Myers 2001), segmental stiffness (Grimbergen et al 2004), postural instability (Bilney et al 2003) and motor impersistance (Pollard 2000). As their righting reflexes are delayed and they have impaired spatial awareness they frequently fall and sustain a significant number of injuries, particularly of the head as they cannot use their arms effectively in protective reactions (Grimbergen et al 2004; Moskowitz and Marder 2001; Tian et al 1991; Huttunen and Hömberg 1990; Horak et al 1989). As the disease progresses the episodes and duration of 'freezing' increase and the individual gradually becomes more and more immobile until there is a total inability to walk (Grimbergen et al 2004; Fletcher 2001) and eventually becomes wheelchair-bound (Askin-Edgar et al 2004; Grimbergen et al 2004; Jankovic and Lang 2004; Quarrel and Cook 2004; Shannon 2004; Bilney et al 2003).

Oculomotor dysfunction: Eye movements are slowed at an early stage and individuals have difficulty in initiating saccadic eye movements. Their pursuit movements are irregular and they have impairment of gaze fixation (Quarrell and Cook 2004; Jacobs et al 2003; Hankey and Wardlaw 2002; Fletcher 2001; Hayden and Kremer 2001).

Voluntary motor dysfunction: An early sign in HD is impairment of voluntary motor function as a result of the bradykinesia and akinesia rather than chorea (Van Vugt et al 2004; Bilney et al 2003; Thompson 1988; Hefter et al 1987). Functional ability and motor slowness is correlated with cognitive impairment rather than the effects of chorea (Van Vugt et al 2004; Moskowitz and Marder 2001).

Musculoskeletal and cardiovascular changes: Secondary musculoskeletal and cardiovascular changes occur as a result of cognitive and behavioural disorders, inactivity and disuse (Bilney et al 2003).

Reflexes: Hyperactive reflexes occur early in up to 90% of patients, while clonus and extensor plantar responses occur late and are less frequent (SuttonBrown and Suchowersky 2003; Young et al 1986; Hayden 1981).

2.10 COGNITIVE DISTURBANCES

The nature, sequence and severity of the impairment vary from person to person (Jacobs et al 2003). Before the definitive diagnosis is made there is often a history of failing work performance and retrenchment or bankruptcy if the person is self-employed. These are due to cognitive decline (LoGiudice and Hasset 2005). Generally, the first cognitive deficits are focal and appear early, but as the disease progresses most individuals have a global decline in cognitive capabilities (Shannon 2004; Hayden and Kremer 2001). The dementia that gradually develops is profound and is responsible for the majority of the morbidity associated with the disease (Bertelson and Price 2004; SuttonBrown and Suchowersky 2003).

There are conflicting claims of the aspects of cognition which are the most affected. Some authors claim that, once the individual has developed the symptoms of HD, the most prominent cognitive deficit is executive dysfunction. Initially the greatest changes occur in automatic speed-based tasks which have low cognitive demands such as routine tasks of dressing, eating, setting the table and washing. These activities take much longer to execute than usual (Moskowitz and Marder 2001) reflecting the greater changes that initially occur in the basal ganglia rather than the cortex (Snowden et al 2001). Individuals have difficulty in organising, sequencing, planning and initiating a thought process or activity. They are unable to switch from one plan or activity to another. This results in, amongst others, perseverative behaviours and diminished verbal fluency (Moskowitz and Marder 2001). They have a reduced problem-solving capacity; lack judgement, insight and motivation; and are unable to monitor self-behaviour (Quarrell and Cook 2004; Shannon 2004; Jacobs et al 2003; Hayden and Kremer 2001). They become inflexible and demand that their needs are attended to immediately and in a particular way (Pollard 2000).

However, other authors maintain that the most frequently reported cognitive complaints from affected individuals and their families are the acquisition of new information, delayed recall, lack of manual dexterity, decreased speed of processing information and impaired verbal skill (Paulsen and Hoth 2004; Hayden and Kremer 2001; Zakzanis 1998).

The individuals have this insight and are aware of their cognitive decline as the disease progresses and these aspects deteriorate (Hayden and Kremer 2001).

Memory

Memory deficits are common and appear early in the disease often as one of the individual's presenting problems (Caine and Fisher 1985). Individuals have difficulty in learning new information as they have impaired retention of new information and poor spontaneous recall. Memory retrieval is affected more than storage and patients may be able to respond by prompting (Lauterbach et al 1998). Retrieval of both short-term and long-term memory is equally impaired (Askin-Edgar et al 2004; Jacobs et al 2003; Hankey and Wardlaw 2002; Hayden and Kremer 2001; Massman et al 1990; Wilson et al 1987; Beatty et al 1988; Brandt and Butters 1986) and these impairments manifest as forgetfulness (Askin-Edgar 2004; Paulsen and Hoth 2004).

Attention and concentration

Attention and concentration are affected early in the disease resulting in individuals with these symptoms being unable to work, drive vehicles, manage family finances and have difficulty in shifting their attention from one task to a new one. This also entails having to relate differently to their family and friends (Paulsen and Hoth 2004; Quarrel and Cook 2004; Shannon 2004; Jacobs et al 2003; Hankey and Wardlaw 2002; Hayden and Kremer 2001; Pollard 2000; Phillon et al 1991).

Language and related functions

Verbal memory, semantic and syntactic structure, word finding and speech comprehension remain fairly well preserved until late in the disease and individuals are able to remember facts, words or stories for a relatively long period (Askin-Edgar et al 2004; Quarrel and Cook 2004; Jacobs et al 2003; SuttonBrown and Suchowersky 2003; Phillon et al 1991; Folstein 1989; Caine et al 1985). However, dysarthria, slowness and lack of initiative or apathy interfere significantly with their communication and with the fluency of spontaneous speech (Jacobs et al 2003; Wallesch and Fehrenbach 1988; Caine et al 1985), although they are able to comprehend throughout most of the course of the disease (Pollard 2000). Individuals may have difficulty in starting a conversation, staying on the topic and switching from one topic to another. They may also perseverate on one subject and have difficulty in moving onto another topic (Hayden and Kremer 2001; Pollard 2000).

Problem-solving

There is marked impairment of the ability to synthesise information and produce flexible strategies for problem solving (Askin-Edgar et al 2004). Individuals are unable to organise, plan and sequence information necessary to perform complex tasks which require components to be completed in a specific order (SuttonBrown and Suchowersky 2003; Caine et al 1978) such as routine activities of daily living.

2.11 PSYCHIATRIC DISTURBANCES

Although psychiatric disturbances are as characteristic of the disease as motor and cognitive abnormalities, they present less consistently and are not necessarily related to the severity of chorea or dementia present (Zappacosta et al 1996; Coleman et al 1990; Caine and Shoulson 1983). They are the most variable symptoms of the clinical manifestations of HD (Bonelli et al 2004; Margolis and Ross 2003; Witjes-Ané et al 2002; Webb and Trzepacz 1987; Caine and Shoulson 1983). There is no correlation between CAG repeat length and the extent or severity of psychiatric symptoms as these disorders progress nonlinearly (Soliveri et al 2002; Zappacosta et al 1996). However, affective disorders in offspring have been found to be highly associated with the presence of similar symptoms in the HD parent (Folstein et al 1983).

Approximately 80-98% of individuals with HD develop psychiatric disorders within 10-15 years of onset of disease (Leroi et al 2002; Paulsen et al 2001a; Moskowitz and Marder 2001; Sobel and Cowan 2000; Folstein 1989). However, behavioural and psychiatric disorders may predate the motor symptoms of HD by as long as a decade, reflecting early pathological changes in the nonmotor areas of the striatum and do not usually manifest for the first time late in the disease (Shannon 2004; Soliveri et al 2002; Morris 1991; Folstein 1989). Behavioural dysfunction can impede the ability to use motor or cognitive skills that are still present in the early stage of the disease and thus contributes significantly to functional disability (Hamilton et al 2003).

These psychiatric disturbances cause the relationships with family and friends to be negatively affected (Bilney et al 2003; Pollard 2000) and are most strongly associated with caregiver stress and the need to have the family member placed in a care centre (Paulsen and Nehl 2004).

Mood and affect

The most frequent psychiatric problems are apathy, depression, anxiety, agitation, suspicion, difficult to get along with, irritability, violent temper outbursts, aggression, obsessive-compulsive tendencies, sexual disorders, social disinhibition and long periods of depression. Symptoms such as delusions, hallucinations and mania occur less frequently (Marshall et al 2007; Askin-Edgar et al 2004; Bertelson and Price 2004; Shannon 2004; Soliveri et al 2002; Paulsen et al 2001a; De Marchi and Mennella 2000; Rosenblatt and Leroi 2000; Morris and Tyler 1991).

Depression

Depression occurs in 9-70% of patients and involves considerable emotional distress evidenced by tearfulness, sadness, anxiety, agitation, insomnia, anorexia, feelings of worthlessness and hopelessness, and recurrent thoughts of death (SuttonBrown and Suchowersky 2003; Hankey and Wardlaw 2002; Hayden and Kremer 2001; Morris and Scourfield 1996; Folstein et al 1983). Depression can precede the onset of neurological symptoms in HD by 2 to 20 years and is most common immediately before the diagnosis has been made and when neurological soft signs and other subtle abnormalities become evident. Following a definitive diagnosis of HD, however, depression is most prevalent in the middle stage of the disease when the individual loses independence and may diminish in the later stages (Paulsen and Nehl 2004; Quarrell and Cook 2004; Witjes-Ané et al 2002; Kirkwood et al 2000). There is conflicting evidence about whether the depression is secondary to pathological changes in the basal ganglia or whether it is a response to the debilitation that occurs during the progress of the disease; being part of a family in which there have been affected individuals during the childhood years of the individual; in anticipation of being at-risk; fear of the onset of disease; or fear of losing independence (Paulsen et al 2005; Witjes-Ané et al 2002; Paulsen et al 2001a; Wells and Warren 1998; Huntington Study Group 1996).

Behavioural disturbances

Sexual disinhibition, inappropriate social behaviour; paranoia and alcohol abuse are other symptoms frequently seen in HD patients (Mueller and Young 2001; Kirkwood et al 2000; Jensen et al 1998; Morris 1991) and, whilst these symptoms might not be perceived as problems by the individuals, they are difficult for the family to manage (Quarrell and Cook 2004). They may be either a sign of the progressive cognitive decline in the disease or a manifestation of the mood disturbances related to the premorbid personality. Alcohol abuse appears to be a greater problem in males and is probably linked to the personality changes of poor self control, impulsive behaviour, irritability and physical aggressiveness or secondary to depressive reactions or difficulties in coping with the symptoms (Jensen et al 1998).

Suicide and suicide ideation

The suicide rate of people with HD is estimated to be as high as 5% to 13%, which is higher than in patients with other medical and neurodegenerative diseases (Paulsen et al 2005; Askin-Edgar et al 2004; Bradley et al 2004; Bertelson and Price 2004; Paulsen and Nehl 2004; Druss and Pincus 2000; Harris and Barraclough 1997; Cummings 1995) and four times the rate in the general population (Paulsen et al 2001a; Meiser and Dunn 2000). It generally occurs around the time of the onset or early in the second stage of the disease and may be associated with depression (Askin-Edgar et al 2004; Hankey and Wardlaw 2002; Di Maio et al 1993). It is a significant cause of death of patients in the earlier stages (Askin-Edgar 2004; Farrer and Yu 1985). Depression paired with a lack of impulse control makes suicide a major risk for affected individuals in all stages of the disease (Pollard 2000).

Suicide ideation is highly prevalent throughout the disease, with 9.1% of all individuals diagnosed with HD having active ideation. Two primary "critical periods" are evident during which suicide ideation dramatically increases. The first is at the time at-risk persons develop soft neurological signs (19.8%) which is the time immediately before a formal diagnosis is made. Schoenfeld et al (1984) found that more than half of the suicides amongst individuals affected with HD were in individuals showing early symptoms of the disease. Paulsen et al (2005) claim that at the time the diagnosis is made suicidal ideation decreases, but that at the time immediately before this there is an increased risk of suicide when individuals have a greater uncertainty about whether they have the disease. They suggest that the frequency of suicide may actually be reduced by the confirmatory diagnosis of HD combined with appropriate treatment of depression. The second critical period is at the second stage of disease when the individual starts to lose independence (21%) such as when he/she is no longer able to work, drive or manage domestic finances and has to depend on others to take over these roles. Suicidal ideation diminishes after the second stage of disease (Paulsen et al 2005).

Psychosis

Psychosis occurs with increased frequency in HD, with estimates ranging from 3-25% and can manifest itself as hallucinations, delusions in the form of paranoia, or both and tends to abate as the dementia progresses (Askin-Edgar et al 2004; Bertelson and Price 2004; Paulsen and Nehl 2004).

Obsessive-Compulsive traits

Obsessive and compulsive behaviours are prevalent in 13-30% of individuals (Moskowitz and Marder 2001). Obsessional thinking often increases with proximity to disease onset and then remains somewhat stable throughout the illness. It is similar to perseveration as individuals get 'stuck' on a previous thought, activity or need and they are unable to shift their thought processes (Paulsen and Nehl 2004).

Aggression

An array of behaviours ranging from irritability to intermittent explosive disorders occur in 19-59% of individuals with HD. Irritability may be accompanied by verbal or physical aggression. Individuals are volatile and have a low threshold for stress and anger. They react to minimal provocation with an explosive response as they lack impulse control (Shannon 2004). Witjes-Ané et al (2002) suggest that aggressive behaviour may be the initial sign of the onset of disease. Tamir et al (1969) found that males were more likely to show aggressive behaviour and females to show depression.

Apathy

Apathy is the most significant behavioural abnormality to develop in individuals with HD and is present in about 59% of individuals (Hamilton et al 2003; Hankey and Wardlaw 2002; Hayden and Kremer 2001). Early signs of HD may include withdrawal from activities and friends and a decline in personal appearance and lack of emotional expression. There is decreased spontaneous speech and motivation which is not attributable to cognitive impairment, emotional distress, or decreased level of consciousness (Paulsen and Nehl 2004). Apathy increases with disease severity and is found in almost all individuals in the late stages of the disease (Shannon 2004).

It appears to be a specific neuropsychiatric syndrome that is distinct from depression (Hamilton et al 2003; Levy et al 1998), as a result of the frontostriatal neuropathology that occurs in HD and leads to a primary reduction in drive and motivation manifesting as behavioural apathy (Hamilton et al 2003). Individuals with hypokinesia are more likely to have apathy (Litvan et al 1998).

2.12 OTHER ABNORMALITIES

Weight loss

Gradual weight loss is common, but not consistent, as the motor symptoms of HD progress (Hamilton et al 2004; Quarrel and Cook 2004; Trejo et al 2004; Fletcher 2001) and there is striking emaciation and generalised muscle wasting in individuals with advanced disease (Jankovic and Lang

2004; Moskowitz and Marder 2001; Bruyn 1968). The weight loss may occur in spite of an adequate dietary intake (Quarrell and Cook 2004; SuttonBrown and Suchowersky 2003; Morales et al 1989; Sanberg et al 1981) or even with an increased carbohydrate intake (Farrer and Yu 1985). Some studies have found that weight loss is not related to either the hyperkinesis or the nutritional intake (Quarrell and Cook 2004; Djoussé et al 2002; Fletcher 2001), whilst others have found that weight loss is associated with severe chorea (Mahant et al 2003; Pratley et al 2000). Other reasons for weight loss can be attributed to dysphagia, loss of appetite due to apathy, depression and to cognitive impairment (Mahant et al 2003; Moskowitz and Marder 2001).

Sleep

In the early stages of the disease sleep is essentially normal (Bollen et al 1988; Emser et al 1988; Hansotia 1985). Later, however, many individuals develop insomnia at night and somnolence by day (Morton et al 2005; Fletcher 2001; Kirkwood et al 2000; Hansotia 1985) which become difficult aspects for the family to manage (Quarrell and Cook 2004).

Incontinence

Approx 20% of all patients are incontinent of urine and faeces in the terminal phases of the illness (Quarrell and Cook 2004; SuttonBrown and Suchowersky 2003; Fletcher 2001; Hayden and Kremer 2001). This may be partly explained by choreic contractions of the perineal musculature (Quarrell and Cook 2004). The condition makes home care both difficult and physically demanding for the family and is a cause of discomfort and indignity to the individual (Pollard 2000).

Functional decline

Although some authors argue and many affected individuals may complain that their voluntary movements are clumsy and that their physical activities are impaired by their hyperkinesia and ataxia (Fletcher 2001; Hayden and Kremer 2001; Gordon et al 2000;Ross et al 1997; Phillips et al 1994; Folstein 1989), other authors claim that, unless the chorea is very severe, it does not appear to constitute a major impairment to normal function (Mahant et al 2003; Fletcher 2001; Hayden and Kremer 2001; Moskowitz and Marder 2001). Functional disability is best correlated with bradykinesia, rigidity and cognitive decline (Mahant et al 2003; Moskowitz and Marder 2001; Snowden et al 1998).

There is also motor impersistance, where there is an inability to maintain a body position or sustain a movement. The individual is unable to maintain tongue protrusion, maintain forced eye closure, hold the mouth open for long periods or maintain a constant handgrip (Jankovic and Lang 2004; Shannon 2004). This affects the individual's functional capacity and is frequently mistaken for misbehaviour. For example, an individual feeding himself might suddenly drop the spoon midway between the plate and his/her mouth or suddenly fall while he is walking with assistance. Another part of movement dysfunction which affects daily activities is the inability to modulate or regulate the force of movements making the individual appear clumsy and as this aspect progresses so the level of functional ability declines (Pollard 2000).

Behavioural disorders contribute significantly to functional disability (Hamilton et al 2003). The level of functional impairment is moderately related to the severity of motor and cognitive dysfunction (Rothlind et al 1993; Bamford et al 1989; Young et al 1986).

Epilepsy

Epileptic seizures occur in adult HD patients with a slightly increased frequency than the general adult population (1%) (Fletcher 2001; Hayden 1981).

Criminal behaviour

Criminal behaviour involving various sorts of crime including assault, arson and murder has been considered a significant problem in HD in South Africa (Hayden et al 1980c) and other countries (Dewhurst 1970; Parker 1958; Reed and Chandler 1958). A large study conducted in Denmark, however, shows that criminal behaviour of low severity appears to be increased in men only. It appears to be related to the disease process and not due to environmental and familial factors. They found that there was no increased incidence in females or in male and female first degree relatives (Jensen et al 1998).

2.13 DIAGNOSIS

The definitive diagnosis of HD is based on:

- a confirmed positive family history (Shannon 2004);
- the presence of an unequivocal movement disorder involving both voluntary and involuntary movements (Paulsen and Hoth 2004; Hayden and Kremer 2001);
- mental disturbances including cognitive decline, affective disturbances, and/or changes in personality (Hayden and Kremer 2001);
- neurophysiological and neuroimaging studies such as EEG (Cummings and Benson 1992); CT or MRI (Hohler and Samii 2004; Shannon 2004); PET (Askin-Edgar et al 2004; Fletcher 2001); and SPECT (Askin-Edgar et al 2004); and
- confirmation by direct DNA testing for the CAG repeat expansion in the huntingtin gene (Hohler and Samii 2004; Myers 2004; Shannon 2004; Fletcher 2001). Although the diagnosis can be confirmed by means of this test it cannot establish the presence of symptoms of HD and an individual who has tested gene-positive may exhibit symptoms of some other disease (Myers 2004).

Various studies indicate that caution has to be taken when providing an individual with a diagnosis of HD as up to 31.5% of their cohorts with apparent "unequivocal" HD-like symptoms were found to be gene-negative (Creighton et al 2003; Goizet et al 2002; Witjes-Ané et al 2002; Siemers et al 1996). For example, Huntington disease-like 2 (HDL2) has similar neurological symptoms, but these individuals test gene-negative for HD. This disorder is caused by a CTG/CAG expansion mutation on chromosome 16q24.3, with normal and expanded repeat ranges similar to HD, and a correlation between repeat length and onset age very similar to HD. HDL2 is generally rare, accounting for only a few percent of HD-like cases in which the HD mutation has already been excluded (Margolis et al, 2004). This demonstrates the importance of a detailed neurological examination without providing a diagnosis of HD even if there is a positive family history present. A DNA test will either confirm that the individual is gene-negative and the cause of the symptoms is unknown or it will indicate that the individual has inherited the mutation, but cannot give any indication as to whether the individual is affected at the time. It is only a detailed neurological examination that can determine the presence of symptoms (Hayden 2003).

Although there may be apparent sporadic cases of individuals developing HD without a family history these are frequently as a result of an unknown family history, premature death of a parent, suspected non-paternity, or an expansion in CAG repeat length from a parent with a CAG repeat length between 27 and 35 (Margolis and Ross 2003; Moskowitz and Marder 2001).

2.14 MANAGEMENT OF HD

Although HD was first described in 1872 (Shannon 2004; Quarrell and Cook 2004), understanding of the underlying neuropathology that causes its devastating symptoms are still rudimentary. This has thus hindered the development of pharmacological agents to prevent or effectively treat the disease (Bonelli et al 2004; Shannon 2004; SuttonBrown and Suchowersky 2003; Fletcher 2001). However, the recent completion of the Human Genome Project promises hope for new ways of diagnosing, treating and ultimately preventing the disease (Weinshilboun 2002). Death often occurs as a result of subdural haematoma (Folstein 1989); aspiration pneumonia following choking (33%); cardiovascular disease (24%) (Harper 1996); or suicide (1-3%) (Hayden and Kremer 2001).

The management of HD currently, therefore, remains only as symptomatic relief by means of drug and rehabilitation therapies (Bonelli and Hofmann 2004; Bilney et al 2003).

Pharmacological agents

There is poor evidence regarding the use of pharmacological agents in the management of HD (Bonelli et al 2004). Some of the symptoms such as depression, psychosis, hypokinesia, rigidity and chorea can be partly alleviated by pharmacological agents (Bonelli and Hofmann 2004; Lindvall and Bjőrklund 2000; Evers-Kiebooms and Decruyenaere 1998), but these drugs do not generally lead to a significant improvement in the level of functional ability (Hayden and Kremer 2001).

There are several medicines available for the treatment of chorea and dystonia, but the side effects of these are associated with a high risk of adverse events (Bonelli and Hofmann 2004; Quinn 2003). Psychiatric manifestations frequently respond to psychotrophic treatment and the amelioration of these symptoms can contribute significantly to an improvement in the quality of life (Bonelli and Hofmann 2004; Margolis and Ross 2003; SuttonBrown and Suchowersky 2003). Cognitive impairment cannot be managed by pharmacological agents (Hayden and Kremer 2001). Epileptic seizures require similar drugs as in other forms of convulsive disorders, but they are often difficult to control, particularly in individuals with the juvenile form of HD (Hayden and Kremer 2001).

Nonpharmacological management

As the pharmacological management of symptoms does not lead to a significant improvement in activities of daily living (Bonelli et al 2004; Fletcher 2001; Hayden and Kremer 2001) the well-being of individuals with HD and their families is dependent on other approaches that aim to optimise participation in family, work, leisure and community activities (Bilney et al 2003). This requires the multidisciplinary input of and support from physiotherapy, occupational therapy, speech therapy, psychotherapy, nursing, genetic counselling, community social services and national support societies to varying extents throughout the course of the disease (Bonelli et al 2004; Bilney et al 2003; SuttonBrown and Suchowersky 2003; Fletcher 2001; Hayden and Kremer 2001; Lewis et al 2000).

Research suggests that there is a low referral rate to the various therapies and that these services are not always routinely provided (Skirton and Glendenning 1997; Shakespeare and Anderson 1993). This could be due to the service providers being hesitant about providing rehabilitation programmes to people with neurodegenerative conditions based on an expectation of improvement. However, it could also be due to reservations about recommending therapy when there is limited scientific clinical evidence demonstrating effectiveness (Bilney et al 2003).

Despite these weaknesses in research a common perception of people with HD and their families is that therapy services are beneficial (Skirton and Glendenning 1997; Shakespeare and Anderson 1993). This perceived benefit may be derived from the psychological support provided during the rehabilitation sessions rather than any specific therapeutic techniques provided by the various therapists.

However, recent studies on mouse models for HD has shown that environmental stimulation delays the onset and slows the progression of the motor and cognitive symptoms (Spires and Hannan 2005; Sullivan et al 2001). Horton et al (2004) have also shown that there is a trend for physical, intellectual and social activity to be associated with later onset of the disease although statistical significance was not reached. The findings of these latest studies might provide an explanation for affected individuals and their families claiming that therapy services were found to be beneficial (Skirton and Glendenning 1997; Shakespeare and Anderson 1993). A potential treatment for people with HD is one where there is environmental enrichment such as increased social interaction, space, exercise facilities and novel activities and tasks to increase cognitive stimulation (Bilney et al 2003). Formal studies on affected individuals will have to be conducted which replicate the positive findings of an enriched environment from the mouse model studies where the rate of functional decline was reduced (Hockly et al 2002). By nature of the slow progression of HD, testing of the effectiveness of therapy needs to be done over a prolonged period of time and measurement tools need to be sensitive to be able to detect minor gradations of change (Snowden et al 2001).

Physiotherapy: While the individual is ambulant physiotherapy is directed at reducing impairment and maintaining function in motor activities. This usually involves short courses of rehabilitation to maintain and improve balance, postural stability and gait to prevent falls (Huntington's Disease Society of America 2005; Bonelli et al 2004; Grimbergen et al 2004; Schöls 2003; Churchyard et al 2001; Moskowitz and Marder 2001).

Occupational therapy: It is important that individuals be permitted to remain in some form of employment and fulfill domestic and social roles for as long as possible to avoid exacerbating feelings of inadequacy and loss of power associated with the disease (Hayden and Kremer 2001).

Occupational therapists conduct home visits and workplace assessments to provide advice on work and domestic adaptations and devices to allow the individual to function safely and independently for as long as possible. These assessments include an evaluation of the person's driving skill and ability to continue driving (Huntington Disease Society of America 2005; Rebok et al 1995; Drapo 1981; Hayden 1981).

Speech therapy: Speech therapy can assist with communication by teaching the family strategies to provide the affected individuals with prompts and cues to facilitate their communication (Bonelli et al 2004; Klasner 1990; Drapo 1981; Hayden 1981). Education is given to the individual and family regarding eating and safe swallowing techniques to reduce the risk of choking and aspiration (Paulsen and Hoth 2004; Kagel and Leopold 1992; Leopold and Kagel 1985).

Psychiatry/psychology: Effective management of mental disturbances in individuals may require psychiatric intervention and such care should be part of the treatment plan for all patients (Bonelli et al 2004; SuttonBrown and Suchowersky 2003). Van Duijn et al (2005) found that electroconvulsive therapy is effective in treating patients who are substantially limited in their daily functioning and have therapy-resistant major depression.

Dentistry: Dental and oral hygiene are important aspects, but often neglected in the care of individuals with HD. The patient should have regular quarterly dental care to maintain original teeth and to ensure the correct fitting of dentures. Not only is dental hygiene difficult to manage with chorea and dysphagia, but dental care is necessary to decrease episodes of choking (Moskowitz and Marder 2001). Effective cleaning minimises the bacteria that can be aspirated and reduces the risk of infection. Bruxism is common due to involuntary movements and changes of muscle tone in the mouth. When choking individuals frequently regurgitate their food and over time the gastric acid damages the tooth enamel leaving them susceptible to decay (Pollard 2000).

Dietician: Because individuals with HD need to have a much higher calorie intake than normal and they have dysphagia the dietician provides advice on techniques of increasing calorie intake with meals that are the correct texture and easy to swallow (Kent 2004).

Social worker: Social workers play an invaluable role in the management of individuals with HD and their families. They have contact with employers, advise on disability payments, and arrange respite and placements admissions. They conduct home visits to assess the individual within the home environment and provide support and advice to the families on the management of their affected relatives who have behavioural problems which affect the functioning of the family as a system (McCusker 2003; Sullivan et al 2001).

Support groups: Where available regional and national support societies provide opportunities for information, advice and support for both individuals and their families (Hayden 1981).

Family: Attention should also be paid to the well-being of the family members. Apart from possibly being at-risk for HD they also suffer from their daily burden of providing adequate care for the patient. Genetic counselling can be provided to counsel the family with regards to managing the psychological and social aspects of their lives (Williams et al 2007; Bennett et al 2003). Physiotherapy management includes teaching carers how to lift and transfer the affected individuals to minimise physical strain (Bilney and Morris 2003). Intermittent, temporary relief from this burden of caring through respite care may assist the family in providing long-term care in the home environment. A crisis at home is the most common reason for forced institutionalisation of HD patients (SuttonBrown and Suchowersky 2003; Hayden 1981). When the symptoms progress to the extent that the individual is severely incapacitated and the demands on the carer are extremely burdensome and disruptive to the family, home care may not be realistic and admission to a nursing home is essential (Fletcher 2001).

Homecare: In order to ensure that the individual receives optimal care and the caregiver experiences minimal burden, it is essential that caregivers receive regular contact with home services that can provide advice and support to the caregiver and, in this way, minimise the patient's psychiatric and physical problems and avoid or delay the need for institutionalisation (Moskowitz and Marder 2001).

2.15 PSYCHOSOCIAL IMPACT OF HD

2.15.1 ON THE FAMILY AS A SYSTEM

Although it may be considered a rare disorder the uncertainty and anxiety associated with the mode of inheritance and AOO leads to constant stress in the family, particularly the spouse (Timman et al 2004; Meincke et al 2003; Jensen et al 1998) even if the disease is not discussed frequently (Anderson and Marshall 2005; Quarrell and Cook 2004; Richards and Williams 2004; Brouwer-DukodeWit et al 2002). Families continually have to deal with chronic neuropsychiatric disorders and premature deaths which are disruptive to their structure, roles and functioning. Family roles and relationships change such as when the wife is compelled to become the breadwinner or the husband has to leave employment to take on the role of carer, placing intolerable strain on the family. The resulting unemployment, financial problems and lack of resources exacerbate tensions in the family (LoGiudice and Hassett 2005; Ross and Deverell 2004; Brouwer-DukodeWit et al 2002).

Children coming from such disorganised households have been found to have related antisocial personality disorders in adulthood and a high rate of psychiatric disorders in adolescence (Duisterhof et al 2001). However, they are not found in individuals whose parents developed symptoms when they were already adults (Folstein et al 1983). These result from growing up with a parent who is physically present, but who gradually becomes distanced emotionally, psychologically and cognitively from the child (Forrest Keenan et al 2007; Berrios et al 2002; Brouwer-DudokdeWit et al 2002). Frequently the unaffected parent has to take over the role of the affected parent in addition to caring more and more for them which results in less attention and time for the child (Duisterhof et al 2001).

When the affected individual starts to show symptoms the healthy partner sometimes elects to leave the household (Duisterhof et al 2001). Thus, instead of the child playing a dependent role he/she takes over the parenting role by caring for the parent and dealing with their personality changes and psychiatric symptoms of irritability, short-temper, withdrawal and depression (Forrest Keenan 2007; Brouwer-DudokdeWit et al 2002).

The parents may experience feelings of guilt, shame and remorse at the possibility of having passed on a serious condition to their children (Arnold et al 2005; Knebel and Hudgings 2002). Individuals who are mutation-positive find it difficult to know when to inform their children that they are at-risk and frequently never appear to find the right moment until their children are almost adult. Hence the topic of HD is often not talked about, particularly while the parent is asymptomatic. This secrecy makes it impossible to discuss the subject with the family and each family member lives in isolation with his/her own thoughts, fears and feelings about the disease. This secrecy also leads to loss of contact with the affected individuals' wider family (Lowit and van Teijlingen 2005; Brouwer-DudokdeWit et al 2002; Bloch et al 1992). This inability to talk about HD appears to be one of the reasons the knowledge about the disease held within families is often so distorted, inaccurate and incomplete (Forrest Keenan et al 2003; Harper 1996). However, when the children are informed of their 50% at-risk status it can set off a series of negative emotions resembling an acute grief process after a traumatic event (Evers-Kiebooms and Decruyenaere 1998).

Family members experience ambiguous loss when the affected relative is physically present, but psychologically is fading away as the person they have always associated with a specific role. The grief they experience is known as "disenfranchised" as it is only mourned by the family involved and there is usually very little social support provided for them (Sobel and Cowan 2003). The gradual deterioration of a parent causes the children a great deal of stress as they fear that when they get older their lives will follow the same distressing course (Evers-Kiebooms and Decruyenaere 1998).

Family members use different strategies to deal with their at-risk status or the fact that a family member has developed symptoms. Some use defensive avoidance and denial. Denial involves negating the thoughts and actions of a psychological or external demand while avoidance means that the person is aware of the internal or external demand, but prefers not to think or talk about it. Avoidance takes the form of refusing to think about it or discuss HD within the family or not seeking information about the condition. This occurs at the time of diagnosis, as symptoms progress or at the time a family member undergoes PT. It can also take the form of lack of long-term future planning for care of an affected individual or for the care of the future generation of HD sufferers (Lowit and Van Teijlingen 2005). However, conscious avoidance is often accompanied by symptom-watching and negative avoidance behaviour such as withdrawing from activities before symptoms arise (Lowit and van Teijlingen 2005). Denial and avoidance strategies can be destructive or constructive. If it is necessary to take action to reduce a threat and the person uses denial/avoidance strategies the processes are negative, but if action is not going to change the situation denial/avoidance strategies can be constructive. Some families use 'magical thinking' where there is preselection of a person by the siblings as the person who will develop HD because they resemble the affected parent in appearance or behaviour or simply by gender vulnerability. This illusion reduces their stress as they believe that their own risk is reduced in this way (LoGiudice and Hassett 2005; Lowit and van Teijlingen 2005; Middleton 2005; Evers-Kiebooms and Decruyenaere 1998; Kessler 1988).

When the first family member undergoes PT and discloses the results it can also lead to ostracisation by the rest of the family if they perceive that there is a stigma about the disease and that the family history of it should be restricted to the family and remain confidential (Charmaz 1999; Tibben et al 1990).

2.15.2 ON THE INDIVIDUAL

Uncertainty: Late-onset genetic diseases are surrounded by uncertainty. This begins with the uncertainty of whether they have inherited the gene mutation or not and continues in the form of symptom watching wondering whether vague symptoms are indicative of the onset of disease. In individuals who are considered to have a new mutation the onset of symptoms may be slow and insidious and it may be years before being diagnosed. However, under these circumstances, once the diagnosis of HD is finally made, it can often come as a relief as the medical recognition legitimises the person's symptoms and terminates conflicts with others over the reality of the symptoms. Confirmation of diagnosis can also be a devastating experience for the persons concerned and their families. It can lead to further uncertainty about the course and outcome of the disease (Ross and Deverell 2004; Charmaz 1999). Although the situation is distressing for the person diagnosed with HD, as the illness progresses their insight becomes progressively more impaired and so does their commitment to family responsibilities (LoGiudice and Hassett 2005).

Reconstitution of the self: Individuals with progressive diseases are involved in a constant struggle to lead valued lives and maintain definitions of self that are positive and worthwhile as their level of physical, cognitive and affective function decreases. This "loss of self" can be a powerful form of distress (Ross and Deverell 2004) and depression is highly associated with this experience of loss and powerlessness (Reynolds 2004).

Emotional reactions: A common initial reaction to the knowledge that the individual is at-risk or has been diagnosed with HD is shock, disbelief or numbness as the individual realises that the consequences are likely to lead to change in lifestyle, loss of control, potential loss of role, status, independence and financial autonomy. These reactions are more pronounced if the person has not expected the diagnosis or anticipated the experience (Ross and Deverell 2004; Falvo 1991).

Grief: "Grief is a process whereby a person can separate him- or herself from someone or something significant that has been lost" (Ross and Deverell 2004:36). In HD loss occurs by reduced functional capacity, gradual cognitive impairment, emotional instability and increased financial independence (McAteer 2004; Falvo 1991). Kübler-Ross (1969) delineated the main stages in the grieving process as denial, bargaining, guilt, depression, anger and acceptance. These affective states do not have a specific linear order, but occur in a cyclical fashion and do not necessarily achieve acceptance. In a deteriorating disorder such as HD this means that any additional loss of function can again plunge the individual into an emotional state which had already been successfully achieved in the past (Kolt and Andersen 2004; McAteer 2004; Reynolds 2004; Ross and Deverell 2004).

Denial: Denial is often present early in the grieving stage and can be observed as rejection of the diagnosis provided by the doctor or refusal to recognise the true nature of the prognosis. It is a defense mechanism that protects the psyche from unmanageable levels of anxiety that people are not ready to deal with and allows the patient time to develop, below the threshold of conscious awareness, more adaptive coping strategies. It is not a passive process, but rather an active process in which there is a great deal happening beneath the surface on the preconscious and unconscious levels. When they find the inner ego strength as well as the information and support needed to deal with the problem the denial stage ceases (French and Sims 2004; Ross and Deverell 2004; Reeves et al 1999; Davidhizar and Giger 1998; Livneh and Antonak 1997; Falvo 1991).

Bargaining: The bargaining phase is characterised by the individual or family trying to behave in an exemplary fashion by following the doctors' orders or devoting themselves to the caring of their affected relative if in turn they can expect complete or significant improvement in the individual's problems (Ross and Deverell 2004).

Guilt: The affected individuals experience guilt if they already have children and are concerned about their role in passing on the risk to their children (Ross and Deverell 2004). Guilt can also arise when individuals receive mutation-negative results of predictive tests. They express relief for their personal results, but feel guilty when their relatives test mutation-positive or develop symptoms for HD (French and Sims 2004). The burden of guilt and anxiety of HD families diminishes their capacity to assist and support affected individuals or their carers (Harper 1996; Hans and Koeppen 1980).

Depression: Depression is highly associated with the experience of loss and powerlessness (Reynolds 2004). Reactive depression is a normal grief reaction to a traumatic experience such as inheriting a neurodegenerative disease. Individuals perceive themselves as being impotent, incompetent, incapable and of little value as they are powerless to influence something so close to them that they want so desperately to alter. It is a normal and necessary part of the grieving process, provided it does not persist for too long or become too severe (Ross and Deverell 2004).

Anger: Anger is an integral part of mourning or grieving and can be directed at the self, a parent for passing on a terminal progressive disease, siblings who have tested negative or towards persons who offer unwanted advice (LoGiudice and Hassett 2005; French and Sims 2004; Ross and Deverell 2004).

Anxiety: Anxiety in individuals with HD is often associated with waiting for test results confirming the diagnosis, contemplating changes in lifestyle or uncertainty about the future. Clearly the diagnosis of a life-threatening disease is likely to generate high levels of anxiety and there is a feeling of being totally overwhelmed and the desire to be rescued (Reynolds 2004; Ross and Deverell 2004; Livneh and Antonak 1997).

Acceptance: People move through the grieving process at different rates until they reach the stage of acceptance. However, each time the individual comes to a major life milestone such as being unable to continue working, losing the ability to speak or walk; increasing dependence; or attending a funeral of an affected family member, the grief may resurface (Ross and Deverell 2004).

2.16 COPING

Coping is the cognitive, behavioural and emotional ways in which people manage stressful situations (Baum et al 1997). There are close relationships between the individual's perception of HD and their coping responses and individuals use a wide variety of coping strategies to manage life events (Kolt and Andersen 2004). In the absence of a cure, affected individuals and their families require resourcefulness, tenacity and a broad repertoire of coping strategies if they are to adapt successfully to the ensuing physical, psychological, social and financial changes and achieve an acceptable quality of life (Reynolds 2004). Coping strategies and lifestyle choices are rarely a matter for the individual in isolation (Maes et al 1996) and family support or rejection have major influences on the person's choice of coping strategies (French and Sims 2004).

The availability of physical and environmental resources also affects coping. Financial security empowers choice, for example, enabling the purchase of suitable aids and adaptations in the home or domestic assistance. Social security benefits are generally set so low that they do not allow affected individuals and their families any choices over housing, transport and social activities. Such enabling and disabling factors in the person's environment have a great impact on coping strategies and quality of life (French and Sims 2004).

2.16.1 POSITIVE SELF-MANAGEMENT STRATEGIES

Individuals adjusting successfully to long-term diseases tend to develop a variety of self-management strategies over time. These include:

- acquiring information about the disease, management options, benefits and services available together with active participation in decision-making about the management of the disease, can reduce associated stress by increasing their sense of control (Kolt and Andersen 2004; Livneh and Antonak 1997; Earll et al 1993; Falvo 1991)
- for the chronically ill, information is a significant resource for managing their lives. It reduces uncertainty, helps the individual to come to terms with the illness and allows for the development of strategies for managing the illness in every day life (Felton et al 1984).
- general health promotion activities such as physical exercise and good nutrition;
- increasing social support, for example, joining HD support groups (Coyner and Bolger 1990);
- humour serves an important function in coping with major threats and fears. It enables individuals and their families to minimise intense emotions and to get things into perspective when there are problems that appear overwhelming. However, it may only be an option for those enjoying a secure sense of self and strong social support (French and Sims 2004; Ross and Deverell 2004).
- spiritual and religious values. Finding meaning to life in adversity is a significant resource for coping with stress and achieving a better quality of life (French and Sims 2004). Antonovsky (1990) argues that people with a strong sense of coherence are more likely to find such meaning. Strong religious or spiritual values help to provide people with a sense of meaning or purpose as well as enhancing their experience of social and emotional support during serious illness (French and Sims 2004; Kolt and Andersen 2004).

2.16.2 NEGATIVE COPING STRATEGIES

Severe degenerative diseases can lead people into abandoning constructive strategies through despair and hopelessness (French and Sims 2004) and cause them to:

- become totally passive by regarding their illness as uncontrollable and their treatment options as futile, feeling overwhelmed by helplessness (French and Sims 2004; Kolt and Andersen 2004).
- adopt a 'sick role'. The meaning of the 'sick role' is often associated with excessive dependence by affected individuals who gradually perform fewer duties within the home and hand over their usual roles and responsibilities before they become incapacitated (French and Sims 2004).
- abuse alcohol and illicit drugs. Men appear to be more likely than women to turn to alcohol and substance abuse when stressed (Gianakos 2002). Alcohol abuse is more likely to occur in HD when stress has previously been managed through alcohol consumption (Holahan et al 2001). Emotion-focused strategies such as excessive alcohol consumption tend to be more common in the early stages of the disease when shock, anxiety and grief reactions are uppermost (Reynolds 2004; Livneh and Antonak 1997; Falvo 1991).

2.17 DISABILITY

Disability is a complex concept resulting in a multiplicity of definitions which describe it (Nordenfelt 1997; Oliver 1996; Nordenfelt 1995; Quinn 1995; Barnes 1994; Verbrugge and Jette 1994; Nagi 1991; WHO 1980). The descriptions and explanations vary according to the theoretical framework of the models used by the authors.

Before providing examples of the plethora of definition of the term 'disability' it is necessary to discuss the models of disability to provide a theoretical framework.

2.17.1 MODELS OF DISABILITY

The models of disability can be placed on a continuum with the medical model at one extreme and the social model at the other. At the one extreme disablement is considered by the medical model to have its locus in the individual with the disability; whereas the viewpoint at the other extreme, namely, the social model, disability is seen to be located in the social environment and it is that which places restrictions on the disabled person.

Medical model

The medical model was constructed by medical scientists for the purpose of studying disease. It focuses on individual pathology and physical malfunctioning and is the prevailing model used in the Western world (Marks 1997; Imrie 1997; French 1993d; Zola 1993). As the basic scientific discipline is that of molecular biology the norms are measurable biological values occurring at the molecular level and are considered to be independent of the social, psychological and behavioural factors that may be affecting the disabled person (Mason 1985). The numerical values assigned to the biological measure assume much more importance than the disabled individuals themselves and the only aspects of importance are seen to be ways of curing, preventing or caring for disabled people (Marks 1997; Harrison 1993). Disability is seen only in terms of failure of one of the bodily systems and is perceived as a deviation from the normal (Marks 1997; Davis 1995; Abberley 1993; French 1993d).

Although this approach to studying disease has been most successful, the fact that it has excluded related psychosocial and cultural aspects interferes with effective patient management (Gleeson 1995; Shakespeare 1994;

French 1993d; Harrison 1993; Scheer and Groce 1988). It places more emphasis on technical and laboratory tests than it does on the person's subjective account. Therefore, psychological and behavioural manifestations and descriptions are often not related to the biochemical deficits (Oliver 1990; Bury 1979).

The professionals consider themselves as a group of people who are responsible for working on behalf of the disabled people in order to help them overcome their problems which are perceived to arise out of their disabilities. Thus the focus is on 'normalising' the disabilities rather than accommodating them (Davis 1995; French 1993d; Lonsdale 1990; Chamie 1989; Finkelstein 1989).

Although proven to be effective with many acute conditions, the medical model perspective offers little beyond assessment, information and limited palliative treatment of the symptoms for patients with incurable conditions such as HD. It ignores the family and patient's own experience of the disease and its challenges to the person's identity, lifestyle and relationships because they are considered to be irrelevant to treatment and recovery (Reynolds 2004).

Social model

Proponents of the social model claim that society needs to adjust to impaired people rather than disabled people needing to adjust to their impairment (DPSA 2001; Marks 1997; Lonsdale 1990).

This perspective of disablement demands a great deal of change in the relationship between the professionals and the disabled as the social model accepts that disabled people have medical conditions which may inhibit them and may need medical treatment from time to time, but that most of the day-to-day problems that they have to face are caused by the fact that they live in a disabling world which is largely designed to fit able-bodied people (DPSA 2001; Marks 1997). The social model is, therefore, critical of those professionals whose aim is to change the people with disabilities rather than the environment as the social model gives due weight to the psychological and social as well as the biological aspects of disease and health care (Reynolds 2004).

As opposed to the individualised approach of the medical model, the social model places disablement in a rejecting and oppressive social environment and not in an impaired or malfunctioning body. It maintains that the restrictions encountered by people with disabilities are not a natural consequence of their impairment, but are as a result of the social environment which fails to take account of their differences (Oliver 1990; Longmore 1987; Zola 1985; Finkelstein 1980).

The extent to which disabled people experience their impairments include the nature of the natural and built environments (Imrie 1996; Sibley 1996), social hierarchy (Oliver 1996), legislation (Barnes 1994), attitudes, images and technologies (Roulstone 1996; Finkelstein 1980), aesthetics (Wendell 1996; Mitzoeff 1995; Bogdan 1988) and languages and culture (Davis 1995; Shakespeare 1994; Morrison and Finkelstein 1993; Groce 1985).

2.17.2 DEFINITIONS OF DISABILITY

Quinn (1995: 56) defines disability as a "Physical or mental impairment that substantially limits one or more of the major life activities of an individual" which is based on the medical model; whereas other authors describe it from the point of view of physical and social barriers which negatively affect the person's opportunity of taking part in the normal life of the community (Bickenbach et al 1999; Pope and Tarlov 1991; Haber 1990). For example, Barnes (1994: 88) defines disability as, "The loss or limitation of opportunities to take part in the normal life of the community on an equal level with others due to physical and social barriers".

Wood and Badley (1983) contend that disability cannot be managed in isolation by dealing with the individual's physical or mental state, but requires improving the underlying impairments and disabilities in conjunction with altering the environments in which the individuals live and increasing the resources available to them. In this way they can personally manage the problems confronting them. This requires changes in social security and welfare policies, vocational training, housing, education, transport as well as the provision of comprehensive rehabilitation services (Lang 2000; O'Day 1999; Hahn 1988; Abu-Khalil 1997; Marks 1997; Morris 1993d; Wood and Badley 1983).

Proponents of the social model of disability claim that the level of handicap is a product of both a disabled person's characteristics such as race, gender, socio-economic class, culture and environment (Smart and Smart 1997; Shakespeare 1994; Lonsdale 1990) and the characteristics of the individual's environment such as public transport, accessibility of buildings, availability of services and employment opportunity (Marks 1997; Imrie 1996; Sibley 1996; Oliver 1996; Barnes 1994; French 1993c; Chamie 1989). Thus the person's personal characteristics do not form part of their explanations of disablement.

The International Classification of Functioning, Disability and Health (ICIDH-2) integrates health at biological, individual and societal levels in order to achieve a universal language for functioning and restrictions (WHO 2001). It focuses on strategies people use to live with their health conditions and how these can be enhanced so that their lives are productive and fulfilling by providing a mechanism to document the impact of the social and physical environment on a person's ability to function in society (Kersten 2004). The ICIDH-2 (WHO 2001) categorises disability into

- body dimension which includes the body structures and functions of the body systems;
- activities and participation. This includes the full range of activities performed by the individual such as social, occupational, recreational and activities of daily living; and

- participation which includes areas of life in which the individual is involved, has access to, and has societal opportunities or barriers.

Environmental factors have an impact on all three of the above dimensions and are organised from the individual's most immediate environment to the general environment. The WHO (2001) considers 'functioning' and 'disability' to be umbrella terms covering the three dimensions and that problems such as impairment, limitation of activity or restriction are situated on the one end of a continuum and non-problematic aspects of functional states on the other (Ross and Deverell 2004).

Lack of consensus of the concept of disablement has led to different notions about a person with a disability (Pinder, 1993) and makes it difficult to obtain accurate data and to compare research findings (Smart and Smart 1997; Helander 1993; Zola 1993; Chamie 1989).

2.17.3 SOCIO-ECONOMIC FACTORS BOTH INFLUENCING AND BEING INFLUENCED BY DISABILITY

Disability and employment: To be disabled is also to be disadvantaged. It means being unable to participate in the social and economic activities which most people take for granted and this leads to a state of conditioned passivity (Lonsdale 1990; Sutherland 1981). Rates of poverty, unemployment, domestic violence and suicide are much higher among people with disabilities than in the non-disabled population (Daniels and Mickel 2002; Little 2002; Schneider 1999; Smart and Smart 1997; Lonsdale 1990; Locker 1983; Wright 1983; Sutherland 1981; Syme and Berkman 1981).

Disability and poverty: Disability has a major impact on society. It does not only affect the disabled, but also the family and the immediate community (DPSA 2001; Office of the Deputy President 1997; Jette 1994; McNeil 1993; Alston and Mngadi 1992; Thornhill and HoSang 1988). Unemployment contributes to poverty and without paid employment an individual has fewer social contacts, less status and most importantly, less income (Daniels and Mickel 2002; Smart and Smart 1997; Lonsdale 1990). There is greater poverty in disabled households than non-disabled households (Jelsma and Maart 2004; Martin et al 1999; Smart and Smart 1997) and the occurrence of disability in a family, often places heavy demands on family morale, thrusting it deeper into poverty (Office of the Deputy President 1997).

Disability and social stigma: Social and cultural belief systems determine the extent to which specific illnesses are stigmatised (French and Sims 2004). Stigma of disease often results in discrimination, social isolation, disregard, depreciation and devaluation (Bombard et al 2007; Taylor 2004; Falvo 1991).

There are many social stigmas linked to the physical and psychiatric disabilities of HD and for asymptomatic people associated with a family history of HD. For example, people have been refused employment because of a family history of the disease even in situations where the individual has no symptoms and there is no danger attached to them personally or others in the particular form of employment. In addition sudden events or rapid onset of disease do not occur in healthy people who are at-risk for HD (Bates 2005; Ensenauer et al 2005; Harper et al 2004; Burgermeister 2003; Martin et al 1999).

Disability and self-image: 'Body image' is a central part of self-image and consists of a person's feelings about his/her body which is partly based on how other people react to it (Mitzoeff 1995; Knudson-Cooper 1981). Stereotyped attitudes, particularly negative ones, influence the social expectations and self-image of the disabled (Lonsdale 1990; Ainley 1984; Hastings 1980; Roeher 1961). Those who become disabled in adulthood experience a major change in their identity and a strong sense of loss. This is particularly so in patients with HD (Simon-Meyer 1999; Falvo 1991).

Disability and independence: Disability is a relative concept as society expects people to be independent, but does not provide for the extent of dependence that the disabled frequently require by having to rely on others in their environment (Grundy 1999; Bain 1998; French 1993a; Morris 1993d; Lonsdale 1990). Disability in itself need not impose a state of dependency on someone, but an unaccommodating and hostile environment or prejudicial attitudes do so (Bain 1998; Law et al 1996; Mann and Lane 1995; McCuaig and Frank 1991).

Crucial to anyone's ability to live independently is having acceptable and suitable accommodation that is provided or having the financial means to provide it for oneself (Grundy 1999; Law et al 1996; French 1993a; McCuaig and Frank 1991).

Disability and abuse: Although people with disabilities are particularly vulnerable to abuse there was minimal literature available on the prevalence of abuse of people with disabilities in general and specifically men with disabilities and people with HD.

People with moderate HD, like any person with a chronic disability, can become an easy target for abuse as a result of the mental changes that they have undergone or because they are easily identified victims. The abuse can be either by a carer, member of the family or community and can take the form of verbal, physical or sexual abuse. It can also be as result of the patient abusing the carer or family members. Sexual abuse can take place in a variety of ways in families where there is HD. The patient can become a target for abuse both in and outside the family. Likewise the women and children in the family can be sexually abused by the patient if there is a loss of social and sexual inhibitions. Sexual abuse of children in a family can occur when the mother is symptomatic and unable to carry out her role as a mother to protect her children from sexual advances by family or community members (Dubinsky 2006; Paulsen 2006; Rosenblatt et al 1999).

Disability and powerlessness: Miller (1983: 3) defines powerlessness as "A perception that one's own actions will not affect an outcome".

Powerlessness may occur in people affected with HD as a result of physical or psychological losses and is particularly prevalent at the time of diagnosis. It is frequently expressed as depression or anger towards others (Charmaz 1999). The sense of lack of control may arise from the disease itself where the disabled person is no longer able to continue working, getting from place to place or carrying out basic activities of daily living independently (Swain et al 1993).

With chronic disabilities the feeling of worthlessness occurs when the disabled are no longer able to perform physical functions or fulfill their previous role within the family as earners, parents, caring for children or parents for whom they feel responsible (Kolt and Andersen 2004; Livneh and Antonak 1997; Morris 1993b).

Self-esteem is a power resource and enables the disabled to take an active part in controlling their personal care (Mitzoeff 1995; Oliver 1993; Falvo 1991). For a person with HD this can gradually decline as he or she loses independence and the family has to undertake her/his roles and responsibilities (Falvo 1991). Dimond (1979) and Pattison (1974) both found that the social support network provided by families helped the disabled to maintain psychological strength as a power resource.

2.18 SOCIAL ASSISTANCE IN SOUTH AFRICA

Most Western industrialised countries have state-controlled programmes to provide basic social security for pensions, unemployment, housing, education and health services for those who are permanently or temporarily disadvantaged. However, in developing countries where there are limited resources social assistance is restricted and does not reach all the population in need (Ahmad et al 1991).

In South Africa the welfare system is that of the 'residual' type of service delivery (Luiz 1994) in that it only provides some form of assistance to those in need when the individual, family, community or economic systems can no longer cope and it is regarded only as a safety net. Aliber (2001; 38) describes it as being "less of a net devised to catch an unfortunate few in times of temporary distress, than a major commitment to help a large fraction of the population over a sustained period". Thus it is typical of countries at this stage of economic development and industrial transition (Van Der Berg 1992). Social assistance is not seen as the right of the individual and the services are provided selectively and are viewed as a privilege which may be withheld (Lui; 1994; McKendrick 1987). In other words, only the symptoms of poverty are partly being addressed, but the underlying causes have not yet been dealt with in any meaningful manner (Ministry for Welfare and Population Development 1997; Luiz 1994).

The system of social assistance is controversial. The main arguments against social assistance include the systems' reinforcement of the medical model of disability and the culture of dependency it creates (Government Gazette 1108 of 1997). However, people with HD are obliged to depend on welfare benefits in order to survive. It also acts as a safety net to other members of the disabled patient's family and has been a source of household security for millions of the population, greatly contributing to poverty alleviation (Hunter 2002; Office of the Deputy President 1997; Lund 1997; Budlender 2000; Schneider and Marshall 1998).

The White Paper on Social Welfare (Government Gazette, Notice 1108 of 1997) emphasises that welfare programmes should optimise social functioning and concedes that as resources are limited the welfare expenditure can only be increased when higher economic growth is achieved in the country and that people's expectations, including those of the disabled, cannot be comprehensively met in the short term.

Social assistance programmes provided by the state are presented in Appendix (ii).

2.18.1 DISABILITY GRANTS (DG) IN SOUTH AFRICA

The welfare office in each magisterial district administers social grants on behalf of the provincial Department of Social Services. When applying for a DG the disabled have to submit documentation to prove their need on the basis of a means test and the amount awarded is calculated on a sliding scale. It takes into consideration the material means available to potential recipients in determining both whether they should receive benefits and what level of benefits they should receive (Van Der Berg 2001). The maximum monthly amount that can be awarded is R820.

Several studies have found that the main items that the disabled spend their money on are household necessities such as food and groceries and clothes (Schneider et al 1999; Schneider and Marshall 1998; Lund 1997).

When people with disabilities need to be cared for at home by a family member they are entitled to apply for a grant-in-aid to relieve their financial burden as many of them have had to give up their jobs to stay at home and take on the role of caregiver (Government Gazette No R 417 of 1998).

2.18.2 CRITICISMS OF THE SOCIAL GRANT PROCESS

From the researcher's personal experience and reading the literature available on the social grant system there appear to be problems all along the process of applying and being awarded a DG (Kimani 2000; National Workshop on Social Assistance Policy 2000; Schneider et al 1999; Schneider and Marshall 1998; Lund 1997).

There does not appear to be a standard procedure to follow to activate the application process and there is a lack of information on the actual procedure for the disabled people which makes the application process extremely lengthy and difficult (Kimani 2000; Schneider and Marshall 1998). The majority of people applying for DGs have difficulty in moving independently and they have less education and are less literate. They are sent from one stage of the process to another and from one doctor to another (Kimani 2000). There are few guidelines for assessing the extent of disability and it rests with and is awarded at the discretion of the medical officer (Kimani 200; Lund 1997) who frequently does not complete the forms in full or his/her handwriting is illegible making it difficult for a rational basis for either approval or denying the grant (Kimani 2000). If an application is not successful the officials do not provide the disabled with the reasons for their applications being rejected. Nor do they give them reasons for grants suddenly being stopped (Lund 1997).

Although the benefit is a statutory right, the most recent legislation of 1997 has made it a discretionary award. It is heavily biased towards a medical diagnosis and does not take into consideration the disabled person's external environment (Lund 1997) nor the number of dependents a household may have (National Workshop on Social Assistance Policy 2000).

The Committee of Inquiry (2002) recommends that the disabled who are also poor should receive a basic income grant (BIG) as a first step in a package of benefits. This grant should meet their daily living expenses (Le Roux 2001). Thereafter, consideration should be given to their special needs and provision should be made in the form of 'topping up' in relation to cash benefits, in-kind benefits and other essential services (Committee of Inquiry 2002). President Thabo Mbeki's State of the Nation address on 09.02.2007 announced the foundations for the introduction of a BIG in the near future (Terreblanche 2007).

2.18.3 DISABILITY AND POLICY IN SOUTH AFRICA

Because, historically, the emphasis in South Africa has been on the medical needs of people with disabilities, there has been a corresponding neglect of their wider social needs which has resulted in severe isolation for many of them and their families (Office of the Deputy President 1997).

In the White Paper on an INDS (1997) it is made clear that there has to be a fundamental shift in the way in which disabled people should be viewed and that the previous individual medical perspective should be replaced with an approach which ensures the human rights and development of disabled people (Office of the Deputy President 1997). The Bill of rights in the Constitution of the Republic of South Africa (Act 108 of 1996) provides for both political and socio-economic rights for all South Africans within a framework of non-discrimination and equality. In particular it provides governmental departments with guidelines that enable people with disabilities to:

a) Be as free as possible from needing permanent medical care and rehabilitation services, but having access to such whenever necessary (Office of the Deputy President 1997: 25-26);

b) Have a minimum livelihood, if appropriate by means of social benefits (Office of the Deputy President 1997: 52);

c) Provide disabled people and their families with safe shelter and dwellings of their own through equitable access to a range of options in the housing subsidy scheme (Office of the Deputy President 1997: 54).

2.19 PREDICTIVE TESTING

HD was the first genetic disorder for which PT was offered after the huntingtin gene was mapped in 1983 (Gusella et al 1983). It has enabled asymptomatic individuals to determine whether they will, at some time during their lives, develop the disease or not (Evers-Kiebooms and Decruyenaere 1998) as the CAG analysis can establish, with almost complete certainty, whether the individual is or is not carrying the mutated gene for HD (Hayden 2000; HDCRG 1993). If they have a mutation-positive result they will inevitably develop the disease and their children will be at 50% risk of developing the disease later in life. Individuals who have a mutation-negative test result are assured that they will not develop HD and that their children and their future offspring will be at negligible risk of developing the disorder. The only remaining risk would be due to a new mutation risk (Sarangi et al 2005; Van Riper 2005; Taylor 2004; Brouwer-DudokdeWit et al 2002).

Tests can be performed in the following three circumstances:

1. Individuals who have a family history of HD present themselves at a clinic thinking that they may be showing similar symptoms of the disease to that which their relatives manifested. The testing is then performed to confirm the diagnosis or to provide evidence that the individual is mutation-negative and will not develop the disease.
2. When at-risk asymptomatic individuals wish to know whether they are gene-positive or gene-negative for the disease.
3. Prenatal testing is performed to determine the at-risk status of the foetus (Decruyenaere et al 2007; Myers 2004; SuttonBrown and Suchowersky 2003). This is available only to a prospective parent with a positive predictive test result; an affected parent; or a parent with a CAG repeat length between 27 and 39. For ethical reasons it is not available if the parent's at-risk status is not known. If the test of the foetus is mutation-positive the parents have the option of having the pregnancy terminated (Van Den Boer-Van den Berg and Maat-Kievit 2001; Evers-Kiebooms et al 1998; International Huntington Association and World Federation of Neurology 1994).

Individuals presenting for PT are often self-referred at-risk people who have already made their decision to be tested or they have been referred by clinicians dealing with their symptomatic family members (Taylor 2005a).

Several authors recommend that, because of the seriousness of the possible implications of the testing, presymptomatic testing should be done only at genetic centres with experienced geneticists and counsellors where there is appropriate psychosocial support available for the individuals, partners and their families during the testing programme and for up to one year after receiving the results (Tassicker et al 2006; SuttonBrown and Suchowersky 2003; Hayden and Kremer 2001). It is essential for testing centres to be consistent regarding information given about the implications of mutable, normal and reduced penetrance results to avoid confusion and distress for HD families who may have several individuals attending and receiving results in different national and international centres with regards to the interpretation of the CAG repeat sizes. All centres should adhere to the current *Laboratory Guidelines for Huntington Disease Testing (1998)* to avoid this unnecessary uncertainty and stress (Semaka et al 2006; Tassicker et al 2006; The American College of Medical Genetics 1998).

2.19.1 PROTOCOL FOR TESTING

HD is a late-onset disease for which there is no effective treatment and, therefore, predictive test results, whether positive or negative, can have grave irreversible psychological repercussions such as suicide. For this reason several countries have developed detailed national guidelines and the majority of testing centres have their own formal protocols for PT programmes to ensure that the individuals are optimally prepared to make an informed decision about the test options and to reduce the chance of any adverse emotional events from arising (McPherson 2006; Myers 2004; Pakenham et al 2004; Hayden 2003; SuttonBrown and Suchowersky 2003; Evers-Kiebooms et al 2002; Hayden and Kremer 2001; Mandich al 1998; Craufurd and Tyler 1992).

In general, the protocols require the following inclusion criteria before consideration is given to testing an individual:

- A confirmed family history of HD;
- 50% prior risk or 25% when there is a case of adult exclusion testing;
- 18 years of age and above;
- the individual has not been persuaded or coerced to having the test by family members, partners, or employment or insurance agencies; and
- a substantial period of time has lapsed between the individual hearing for the first time of her/his at-risk status and presenting for the PT programme (Ensenauer et al 2005; Lea et al 2005; Sarangi et al 2005; Taylor 2005a; Myers 2004; Craufurd and Tyler 1992);

Most authors recommend a multidisciplinary team comprising a genetic counsellor/clinical geneticist, neurologist, psychologist and/or social worker and/or genetic nurse to provide preparatory counselling sessions. Although there is no need for them all to counsel the individual simultaneously for the sessions, there should be at least two members present when the results are given (Lea et al 2005; Decruyenaere et al 2003; Craufurd and Tyler 1992).

The counselling sessions should be at well-spaced intervals to give the individual sufficient time to reflect on the risks and benefits of a positive result, negative result or deciding not to undergo testing (Sarangi et al 2005; Taylor 2005b; Hayden 2003; Evers-Kieboorns et al 2002; Houlihan 1999; Evers-Kieboorns and Decruyenaere 1998; Craufurd and Tyler 1992). As confidentiality and privacy are major considerations one of the topics that needs to be discussed, in detail, is the possible reactions and implications of disclosing their test results to family members, friends, employers and insurance agents (Bombard et al 2007; Fulda and Lykens 2006; Sarangi et al 2005; Myers 2004). On the one hand by discussing it with trusted persons they can build a social support network, but, on the other hand the information can be misused to their detriment (Evers-Kiebooms and Decruyenaere 1998).

The main aim of the sessions is for the counsellors to ensure that the individual has sufficient knowledge of the disease, testing programme and possible consequences of the test results in order to make an informed decision about having or not having the PT as this knowledge is irreversible (Arnold and David 2005; Hayden 2003; Evers-Kiebooms et al 2002; Evers-Kiebooms and Decruyenaere 1998). Because of the increased levels of marital stress found in couples who test positive, Richards and Williams (2004) and Quaid and Wesson (1995) recommend that the partners should be included in the pretest counselling and that their expectation of the test result, their coping strategies and possible adverse impact on the couple's relationship should also be assessed and explored (Richards and Williams 2004; Soldan et al 2000; Huggins et al 1992). The test results should be given in person together with the accompanying spouse/significant other.

Individuals need to be reassured that their personal attitudes and responses to the scenarios discussed during these sessions will not prevent them from being eligible for the testing. They should also be confident that they do not have to have specific psychological attitudes and qualities before undergoing testing (Sarangi et al 2005). They should be advised that they can withdraw from the testing programme at any stage and that they will be given the opportunity to delay or decline the test results without any repercussions (Taylor 2005a).

It is essential to provide comprehensive follow-up by means of face-to-face support sessions with the testee after the test results are given to increase their perceived personal control of life and to encourage them to actively cope with their problems. Support sessions might also be required for the partners and family members to whom the test results have been disclosed (Decruyenaere et al 2003; Evers-Kiebooms et al 2002; Evers-Kiebooms and Decruyenaere 1998; Lauterbach et al 1998; Van t' Spijker and Ten Kroode 1997; Decruyenaere et al 1996; Craufurd and Tyler 1992). Follow-up sessions for the mutation-positive individuals, gene-negative individuals and partners should be done on a formal basis and should be pre-arranged for one week, one month, three months, six months and annually after the test result (Codori et al 2004; Richards and Williams 2004; Almqvist et al 2003; Craufurd and Tyler 1992) as most adverse events occur in both mutation-positive and mutation-negative individuals within one year of receiving results (Almqvist et al 2003; Pilnick and Dingwall 2001; Lawson et al 1996). Special attention should be paid to mutation-negative individiauls who have feelings of guilt and those who need assistance with planning for their future. Care has to be taken that individuals may appear and report that everything is in order, but that their emotional reactions might be guarded (Van t' Spijker and Ten Kroode 1997).

2.19.2 UPTAKE OF PREDICTIVE TESTING

Individuals at-risk for HD have been described as living "in a state of silent apprehension" (Wexler 1979:2000) or living with a permanent cloud over their heads as the uncertainty is severely emotionally taxing (Taylor 2004; Tyler 1996). They have difficulty in planning for the future or committing themselves to their careers or current relationships because of the uncertainty caused by their at-risk status (Evers-Kiebooms and Decruyenaere 1998; Kessler et al 1987; Mastromauro et al 1987).

The number of at-risk people in all countries who have utilised PT for HD has been much lower than was originally anticipated before the service became available in 1986 (Harper et al 2004; Creighton et al 2003; Hayden 2003; Brouwer-DudokdeWit et al 2002; Evers-Kiebooms et al 2002; Hayden and Kremer 2001; Houlihan 1999; Evers-Kiebooms and Decruyenaere 1998; Houlihan and Harper 1995; HDCRG 1993; Quaid and Morris 1993). The most likely reason for the uptake being only 5 – 25% appears to be the lack of treatment to delay the onset or treat the symptoms of the disease (Shannon 2004; Hayden 2000; Tyler et al 1994). The majority of people at-risk, therefore, avoid the opportunity of confirming their future by means of PT because of the potential for personal and family disruption should the result be gene-positive (Langbehn et al 2004; Myers 2004; Taylor 2004;

Creighton et al 2003; Sobel and Cowan 2000). Van t' Spijker and Ten Kroode (1997) cite an over-intensive assessment and counselling protocol as another reason for people not undergoing the testing programme.

2.19.3 DECISION WHETHER OR NOT TO HAVE TESTING

The individual's decision to have PT as well as the test result can have overwhelming short-term, mid-term and permanent implications for the planning and future life course, health and happiness of the individual, partner and family members as, once the result is known, the knowledge cannot be reversed (Taylor 2005b; Van Riper 2005). Decisions that individuals must face are often multiple and sequential (Emery 2001) and the decisions to undergo PT is usually just the catalyst that elicits more decisions to be made (Wang et al 2004). Because it is a family disease the decision to be tested is rarely based on the individual's needs, but is often a social situation based on the feelings of responsibility and commitment to partners and other members of the family; their values and attitudes towards PT; and the nature of the family experience of HD (Van Riper 2005; Decruyenaere et al 1995a). The individual's decision to have the test or not often can be considered the ultimate symbol by which relatives perceive and judge each other's loyalty to the family beliefs, values and rules (Sobel and Brookes 2000). Decision-making regarding PT is thus dynamic and changes in accordance with the individual's circumstances and life events at the time. Hence at one stage of their lives at-risk individuals may perceive that they are 'not ready' to be tested. This perception of readiness appears to refer to their emotional state, knowledge of HD, stage of life, availability of time and 'space' of themselves and partners. Later when their circumstances have changed and they are able to deal with the emotional and other demands of the testing programme they may become convinced that it would be in their best interests to undergo the test (Taylor 2005a). Decision-making is also influenced by the individual's perception of the onset, cause, predictability, seriousness, progression of the disease and treatment availability (Van Riper 2005).

The counsellor's personal characteristics can play a large part in the individual's decision to undergo testing or not. Although genetic counselling is, in principle, supposed to be non-directive there can be a bias in the amount and type of information provided by a specific counsellor which will encourage decision-making in a particular direction (Evers-Kiebooms et al 2002).

Responsibility

People feel they have certain moral commitments towards their family members, partners and future generations and, therefore, seek PT. This sense of responsibility towards family members is particularly evident when decisions about reproduction are being contemplated, either by themselves or their adult children and they feel there is a need to consider PT. Some individuals who know they are at-risk feel a moral sense of social duty to undergo testing when they enter a serious relationship, so that their partner can be fully informed and decide whether to continue the relationship or not.

Given the same family scenarios responsibility is perceived from different frameworks of reference and decisions are made from the unique viewpoints of the families concerned. Thus the same nominal risk is interpreted differently by and for each member of the family (Kessler 2001). One family member might consider it responsible to terminate a pregnancy to avoid having a child who is gene-positive; whereas another might feel that it is irresponsible to have repeated terminations to ensure a gene-negative child and put the mother's health at-risk as she would not be able to care for her other children adequately (Downing 2005).

Individuals feel they are acting responsibly by having PT so that they can assist their at-risk children in coming to terms with their at-risk status, setting up insurance policies for them and providing for them should they develop the disease. Others justify not being tested as they fear a positive test result would be intrusive and compromise their ability to parent their children.

Individuals who drop out of the testing programme may also consider their actions as being morally responsible in that they have made the necessary arrangements and attended the testing programme in order to make an informed decision about not being tested. They also consider that they have the right not to know (Downing 2005; Sarangi et al 2004; Taylor 2004; Cox 2003; Sobel and Cowan 2000; Hallowell 1999; Quaid and Wesson 1995). The decisions made vary significantly over time as changes occur in their family lives and depend on the individuals' concerned, their life stage and their family circumstances at a specific time. For example, a young mutation-positive couple might consider it responsible to have children by having prenatal testing to ensure that the children are healthy. However, once they have two or three children and they perceive that "time is passing" and the time for the onset of symptoms is drawing closer, they may decide that it would be irresponsible to have further children because of the knowledge that the one parent will be unable to parent another child for any length of time (Taylor 2004).

2.19.4 REASONS FOR UNDERGOING PREDICTIVE TESTING

The main reasons for individuals requesting testing are behaviours they identify in themselves as being symptoms of HD; the need or want to know in order to relieve their uncertainty; to enable them to plan for the future with regards to relationships and marriage; decision-making about careers; to clarify the risk status of their children; and for family planning purposes (Etchegary 2006; Taylor 2005a; Myers 2004; Shannon 2004; Decruyenaere et al 2003 ; Almqvist et al 2003; Hayden 2003; Evers-Kieboorns et al 2002; Hayden 2000; Decruyenaere et al 1996; Tyler et al 1992; Meissen et al 1991; Jacopini et al 1987; Mastromauro et al 1987). Other issues such as concern about insurance and employment rank lower on the underlying reasons for testing (Evers-Kieboorns et al 2002; Tyler et al 1994; Harper 1996). People over 50 years of age requesting tests often do so for the benefit of their adult children who may be considering family planning as much as for their personal information. Others consider it their moral responsibility for all family members to be informed of their status (Taylor 2004).

2.19.5 CHARACTERISTICS OF INDIVIDUALS REQUESTING TESTING

Individuals requesting testing are reported to be a self-selected group who are more resourceful, have more positive coping strategies and are less anxious and depressed than the general population. There appears to be no difference with regard to baseline measures of depression, anxiety and coping strategies between mutation-positive and mutation-negative individuals. Persons requesting testing also have significantly higher ego strength and are more socially extraverted. They believe that they will be able to cope with the social, emotional and psychological outcomes of the test result should it be positive and would enjoy the healthy period of their lives more (Van 't Spijker and Ten Kroode 1997; Decruyenaere et al 1996; Codori et al 1994; Kessler 1994; Van der Streenstraten 1994; Bloch et al 1989). Approximately 60% of people undergoing PT are women (Shannon 2004; Creighton et al 2003, Hayden 2003; Goizet et al 2002; Mueller and Young 2001; Harper et al 2000; Almqvist et al 1999; Tibben 1997a; Simpson et al 1992; Craufurd et al 1989). A reason for this might be that men have a greater capacity to avoid examining the unpleasant implications of the situation and they may be more afraid of developing the disease (Bloch et al 1989). Generally individuals requesting testing are in their mid-30s, married or in stable relationships and have already had children (Creighton et al, 2003; Hayden 2003; Decruyenaere et al 1995b; Hayden 2000). People who undergo testing are more likely to have higher educational levels than the general population (Craufurd et al 1989).

Although persons who drop out of the programme do not differ in their psychological characteristics from those who participate (Decruyenaere et al 2003), they claim that they would not be able to cope with a gene-positive result. This is, therefore, a form of avoidant behaviour (Bradley et al 2004; Decruyenaere et al 1997). Up to 40% of individuals abandon testing after their first visit highlighting the importance of pre-test counselling to ensure that individuals are adequately prepared to make an informed choice (Bradley et al 2004; Myers 2004; Hayden 2003; Duisterhof et al 2001).

2.19.6 REASONS FOR NOT UNDERGOING PREDICTIVE TESTING

The reasons given most frequently for not availing themselves of the opportunity of PT are that there is no effective treatment, the risk of compromising insurance and the possibility of increasing their children's risk if they are found to be gene-positive (Bombard et al 2007; Creighton et al 2003; Evers-Kiebooms et al 2002; Quaid and Morris 1993; Morris et al 1989). Individuals also fear that they might not be able to cope with the psychological and emotional consequences they might experience if they receive a positive result (Brouwer-DudokdeWit et al 2002; Van 't Spijker and Ten Kroode 1997; Babul et al 1993; Tyler et al 1992) and would prefer to live a happier life living with the possibility that they might not have inherited the HD gene (Evers-Kiebooms et al 2002). Many see that a positive test result would cause them to lose the little hope that they could retain and anticipate that they would spend the rest of their lives searching for symptoms of the disease (Lowit and Van Teijlingen 2005; Creighton et al 2003; Evers-Kiebooms et al 2002; Decruyenaere et al 1997; Babul et al 1993). This is particularly so when an individual has been exposed to traumatic experiences with affected relatives (Pakenham et al 2004). Some are apprehensive that having the test would elicit negative reactions in their children or partners should they test gene-positive whilst others have cited the extensiveness of the pre-test counselling sessions as a reason for not undergoing testing (Evers-Kiebooms et al 2002; Decruyenaere et al 1995b; Kessler 1994). Testing may be declined because of religious and philosophical convictions or because individuals believe it is not relevant for their age or stage of the life cycle. Some individuals, for no objective reason, believe that they do not have the gene mutation and so testing is unnecessary (Lea et al 2005; Taylor 2004; Tercyak et al 2001).

The most important reasons for couples not wanting antenatal testing is objection to termination of pregnancy and the hope that a cure will be found before their children develop symptoms of HD (Downing 2005; Lea et al 2005; Evers-Kiebooms et al 2002; Hayden and Kremer 1995). Sobel and Cowan (2003) warn against labelling individuals who do not take up the option of PT as being "in denial" when they choose to live with the uncertainty of being at-risk.

2.19.7 CHARACTERISTICS OF INDIVIDUALS DECLINING TESTING

People who declined testing were found to be significantly more depressed and pessimistic than people undergoing testing. They expected to be mutation-positive and anticipated more negative repercussions from a mutation-positive result. They were also more likely to have learned about their risk of HD when they were adolescent rather than during their adulthood (Van der Steenstraten et al 1994).

2.20 ETHICAL CONSIDERATIONS WHEN TESTING FOR THE HUNTINGTIN GENE

"Ethics is a set of guidelines for morally acceptable conduct within a theoretical framework" (Schmerler 1998:250). Genetic testing has substantial moral and ethical implications for the individual, family and health professionals involved, as genetic information is directly related to the individual's identity and is, therefore, extremely private (Fulda and Lykes 2006). The principles of ethical theories such as utilitarianism and libertarianism form the foundation of the guidelines for professional behaviour of the different health care disciplines (Fulda and Lykes 2006). The ethical issues of testing for the HD gene are underpinned by the principles of autonomy, informed decision-making, privacy and confidentiality, beneficence, nonmaleficence and justice (Schmerler 1998).

Autonomy signifies the principle that the individual is free to make meaningful choices without external controlling interferences (Fulda and Lykens 2006; Schmerler 1998). In the genetic context of HD it is the right of the individual to choose or refuse PT after adequate information has been provided in a non-directive manner (Fulda and Lykens 2006). Autonomy ensures that the decisions made by individuals have been made independently without persuasion or coercion by family members or the counselor, and in accordance with their personal values and preferences (Ensenauer et al 2005; Lea et al 2005). Families frequently have a strong emotional influence on the individual's choice to be tested or not by making it known what their individual attitudes to PT are (Lea et al 2005).

Parents do not have the autonomy to make decisions regarding their children being tested if they are younger than 18 years of age as genetic testing for adult-onset diseases is generally deferred until the individual is at least 18 years of age. It is the child's right not to know until he/she has reached legal majority and can make his/her own free decision. Many fully informed at-risk adults, after due consideration, opt never to be tested and this right not to know is violated if parents are authorised to make the decision for testing on behalf of the child (Duncan and Delatycki 2006; Myers 2004; Taylor 2004; Evers-Kiebooms and Decruyenaere 1998; Craufurd and Tyler et al (1992). However, if an adolescent actively requests PT, Binedell et al (1996) suggest that he/she has the right to know and that it should be done, provided the individual is competent to make the decision about the predictive test. This competency would include an evaluation of the adolescent's cognitive, emotional and social maturity; ensuring that there is no third party coercion; exploring the role of the family context in the decision; and assessing the adolescent's experience and confidence in making major personal decisions.

Prenatal testing allows couples increased autonomy about their reproductive choices as they can decide whether to continue a pregnancy or not, depending on the risk status of the foetus (Ensenauer et al 2005). With regards to the rights of family members who do not wish to know their situation, a positive test result of one individual may reveal the risk status of other biological family members and, therefore, violate their autonomy of not wishing to know their at-risk status (Fulda and Lykens 2006; Ensenauer et al 2005; Evers-Kiebooms and Decruyenaere 1998). On the other hand, according to international guidelines, adults at 25% prior risk have the right to know and should have access to testing even if their at-risk parents do not want to know their status (Benjamin and Lashwood 2000). According to the utilitarian perspective if there is a group of people at-risk they should be informed by either the mutation-positive individual or physician as the information may, in the long-term, increase their quality of life through adequate planning and lifestyle changes and by preventing the disease from being passed onto their offspring. The decision not to have children could also save society large amounts of money which would have been spent on care once the symptoms have developed (Fulda and Lykens 2006; Miller 2002). This situation also arises when individuals do not disclose information to their relatives and deny the relatives the opportunity to seek testing and making informed health choices for themselves such as reproduction risks (Gilbar 2007; Fulda and Lykes 2006; Clarke et al 2005).

As genetic test results have extensive and non-reversible implications for the individuals tested and their families, it is essential that truly informed consent be obtained from the person being tested before the genetic test is performed (Ensenauer et al 2005). This means that, not only must all the relevant information be provided to the individual during the counselling sessions, but the counsellees must have a thorough understanding of the material supplied and the information that has been explained during the counselling sessions (Van Den Boer-van den Berg and Maat-Kievit 2001; Baker et al 1998). It is the responsibility of the counsellor, therefore, to identify barriers such as the counsellees having a different home language or a low level of education, and to endeavour, to the best of her/his ability, to overcome the specific barriers before obtaining permission to have the test performed (Middleton 2005; Baker et al 1998).

Informed decision-making is brought about by means of the discussions that take place between the individual and members of the genetic testing programme before the genetic test is conducted (Lea et al 2005) to ensure a comprehensive understanding of the seriousness and permanence of the test results and the impact of a positive or negative outcome on the individual and her/his family (Van Den Boer-van den Berg and Maat-Kievit 2001).

Privacy and confidentiality of genetic information are complex issues that can have far-reaching harmful effects on the individuals, families, communities and society with regard to health care, insurance and employment (Fulda and Lykes 2006; Ensenauer et al 2005; Lea et al 2005). The relationship formed between the genetic health care provider and the individual is a confidential one where it is obligatory not to share any privileged communication without the person's consent. Individuals waive the principle of confidentiality when they knowingly share confidences in front of a third party who is not a member of the health care team, such as that which occurs when a partner attends the PT programme. Privacy is different from confidentiality and is controlled by the individuals themselves. It refers to the information they choose to share with other persons who are not necessarily health care providers. Any breach of the information in this situation is a violation of privacy rather than that of confidentiality (Schmerler 1998).

Disclosure of an individual's genetic test results can be critical and thus the principle of confidentiality can place the medical practitioner in a vulnerable situation. On the one hand, by keeping the positive test result of one family member confidential it might mean that other family members at-risk for HD may not be made aware of their personal or their future children's risk status. However, certain proponents claim that it is the duty and right of the medical practitioner to alert family members to their status of a late-onset life-threatening disease (Ensenauer et al, 2005; Lea et al 2005; Weil 2000). In these situations the harm due to failure of disclosure should outweigh the harm that may be caused by disclosure if the information is not made available to the family members. Individuals may consider that the information might cause their relatives anxiety or alarm and, therefore decide against disclosing their test result (Clarke et al 2005; Ensenauer et al 2005). On the other hand, disclosure to family members by the medical practitioner without consent of the individual tested violates the individual's privacy and breaks the rule of confidentiality which may negatively affect the counselling process (Fulda and Lykens 2006; Clarke et al 2005). It can also damage relationships and destabilise family values. Any strategies taken by the counsellor to encourage the individual to disclose information to other family members is undoubtedly a form of directive counselling which is against the most basic principal of genetic counselling. Family members also have the right not to know about their risk for HD and so live without the fears of discrimination and social stigmatism that society frequently imposes on those known to have the disease in their family. They may live a happier life without knowing that there is the possibility of them developing symptoms or having to make decisions about relationships, having children and career choices (Bombard et al 2007; Fulda and Lykens 2006).

Beneficence and non-maleficence "is the promotion of well-being in others" (Baker 1998:253) while non-maleficence is an obligation derived from the principle of beneficience, inferring that no harm should be done. The balance of these two principles is assessed to ensure that the decision to have a predictive test will have overall greater good than harmful outcomes for the individuals or their children. Genetic testing for HD, where there is no treatment available, has the potential to cause psychological harm, societal stigmatisation, and discrimination (Bombard et al 2007; Duncan and Delatycki 2006; Lea et al 2005). On the one hand the right not to know about the risk for HD is supported by the principle of non-maleficence, if it were to affect individuals negatively should they know their status. On the other hand if the physician maintains confidentiality of the individual and does not inform the family members who may be at an increased risk for HD, the principle of non-maleficence may be violated as they may have wished to avoid having offspring or may not have planned adequately for their future (Fulda and Lykens 2006). The recommendation for not testing children in most guidelines is because of the potential psychological harm it may cause them should they test gene-positive. However, there is some evidence that testing is occurring in some countries in rare circumstances and there are proponents who claim that by not testing in certain circumstances may fail to promote benefits and that this can cause harm (Duncan and Delatycki 2006; Clarke 1994).

Justice ensures that all people have equal access to and allocation of medical and genetic testing services regardless of their socioeconomic, ethnic and geographical regions (Lea et al 2005; Baker et al 1998). As genetic tests are expensive there is concern that they may become elitist and only accessible to a select group of families who have the financial means or medical insurance to cover the costs of the resources involved in the PT process (Ensenauer et al 2005).

As can be seen from the above review of the literature each situation has to be considered within its unique set of circumstances. This will depend to a large extent on the amount of information regarding the family members that the individual has disclosed to the counsellor or physician and the extent to which the counsellor or physician knows the family members. In one situation the principle of autonomy might be violated while the principle of non-maleficence is supported. In another situation the principle of confidentiality of the individual might be supported at the expense of beneficence to the family members.

2.21 IMPLICATIONS ASSOCIATED WITH PREDICTIVE TESTING

Most studies investigating the psychological consequences of PT have found it to be beneficial for the majority of individuals tested, regardless of whether they tested positive or negative (Almqvist et al 2003; Brouwer-DudokdeWit et al 2002; Horowitz et al 2001; Meiser and Dunn 2000; Bundey 1997; Tibben et al 1997b; Decruyenaere et al 1996; Lawson et al 1996; Wiggins et al 1992). Generally, most individuals who have a negative test result show a marked improvement in psychological health. Long-term follow-up research of individuals who have tested positive shows that their psychological well-being has also been maintained or even improved (Decruyenaere et al 1996; Hayden and Kremer 1995). However, either a favourable or unfavourable result can cause adverse emotional and psychological reactions, and in 10% to 15% of individuals these adverse reactions can be severe such as major depression and hospitalisation for psychiatric illness (Almqvist et al 1999; Taylor and Myers 1997; Lawson et al 1996; Huggins et al 1992).

Decruyenaere et al (2003) and Meiser et al (2000) maintain that psychological tests should be conducted by the counselling team before the predictive tests are done as the psychological state of the individual has a significant effect on the amount of distress they experience with either a positive or negative result.

2.22 ADVERSE REACTIONS FOLLOWING PREDICTIVE TESTING

Individual

An adverse or catastrophic event is considered to be a suicide attempt or suicide ideation; psychiatric hospitalisation or an episode of depression lasting for more than two months; marked increase in alcohol or drug abuse; or breakdown of a significant relationship (Almqvist et al 1999; Lawson et al 1996). It does not include significant levels of distress and anxiety which involve a major emotional adjustment and need for significant support and skilled counselling (Tassicker et al 2006; Codori et al 2004). When PT originally started it was envisaged that there would be a large number of individuals testing positive who would develop adverse events (Ensenauer et al 2005; Shannon 2004; Mueller and Young 2001; Hayden 2000; Hayden and Kremer 1995; Kessler 1994), but this has not occurred (Timman et al 2004;

Almqvist et al 1999). Several authors warn that the dropout rate of people who test positive is a significant problem in long-term studies, and that the longer the follow-up is maintained the more people drop out of the study and the more the test results are likely to be positively skewed (Tassicker et al 2006; Timman et al 2004; Almqvist et al 2003; Bijleveld et al 1999). The high drop-out rate in Timman et al's (2004) study after 10 years was 69% which may, in part, be due to the fact that the suicide rate in HD is increased at the time of onset of the first symptoms (Timman et al 2004; Almqvist et al 1999). A later study by Codori et al (2004) of the 1-year prevalence of depression showed that as many as 20% of those who tested positive experienced depressive symptoms of sufficient severity to warrant further evaluation and possible treatment by a mental health professional.

Other authors have found that approximately 10% to 20% of both mutation-positive and mutation-negative individuals have psychiatric symptoms after PT (Tibben et al 1997b; Bloch et al 1992; Huggins et al 1992; Brandt et al 1989). Those who have tested positive focus on 'symptom watching' and continually seek medical assessments for confirmation of the disease or reassurance that they do not have symptoms (Hayden and Kremer 1995). Those who have tested negative have serious difficulties in adapting to the fact that they have to plan for an unplanned future which they previously thought would be consumed by the disease (Van t' Spijker and Ten Kroode 1997; Hayden and Kremer 1995). It appears that being at-risk has been an important part of their identity, and personality and they have made life decisions based on the belief that they would someday be symptomatic. The news that they are gene-negative causes confusion as the result has not brought the expected effect of relief on their life perspective (Evers-Kiebooms and Decruyenaere 1998).

However, adjustment to results has been found to depend more on psychological adjustment before testing than the testing itself and, therefore, no obvious contraindications have been identified for PT itself (Meiser and Dunn 2000). However, Folstein et al (1983) found that affective disorders in offspring were highly positively associated with similar symptoms in the HD parent and Larsson et al (2006) warn that, based on these findings, psychiatric symptoms of relatives should be taken into consideration during pretest counselling.

Various researchers have studied adverse events at different stages after test results have been delivered. The majority of these adverse events occur in the first year after receiving results (Almqvist et al 2003; Tibben et al 1997b; Decruyenaere et al 1996; Brandt et al 1989). At seven days post-test mutation-positive individuals showed more depression, hopelessness and a decrease in well-being. The depression and hopelessness returned to baseline levels in the first month and the general well-being within six months (Decruyenaere et al 1996; Tibben et al 1994; Brandt et al 1989). In a long-term study of adverse events Almqvist et al (2003) found that in mutation-negative individuals the improvement in the quality of life was maintained for five years while that of the mutation-positive individuals returned to baseline levels after five years.

Psychological reactions such as depression seen in individuals testing positive, and less frequently in those testing negative, may last up to one year after receiving results (SuttonBrown and Suchowersky 2003; Hayden and Kremer 2001). However, different reactions can occur from that which one would expect. 'Survivor guilt' is sometimes found in individuals who are gene-negative (Ensenauer et al 2005; Shannon 2004; Brouwer-DudokdeWit et al 2002) and a mutation-positive result can produce positive reactions of feelings of relief from the uncertainty and empowerment by being able to take charge of their lives and plan for their future (Ensenauer et al 2005; Van t' Spijker and Ten Kroode 1997). However, it can also cause adverse effects in individuals testing mutation-positive as they may feel that control has been taken away from them as the disease will run its own course over which they have no control (Van t' Spijker and Ten Kroode 1997). Likewise mutation-negative individuals may mourn lost opportunities due to previous decisions based on the fact that they would ultimately develop the disease (Downing 2005).

Because individuals often believe themselves to be either positive or negative when presenting for the test, many mutation-positive as well as mutation-negative individuals have difficulty in adapting to their new status when the outcome is contrary to expectations and they can experience major adverse effects (Tassicker et al 2006; Van Riper 2005; Myers 2004; Taylor 2004; Brouwer-DudodeWit et al 2002; Meiser and Dunn 2000; Van t' Spijker and Ten Kroode 1997). These adverse effects are not reflected in the findings of studies based on average group responses showing overall positive psychological adjustment (Tassicker et al 2006). Brower-DudokdeWit et al (2002) found that the reaction to the test result is influenced by the meaning of the disease to the individual. If the family have been able to deal with affected relatives in a matt-of-fact way, then the impact of a positive result is easier to accept. Those with an unexpected negative result sometimes experience stress in their marriage relationships that has previously been attributed to their HD risk (Richards and Williams 2004). Others have made irreversible reproductive decisions or career choices on the basis of their belief. Some individuals who have learned of their HD risk status during adolescence at the time of their identity formation and who test negative, also have adjustment difficulties severe enough to warrant post-test psychological support (Tassicker et al 2006).

Intrusive thoughts and avoidance behaviour are found more frequently immediately after the test result and gradually decrease over time. Mutation-negative individuals have less avoidance and intrusion immediately after the test result, reach a maximum after a few years, but then decrease and in the long-term they have less avoidance and intrusion. Timman et al (2004) suggest that there may be increased avoidance behaviours in mutation-positive individuals who may not return for follow-up appointments to the genetic centre as they wish to avoid seeing the people who were involved in their testing. This avoidance behaviour of mutation-positive individuals can be considered as a beneficial choice to escape from pessimism and negative consequences of the test result. It is a strategy to keep stress and anxiety at a manageable level (Decruyenaere et al 2003).

In mutation-positive individuals there is increased hopelessness immediately after the result which declines after a while and then increases again as the onset of the disease approaches or possibly as a result of the disease itself (Timman et al 2004; Duisterhof et al 2001; Horowitz et al 2001).

Bundey (1997) explained the low rate of adverse outcomes as an outcome of the careful protocols followed by the various testing centres, but Evers-Kiebooms and Decruyenaere (1998) caution that the supposed low incidence may be partly due to denial of distress.

Couple relationships

Partners of mutation-positive individuals receiving predictive test results often experience sadness, anger and despair, but for the majority of mutation-positive and mutation-negative individuals there is no adverse impact on the relationship, although there are a few cases where there is a breakdown of the marital relationship (Richards and Williams 2004; Tibben et al 1997a; Quaid and Wesson 1995). Adverse effects appear to occur more frequently in mutation-positive-partner relationships than that of mutation-negative partnerships (Decruyenaere et al 2004; Richards and Williams 2004; Tibben et al 1997a; Quaid and Wesson 1995; Codori and Brandt 1994) and partners generally show similar patterns of distress as the mutation-positive individuals themselves (Van t' Spijker and Ten Kroode 1997). Richards and Williams (2004) found that there is no difference between the level of marital adjustment in couples who had undergone testing and those who had not.

The mutation-positive individuals appear to cope by restricting themselves to living day-by-day, but their partners are much more concerned and distressed about the future with regards to caring for the affected partner, raising children and the fact that the children might also eventually develop the disease (Decruyenaere et al 2005; Brower-DudokdeWit et al 2002; Sobel and Cowan 2000; Codori and Brandt 1994). Partners of mutation-positive individuals with children are significantly more distressed than those without children, while partners of mutation-negative individuals are significantly less distressed than the mutation-negative individuals after three years, regardless of whether they had children or not (Duisterhof et al 2001; Tibben et al 1997a). After three years mutation-positive individuals and their partners showed more avoidance behaviour, intrusion and hopelessness than partners of mutation-negative individuals (Duisterhof et al 2001; Tibben et al 1997b). Tibben et al (1997b) alert readers to the fact that partners of mutation-positive individuals frequently deal with their problems stoically and do not seek professional support. Partners, generally, may also have difficulty in adjusting to unexpected test outcomes. Those who had planned a caring responsibility for the affected person and their children may feel insecure about their relationship when they realise that their partners will not need that support in the future (Downing 2005).

Family

Test results can have a substantial impact on 'family membership', that is, who is considered to be part of the family and how they relate to each other (Atkinson et al 2001). Bonds between siblings can become divided when test results are different or when one sibling tests negative and the other does not undergo testing as their shared 'bond of risk' is broken (Meiser et al 2001). These 'bonds of risk' may also be altered and have an affect on relationships between parents and children and even more distant relatives (Duncan and Delatyicki 2006). When a 'preselected' individual tests mutation-negative it can also have an adverse affect on relationships as preselection is a coping strategy families use to deal with uncertainty and anxiety and the individual has been raised throughout childhood in a "sick-role" (Van t' Spijker and Ten Kroode 1997; Kessler 1988). Some individuals think that their personal risk is greater once a sibling is tested mutation-negative and might, therefore, blame the healthy sibling for increasing their risk and distance themselves from them (Van t' Spijker and Ten Kroode 1997; Fox et al 1989). When individuals are tested mutation-positive they are sometimes treated as though they are already dying and so lose their role in the family while those who test mutation-negative are sometimes excluded from the family. If an individual in a family, where there are several members who have the disease or are at-risk of developing it some day, is found to be mutation-negative they are perceived to be different and not share the same characteristics of the family. Some families deal with their grief of knowing that a member has tested mutation-positive by either distancing themselves from each other or increasing their connections between members (Sobel and Cowan 2003). Intrafamilial relationships may be influenced when an individual is tested as there may be changes in the anticipated future patterns of care giving (Sobel and Cowan, 2000).

PT has to be viewed from a family perspective and not from that of an individual (Sobel and Cowan 2003). It reflects the disease probability and risk factors in other blood relatives, some of whom may not wish to know this information. Genetic testing of a healthy parent may disclose that the children have a predisposition to the disease and this may lead to a significant change in the way they raise their children and their ensuing quality of life (Ensenauer et al 2005; Sobel and Cowan 2000). Should the information regarding the parent's genetic status become public knowledge, it might also have implications on the child's ability to obtain insurance and employment later in life. Family members may also inadvertently provide information on an affected family member without having been authorised to do so (Ensenauer et al 2005).

Family members who test mutation-negative as well as those who test mutation-positive may experience guilt. Those who have children feel guilty because they might have passed on the mutated gene to their children. Individuals with CAG repeat lengths in the intermediate range also experience guilt as they know they will not develop the disease, but that their children remain at-risk of developing HD. Children who learn that they are mutation-positive feel guilty because the unaffected parent will have to witness their symptoms and ultimately death. If they have children themselves they may feel guilty about the risk of passing on the disease to them as well as not being able to be a parent to the children because of symptoms that they will develop. Those who test mutation-negative may experience guilt when they know that a sibling has tested mutation-positive (Semaka et al 2006; Sobel and Cowan 2003; Maat-Kievit et al 2001).

Prenatal testing

Knowing the child's at-risk status may have a negative effect on the childrearing practice of the parents by overindulgence, overprotection or possible neglect of the child. In addition the child's right not to know will be violated even before her/his birth. All these issues should receive attention during counselling before the parents engage in a pregnancy with prenatal diagnosis (Evers-Kiebooms and Decruyenaere 1998).

Discrimination

Genetic information can be used to discriminate against individuals who have had PT and their families in spite of there being no phenotypical differences. This is particularly so when affected relatives of those at-risk have had to be institutionalised (Taylor 2004). People are refused insurance, forced to pay additional premiums or losing their current insurance coverage should they or their family members undergo testing (Blain and Brooks 2007; Bombard et al 2007; Lea et al 2005; Tassicker 2005; Van Riper 2005; Harper et al 2004; Smith et al 2004) and there has been a report of employers being allowed to introduce gene testing for job applicants in future (Burgermeister 2004). Predictive test knowledge can also be a disadvantage with regard to family law and child custody (Tassicker et al 2006).

2.22.1 PREDICTORS OF ADVERSE OUTCOMES

A history of depression before PT increases the risk of an adverse outcome, regardless of whether the result is favourable or unfavourable (Blain and Brooks 2007; Pakenham et al 2004; Shannon 2004; Hayden and Kremer 2001; Meiser et al 2000; Brouwer-DudokdeWit et al 1998; Evers-Kiebooms and Decruyenaere 1998; Decruyenaere et al 1997). Individuals who are unemployed are also at-risk of a less satisfactory outcome after testing (SuttonBrown and Suchowersky 2003; Decruyenaere et al 1997). Adverse outcomes are more likely in those who receive a test result contrary to what they had previously expected (Decruyenaere et al 1997). Those with a history of adverse advents prior to entering the PT programme and who receive a positive result are also more likely to be associated with an adverse event after testing (Almqvist et al 2003).

The process by which the individual develops a sense of self, known as identification, can have a negative effect on the test outcome, depending on whether they identified with the affected or unaffected parent and believe that they have the same HD genetic make-up as the same sex parent because of similarities in appearance or nature. As knowledge of HD becomes integrated into the individual's psyche over a long period of time if they have been raised in a family with an affected parent, so it can take a long time for the reality of a different test result from what they were expecting to be integrated and internalised and this can have adverse outcomes (Tassicker 2005).

There is a subgroup of people tested in which psychological distress remained high after testing or even reappeared after some years after testing. These were mutation-positive or mutation-negative individuals whose reasons for testing were simply to relieve the uncertainty of not knowing and they had no specific reason such as needing to know for family planning, relationships or career planning (Decruyenaere et al 2003).

2.23 GENETIC COUNSELLING

"Genetic counselling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following:

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
- Education about inheritance, testing, management, prevention, resources and research.
- Counselling to promote informed choices and adaptation to the risk or condition" (Resta et al 2006:77)

Genetic counselling is generally provided by neurologists, psychologists, social workers; genetic nurses, medical geneticists and genetic counsellors (Ensenauer et al 2005; Lea et al 2005; Decruyenaere et al 2003; Craufurd and Tyler 1992).

2.23.1 GOALS OF COUNSELLING IN PREDICTIVE TESTING

The goals of counselling in PT are to

- Ensure that individuals comprehend that the test results will stay with them throughout their lifetime;
- Ensure that individuals are comprehensively prepared to make an informed choice about the testing (Shiloh et al 2006; Myers 2004; Houlihan 1999);
- Ensure that the individual has not been coerced, persuaded or pressurised by a person or institution to undergo PT (Decruyenaere et al 2007);
- Ensure that individuals have balanced the realities of HD inheritance with their religious and personal beliefs; personal wishes and family goals (Decruyenaere et al 2007; Miller 2002);
- Discuss the benefits and limitations of the test, including limited detection rates of testing (Arnold et al 2005; Ensenauer et al 2005);
- Enable individuals tested to disclose their condition and alert other family members to their inheritance risks, and support them in the decisions that they make (Miller 2002);

- Offer opportunities for partners, couples and families to be included in some genetic counselling sessions to explore their expectations of the test outcome and their coping strategies (Richards and Williams 2004; Miller 2002);
- Ensure individuals have carefully considered the details of the impact both negative and positive testing outcomes will have on themselves, their partners and families (Shiloh et al 2006; Ensenauer et al 2005; Bennett et al 2003; Pilnick and Dingwall 2001; Houlihan 1999);
- Explore the couple's attitude to therapeutic termination of pregnancy (Bradley et al 2004; Hayden and Kremer 2001); and
- Ensure informed consent for testing and procedures is obtained and that the counselling sessions have discussed information about:
    - HD, its progress and its treatment options;
    - The transmission of the disease;
    - Tests for prediction and confirmation of diagnosis;
    - Prenatal testing and risks to mother and foetus;
    - Procedure for giving information about tests results;
    - Views about termination of pregnancy;
    - Implications for future and existing children and other family members;
    - Implications for employment and insurance; and
    - Assisting individuals adjust psychologically to the genetic information received to minimise psychological distress and increase personal control (Arnold and David 2005; Ensenauer et al 2005; Harper 2004; Bennett et al 2003; Miller 2002; Biesecker and Peters 2001; Van Den Boer-van den Berg and Maat-Kievit 2001; Shiloh et al 1997).

Miller (2002) maintains that there is a difference between giving information and discussing the disease with the patient, and that which generally occurs in counselling. With counselling the implications of the information provided are discussed between the counsellor and the individual within the context of the individual's understanding, beliefs and values. Through the counselling process the individual is encouraged to reflect on his/her thoughts, emotions and possible reactions given the different scenarios, namely, should he/she decide not to undergo the test; the result be positive or negative; develop clinical symptoms in the future; or should clinical symptoms not appear in the future (Shiloh et al 2006; Ensenauer et al 2005). Studies have shown that individuals who become intensely engaged, both cognitively and emotionally, with their at-risk status during these reflective sessions and who receive an unfavourable result adjust better than individuals who only engage cognitively and not emotionally (McAllister 2001; Binedell et al 1998; Brouwer-DudokdeWit et al 1997).

It should be made clear to the individuals that the predictive test will only indicate whether they are gene-positive or mutation-negative, but it cannot reveal at what age the symptoms will appear should the test result be positive (Myers 2004). Should they test mutation-positive they will need continued counselling, support and reassurance that they have been tested as mutation-positive, but that that does not mean that they have been diagnosed with the disease (Hayden and Kremer 1995) and that, on average, individuals who inherit the mutant gene, spend an average of two-thirds of their shortened lives in a healthy state before developing any symptoms of HD (Harper 1992). Horowitz et al (2001) warn genetic counsellors that approximately 50% of individuals who undergo PT already have symptoms of the disease, although they are unaware of it, and that these individuals may be depressed and at-risk of harming themselves at the time of seeking testing.

Couples who decide that they do not wish to put their children at-risk for inheriting the disease can be informed about the possibilities of adoption; artificial insemination by donor if the husband is at-risk; surrogate gamete in vitro fertilisation if the wife is at-risk; or embryo genotesting if they have funding and if the service is available (Bradley et al 2004).

If an individual has a history of depression or is unemployed the counsellor needs to be particularly careful about the counselling process to minimise the chances of adverse events, regardless of whether they have positive or negative results. If necessary, they should be referred for further counselling by a psychologist or psychiatrist (Ensenauer et al 2005; Shannon 2004; SuttonBrown and Suchowersky 2003).

When an individual has strong feelings regarding their at-risk status and they receive another result, they require a significant amount of counselling too for the reality to become internalised and a different future envisaged from the one they had been anticipating. If they test negative there is the fear that they may be abandoned by their family who have the disease or who have made the decision not to undergo testing. On the other hand, if their test result is positive they may also feel threatened by being abandoned by their partner. This is particularly so if the individuals have observed the affected parent being abandoned during the course of the illness or have recalled feeling abandoned by a parent throughout their childhood and eventually by their deaths (Tassicker 2005).

Several authors have suggested that the low rate of catastrophic psychological and emotional outcomes after testing, with a positive or negative result, are due to the extensive pre- and post-test counselling provided by the genetic testing centres (Blain and Brooks 2007; Tassicker et al 2006; Myers 2004; Sarangi et al 2004; Shannon 2004; Soliz-Perez et al 2001; Evers-Kiebooms and Decruyenaere 1998; Bundey 1997; Kessler 1994) indicating that genetic counselling facilitates clients' coping with and adapting to genetic risk information (Biesecker and Peters 2001; Biesecker 2000). In the USA where programmes exist where there is no PT protocol there are many unpublished reports of psychological distress arising from results being given telephonically or without counselling and support before and after PT (Tassicker et al 2006; Hayden 2000).

By virtue of the title "genetic counsellor" and its concomitant connotation of receiving psychological counseling, there is increasing recognition that in addition to being an important source of genetic information to clients, genetic counselling plays a key role in providing psychological support to clients. This is particularly so in the first few months after the results have been received in order for successful psychological adjustment for individuals with positive outcomes or whose results have been contrary to their personal beliefs of their at-risk status (Van Riper 2005; Davey et al 2005; Myers 2004; Taylor 2004; Hayden et al 2001), and should continue for as long as it takes the individual to reach a new psychological equilibrium (Van t' Spijker and Ten Kroode 1997).

After the test result an essential component of counselling is in assisting spouses of mutation-positive individuals to deal with feelings of guilt and role shifts (Kessler 1993). Interventions also have to be planned to promote coping with gene-negative results in individuals who live within at-risk families. They have to learn how to develop new self-definitions and relationships with family members and new roles in society (Williams et al 2000b). Families whose members test positive need to be prepared for the different grief responses among family members and the lack of social support and isolation they may experience. There should also be support provided for family members who may be encouraged by the family to be tested, but who choose not to be tested (Sobel and Cowan 2003).

Lea et al (2005) maintain that, in genetic counselling, it is critical that the information provided and discussions are conducted in a language in which the counsellees are comfortable with communicating.

In "minimal risk" situations, such as technological failure or the risk of an intermediate allele result, as can occur in PT for HD where there is a "chance" of an outcome being out of the ordinary, to avoid confusion, it is advisable for the counsellor to refrain from dwelling on this uncertainty and to focus on information which is based on certainty (Semake et al 2006; Van Den Boer-van den Berg and Maat-Kievit 2001).

Davey et al (2005) recommend that it would be beneficial for genetic counsellors to establish links with other health professionals and allied health services to assist in meeting those clients needs which cannot be met within the genetic service setting. These would include aspects such as comprehensive information about the clinical symptoms and course of the disease; overall management of affected family members to enable their independence for as long as possible and to deal with their day-to-day activities such as exercise, eating, swallowing, general functional and mobility difficulties.

2.23.2 NONDIRECTIVE COUNSELLING

"Nondirectiveness refers to any procedure used in genetic services that promotes the autonomy and self-directedness of the client" (Kessler 2001:188). It is an active strategy to assist clients to achieve their personal health-related goals by assisting them to think through the various options available to them and their families to make relevant decisions. It gives them an opportunity to consider what the choices are and to explore what meaning they have for themselves and their families in the short- and long-term. They are able to identify obstacles and to consider strategies to minimise them so that are able to make decisions autonomously (Kessler 2001).

Many articles on nondirective genetic counselling do not define the term "nondirectiveness", but describe it as a guiding principal of practice or a primary goal to be aspired to for genetic counselling and as the preferred style of counselling that should be used during a counselling session in order to develop a unique individualised two-way interaction between the client and counsellor (Weil et al 2006; Kessler 2001; Weil 2000; Bartels et al 1997). Thus the terms have been used differently, but in essence, they describe genetic counselling as being noncoercive, nonprescriptive, and value-free where the counsellor remains neutral and refrains from advising the counsellees what to do or making therapeutic recommendations (Blain and Brooks 2007; Visser and Bleiker 1997; White 1997). The counsellor and the counselling process should not impact the personal, cultural, ethical and religious beliefs and values of the counsellee, but should support the counsellees' decisions unequivocally by being empathic and non-judgemental, regardless of their personal attitudes, values and beliefs (Weil 2000; Bartels et al 1997). If counsellors do not agree with the counsellees' decisions and distance themselves emotionally it may cause the clients to feel abandoned (Hodgson and Spriggs 2005).

In spite of the goals of nondirectiveness and autonomous decisions the counsellor has to make a conscious effort in providing a balanced objective presentation of all the information and be sensitive to and respect the clients' values when making decisions after receiving the genetic information. Neutrality is lost when there is overemphasis of selected information; information is focused on negative consequences of certain options omitting to provide possible positive consequences; or when the counsellors' personal beliefs influence their counselling. Neutrality can also be affected by the counsellor's body language, tone of voice and choice of words (Weil 2000; Bartels et al 1997; White 1997).

Initially nondirective counselling involved clients having to make reproductive decisions and the approach was also most appropriate with the evolvement of genetic counselling into many different areas. However, nondirective counselling has many problems associated with it. There are situations where providing direct advice can be considered nondirective. For example, a counsellor may have a great deal of experience in specific medical conditions and by providing this information to the client might enable the client to make a well-reasoned decision that he/she would not have been able to do without the additional knowledge of the counsellor's experience (Hodgson and Spriggs 2005: White 1997). In other situations where no advice or direction is provided, in order to comply with the principle of nondirectiveness, the client gains nothing from the session and it can be considered to be poor counselling. By being inhibited by the requirement of nondirectiveness the counsellor may not be able to use the full range of genetic counselling knowledge and skills to become actively engaged emotionally (Weil et al 2006; Biesecker 2000; Kessler 1997).

There need to be goals and direction set for each session so that counselling does not become directionless (Hodgson and Spriggs 2005; Weil 2000). If the counsellor does not question the client's beliefs it may not meet the requirements of facilitating the client's autonomy as the client may not have understood an adequate amount of relevant information; may not have adequate skills to apply the information to his or her own life situation, values and wishes; or may lack decision-making skills (Hodgson and Spriggs 2005). In certain clinical situations the counsellor may be required to make essential recommendations of treatment or screening based on the particular clinical circumstances of the client which means that the counsellor would not be adhering to a strictly nondirective counselling style. This same argument would apply to exploring ethical and moral issues as they have profound individual and societal meaning and implications (Weil et al 2006; Hodgson and Spriggs 2005; Elwyn 2000; Bartels et al 1997; Clarke 1997). In addition, not all countries consider nondirective counselling appropriate in all situations (Marteau et al 1994).

It is not possible to be totally nondirective in the choice of information included and excluded from a counselling session as information has to be tailored to the specific clients with regards to the their level of knowledge gained by living in a family with the disease or not having had any previous contact with affected persons; level of education; language proficiency; and economic circumstances. Directive counselling tends to be used when recommending genetic or medical testing and in recommending medical treatment when it is available and relevant to the clients own health or that of the clients' children (Bartels et al 1997). This is considered to fall into the category of beneficence/non-maleficence of the ethical principles of practice.

There is a difference between directiveness in process as opposed to directiveness in outcome. When clients are unable to make a decision the counsellor can be directive in telling them how to go about making their decision, but should be not be directive about what the decision should be (Bartels et al 1997). Several authors argue that, in rare situations, when the client is unable to make a decision, it is permissible for a counsellor to disclose personal biases and values if it assists the counselling process and the decision making of the client (Brunger and Lippman 1995; Wachbroit and Wasserman 1995), but that the timing of this should be so that it does not lead to premature closure of the client's decision-making process and does not unduly influence the decision outcomes. Most articles on genetic counselling indicate that what the counsellor decides to do is irrelevant to the client's decision since the values and circumstances of each are unique (Bartels et al 1997).

There are also situations when the clients expect, based on their cultural backgrounds, to be advised and told what to do and counsellors have had to be directive about decision outcomes as opposed to the decision-making process. However, Bartels et al (1997) warn that this should not be done automatically, but that counsellors should make every effort to refer the client to other resources such as a religious or community leader to assist with the decision-making first before using the directive approach to ensure that the decision is consistent with the client's values and the cultural context in which she or he lives.

In view of the many dilemmas that nondirectiveness brings as a counselling style or technique, many authors suggest that it be considered only as a value to guide genetic counselling practice or as a paradigm for them to assist clients to make autonomous choices (Weil et al 2006; Hodgson and Spriggs 2005). At the National Society of Genetic Counsellors Annual Education Conference (2003) several additions to nondirectiveness were recommended in order to improve the effectiveness of genetic counselling sessions (Weil et al 2006). These included satisfying the wants and needs of the counsellees; interacting with the counsellees to facilitate active decision-making; identifying and processing genetic counsellors' personal and professional values and experience; and flexibility in the use of different counselling techniques and the application of nondirectiveness (Weil et al

2006; Biesecker and Peters 2001; Kessler 1997; Weil 2000; White 1997). The recognition of flexibility is particularly appropriate since genetic counselling has moved from its historical role in reproduction only to counselling in diverse medical settings such as PT for late-onset neurogenetic diseases; prenatal diagnosis; cancer risk counselling; at-risk status for genetic diseases in general; and neurogenetics (Weil et al 2006).

The above findings indicate that generally non-directiveness is the desired approach to maximise client understanding of information and their personal values so that their decision-making can be autonomous. However, in rare situations, the directive approach may have to be used in order to minimise potential harm to the clients or their children.

2.23.3 RISK PERCEPTION

One of the most important goals of genetic counselling is to calculate and inform clients of their risk status and that of their family members in a manner that is tailored to the individuals' needs and that they are easily able to comprehend. As the term "risk" implies a negative connotation, it should be avoided and more neutral words such as "chance", "likelihood" and "probability" should be used in the discussions. When providing objective risk estimates the person's educational level has to taken into consideration. People with little formal education may not be able to interpret abstract probability estimates such as 1/10.000. Changing the numeric data to fractional risks (1 in 4 chance) or percentages (50%) makes the information more meaningful. Words such as "very', "often", "low", "never" and "high" should also be avoided when referring to numerical figures due to the subjectivity of interpretation (Brunger and Lippman 1995). Even if the individual has achieved a good level of education, risk information should be presented in several different ways for optimal cognitive processing and emotion impact (Middleton 2005; Baker et al 1998). The probabilities of carrying the mutation or not carrying it should both be presented to the individual so that both outcomes receive the same amount of attention and the individual has a better chance of understanding the concept (Baker et al 1998). Multiple risk figures presented in conditions for being a carrier of the mutation and the reproductive risk often cause confusion and it may take considerable time to clarify the concepts (Weil 2000; Baker et al 1998).

In late-onset diseases such as HD certain individuals may perceive they are at a greater risk for inheriting the disease as the family dynamics might have "preselected" a particular family member as being the one most likely to inherit the disorder. There is often also confusion about the number of siblings who will develop the disease. Siblings may have difficulty understanding that the risk of carrying the HD mutation is the same until a predictive test has been performed or a diagnosis has been made. They sometimes believe that because two of their siblings have tested mutation-positive they will be mutation-negative (Weil 2000). Individuals view risks differently depending on their experience of family members who have had the disease (Baker et al 1998).

The explanation of Mendelian inheritance can be aided by visual diagrams and tactile objects illustrating the patterns of inheritance (Weil 2000; Baker et al 1998). Counsellees can be asked to restate what has been discussed at the session and that which needs to be passed on to the family in their own words. This assists in clarifying concepts immediately if there is confusion and consolidating the information discussed (Weil 2000; Baker et al 1998). Kupst et al (1975) found that patient restatement with feedback from the counsellor is the most effective means of increasing long-term patient recall.

2.23.4 RECALL OF INFORMATION

Recall of information requires an intact memory, thorough understanding of information and satisfaction with quality of treatment. Between 40-80% of medical information provided by healthcare professionals is forgotten immediately (Kessels 2003) and the more content provided the less is correctly remembered (McGuire 1996). Of the information that is remembered nearly 50% of it is incorrect (Anderson et al 1979). Individuals retain more information immediately after they are given it rather than after one week and even less after four weeks. (McGuire 1996). When medical information evokes anxiety it interferes with the processing of information and not all information receives the same amount of attention and is remembered to the same extent (Kessels 2003; Hammond 1998). For example, when an individual is informed of her/his risk status for a genetic disease, this aspect becomes the primary focus and limits the attention paid to other aspects of information such as prognosis, treatment options and follow-up appointments (Visser and Bleiker 1997). Accurate recall also depends on the nature of the information. Michie et al (1997b) found that aspects related to family matters were recalled 100%, but there was only 68%-78% recall of issues related to genetic or medical information. If simple language is used in explanations it will be remembered more accurately than when it is given in medical terms with complex numbers included (Kessels 2003) and vital information may need to be repeated several times in order for it to be remembered (Reynolds 2004).

The method of providing medical information also affects the amount of recall. In individuals who have a good educational level and whose home language is the same as the message, verbal communication is not as effective a means of imparting information as written text for facilitating recall (Blinder et al 2001; Thomson et al 2001). However, for individuals who are illiterate or whose home language is different from the message, visual aids such as clear cartoons displayed during the verbal communication of the information is most effective (Houts et al 2001).

2.24 EVALUATION OF THE EFFECTIVENESS OF GENETIC COUNSELLING SERVICES

Several studies have emphasised the importance of investigating the effectiveness of counselling services in meeting counsellee needs (Bernhardt et al 2000; Clark et al 2000; Collins et al 2000; Jay et al 2000; Clarke et al

1996) and identifying aspects of the service which require improvements to optimise benefits to the counsellees (Biesecker and Peters 2001; Pilnick and Dingwall 2001; Lea 1996).

One of the aims of counselling is to provide counsellees with sufficient medical information regarding the diagnosis, prognosis, course of the disease and current available management to make informed decisions. Studies evaluating the level of recall of information and level of understanding of risk status have been conducted by measuring client level of knowledge and understanding after counselling (Bernardt et al 2000; Berkenstadt et al 1999; De Pina-Neto and Petean 1999). They found that, generally the level of understanding was inadequate as only 40% to 80% of information was retained (De Pina-Neto and Petean 1999; Clarke et al 1990; Somer et al 1998; Abromovsky et al 1980), and that it was significantly higher in individuals with a higher social class and educational level (Eggers et al 1999). There was also a definite inverse relationship between the amount of recall and time interval between the counselling interview and follow-up interview as well as a decrease in knowledge of genetic disease in the family with the progress of time (De Pina-Neto and Petean 1999). Comprehension was found to be higher among individuals who had a family member affected by or deceased from the disease (Eggers et al 1999).

Other studies have focused on the effectiveness of providing information by measuring the impact on potentially reducing affected births (Somer et al 1998; Hildes et al 1992; D'Amico et al 1992). Although almost half of the cohort in one study reported that counselling sessions had influenced their reproductive plans, the approach of measuring effectiveness by reduction in affected births is controversial because of its eugenic nature (Pilnick and Dingwall 2001). In a study investigating which aspects were found to facilitate reproductive decision-making, Shiloh et al (2006) found that information about consequences of options and about possible controlling actions were found to be most useful by counsellees.

Studies examining the impact of counselling on mood change found that there were reduced adverse psychological events when patient expectations were met in the counselling sessions (Michie et al 1997a).

Berkenstadt et al (1999) and Shiloh et al (1997) used perceived personal control (PPC) as an outcome of genetic counselling as it includes the ability to cope with a stressful event in a manner that reduces its perceived threat; the ability to alter the physical characteristics of the threatening event so that its actual threat is reduced; and the opportunity to choose amongst options. They found that PPC is positively associated with knowledge, satisfaction and general evaluations of the genetic counselling session. As the goals of genetic counselling include enabling the individual to cope and adapt to their at-risk status (Biesecker and Peters 2001) and the main aim of undergoing genetic testing is to reduce uncertainty so that the individual and her/his family can cope with the threat of the disease (Clark et al 2000), a measure such as the PPC reflects how effectively the individual is coping with their particular situation (Wang et al 2004).

2.24.1 PATIENTS' EXPECTATIONS OF GENETIC COUNSELLING

Some studies report that individuals often have few expectations of genetic counselling and are uncertain of the role of the genetic counsellor (Bernhart et al 2000); therefore, measuring client satisfaction is difficult (Davey et al 2005). Although it is assumed that it is important to meet clients' needs, little is written about identifying the most essential needs to address during the counselling sessions (Wang et al 2004). One study that looked at specific outcomes such as anxiety, concern about the medical problem and satisfaction with information provided, found that counsellee expectations during counselling were information, reassurance, advice and help in making decisions. There was a greater reduction in concern and anxiety levels when individuals' expectations for reassurance and advice were met (Michie et al 1997a).

In a study on women at high risk for breast cancer many of them considered that emotional support and counselling was an essential part of post-test counselling (Jay et al 2000; Audrain et al 1998).

2.24.2 GENETIC COUNSELLING PROCESSES

Evaluating genetic counselling services involves more than variables such as the psychological and educational outcomes and the impact on reproductive decision-making. The services may have shortcomings with regard to the way in which genetic counselling is provided, namely, the processes of counselling (Wang et al 2004; Pilnick and Dingwall 2001). These, according to Bernhardt et al (2000) are the most appropriate outcomes of genetic counselling for conditions such as HD where there is a valid predictive test available, but no effective treatment. They would include aspects such as the manner in which the information was conveyed by the genetic counsellor, the extent to which the counsellor was concerned about the problems of the counsellee and whether the individual perceived that the counsellor was interested in her/his particular problems.

Various studies have been done on measuring the level of satisfaction with genetic counselling services which have found a general high level of satisfaction (Michie et al 1997b; Clarke et al 1996; Shiloh et al 1990). However, Clarke et al (1996) warns that assessments of global levels of satisfaction are not beneficial in identifying the strengths and weaknesses of the service, but recommend that specific aspects of counsellor performance should be evaluated such as the comprehensive Satisfaction with Genetic Counselling Service Scale developed by Shiloh et al (1990).

Although it might be expected that there would be a high level of dissatisfaction associated with a mutation-positive result (Shiloh et al 1990), Green and Murton (1996) found that more than 50% of their sample indicated that they were very satisfied with the way in which the result was given to them showing that it is possible to deliver negative news in a satisfactory manner. They found that recall; understanding information; obtaining adequate information; presence of a support person; and level of education were also important aspects to be considered when interaction took place. In other words what is required is a genetic counsellor who displays empathy and sensitivity to the individuals' emotional and informational needs (Green and Murton 1996).

If a genetic counselling service is to be evaluated it is only appropriate to do so once the client has sufficient experience of the service to make a reasonable judgement about the quality of its processes (Clarke et al 1996). For HD this would be one year after the results have been given, once the individuals' psychological status has adjusted and stabilised (Almqvist et al 2003; Tibben et al 1997a; Decruyenaere et al 1996).

2.25 CAREGIVING

"A caregiver is any person looking after another, resulting in the caregiver losing personal freedom" (Ayling, 1993: 780). A family carer is a person who, "assumes a cardinal role in looking after a relative with a chronic illness or disability" (Schofield and Bloch, 1998: 405).

The spouse, mother or immediate family members living in the same household usually automatically takes on the role of caregiver (Aubeeluck and Buchanan 2007; Aubeeluck 2005; Boeije et al 2003; Helder et al 2002; Miles, 2002; Abu-Habib, 1997; Petersen 1991) to assist the person affected with HD with activities of daily living and, as the disease progresses, to carry out basic nursing skills. Although most carers appear to have had to take on the caregiving role without choosing to do so and, generally, without having received any training for the tasks involved, they deal with the problems of their relative's HD by accepting the disease and by developing strategies to deal with the problems as they present and progressively become worse (LoGiudice and Hassett 2005; Lowit and Van Teijlingen 2005). The motivation to undertake the caring is provided by family loyalty; affinity for their partners and children, public expectations, financial difficulties and inadequate professional facilities (Boeije et al 2003; Twigg and Atkin 1994; Aronson 1992). Financial difficulties and inadequate professional facilities sometimes result in children and adolescents having to have the dual role of young caregiver and student. Research shows that 67% of these young individuals miss school or after school activities, do not complete their homework and/or are interrupted in their studying careers apart from having to carry out personal hygiene tasks for their relatives which are inappropriate for their age and gender.

Caregiving is a dynamic process. With the long duration of HD the caring tasks develop over time requiring different skills and more and more time devoted to the affected individual, eventually resulting in the carer's life completely revolving around the affected person. Many carers have to leave paid employment as their caring tasks are so time-demanding and burdensome (Siskowski 2006; Boeije et al 2003; Petersen 1991).

Studies of caregivers of family members with HD have found that they face the following common challenges:

- difficulties with the physical and behavioural management of the disabled person;
- chronic worry and stress;
- grief;
- financial difficulties;
- losses in financial opportunities;
- extraordinary time demands;
- difficulties with normal family routines;
- marital discord; and
- absence of formal support services (LoGiudice and Hassett 2005; Cheung and Hocking 2004; Glendenning, 1992; Forster and Young, 1990; Singer and Irvin, 1989).

The additional obligations and sacrifices the caregivers have to fulfill place a major burden on them in terms of psychological, physical and financial resources (Lowit and Van Tuijlingen, 2005; Medeiros et al, 2000; Glendenning, 1992; Forster and Young, 1990; Schultz et al, 1990). Sometimes the caregivers' obligations entail being available all day and all night throughout the week (Boeije et al 2003; Forster and Young, 1990) and it has been found that there is a strong correlation between the number of hours spent on caring and the stress experienced (Taylor et al, 1995; Askham, 1992). This burden and stress can eventually result in a deterioration of their health which leads to a breakdown of family functioning (Elmstahl et al, 1996; Schofield and Bloch, 1998; Anderson, 1988).

HD, as a dementia, poses a unique burden on the family and, in particular, spousal carers as the onset of progressive cognitive impairment during middle adulthood is out of sequence in terms of an individual's life cycle (Aubeeluck and Buchanan 2007; LoGiudice and Hassett 2005). Because it is a genetic disease and the children could be affected, family carers experience distinct and more intense problems than carers of relatives with other dementias as they realise that there is a risk of the disease being transmitted to the children and they are constantly reminded of the reality of the symptoms. The cognitive impairment, aggression, violence, danger, promiscuity and slovenly behaviour of the affected relative cause the caregivers a great deal of stress (Aubeeluck 2005; LoGiudice and Hassett 2005; Williams et al 2000a; Kessler 1993). Many authors (Elmstahl et al, 1996; Schulz et al, 1990; Hayden et al 1980c) report on the frequency of depression and isolation experienced by caregivers as a result of the antisocial behaviour associated with HD which causes social embarrassment to the carer and rejection by their friends. These complex characteristics and symptoms of HD mean that the caring role of affected individuals can be particularly demanding, worrying and challenging, placing a great burden on the primary carer (LoGiudice and Hassett 2005; Lowit and Van Tuijlingen, 2005; Williams et al 2000a). As symptoms typically occur between the ages of 35 to 40 it is at a time of life when they have many family responsibilities such as being a spouse, parent and earning a living to support their families.

It means that the partner who is the carer has to take on these roles and responsibilities in addition to their own (Kessler 1993) and has a significant impact on spousal relationships (Aubeeluck and Buchanan 2007). As HD is a genetic disorder it does not end with the death of the affected individual, but can recur in successive generations with the result that a carer may be responsible for caring for more than one generation of victims at the same time or successively, or the carers may eventually develop the disease themselves (LoGiudice and Hassett 2005; McGarva 2001; Korer and Fitzsimmons 1985).

The greatest problem for carers with children is the fear of their children developing the disease and most of them find it a grueling task to inform their children of their HD risk status. Once the children have been informed of their at-risk status it is frequently not openly discussed again and future caregiving of affected children is not planned for. This avoidance is part of the coping process which is used when individuals are confronted with threats of such a proportion they cannot deal with them directly. Avoidance and secrecy leads to the caregiver becoming isolated and losing contact with potential supportive relationships such as their children, extended family members and friends (Lowit and Van Teijlingen 2005; Krohne 1993).

Lowit and Van Tuijlingen (2005) found that although carers felt that they had a close relationship with their children and that this assisted them emotionally and improved their quality of life, none of the children actively helped with the caring tasks of the affected parent. They also reported that their spouse's parents did not assist in the day-to-day management although they did visit their affected child and that they had no contact with their spouse's extended family or, if they did, it was only on a social basis once or twice a year at a family gathering. These authors also found that many carers avoid contact with other HD families at support meetings as they find that meeting other carers made things worse because they are presented with a picture of their future and they prefer to cope with 'one day at a time". It also served as a reminder of what their children may have to experience later in their lives (Aubeeluck and Buchanan 2007; Lowit and Van Tuijlingen, 2005). However, other studies of long-term progressive diseases found that carers expressed the need to meet and share experiences with other carers experiencing the same difficulties and challenges (Passeti et al 2003; Carers National Association 1992; Culley 1991).

Many carers need information and advice, respite and domiciliary assistance to help them maintain a tolerable quality of life while they perform this valuable social function (LoGiudice and Hassett 2005; Thomson and Thomson, 2001; Taylor et al, 1995). Support is provided to families in Western developed countries in the form of respite services at home, special respite care facilities, day/night respite care services and respite programs based at an institution. However, family care at home has always been the norm in South Africa as there are very few state day care facilities and institutions for the disabled for placement or respite care. They automatically become unpaid workers who save the social service providers' funds (Elmstahl et al, 1996; Petersen, 1991; Forster and Young, 1990; Singer and Irvin, 1989).

2.26 HUMAN ACQUIRED IMMUNODEFICIENCY SYNDROME (HIV/AIDS) IN SOUTH AFRICA

AIDS is "The medical designation for a set of symptoms, opportunistic infections, and laboratory markers indicating that a person is in an advanced stage of HIV infection, with an impaired immune system" (Irwin et al 2003:xv).

A person who is HIV-positive has been infected by the virus, but has not yet developed full-blown AIDS. That is the reason that there are always far greater numbers of HIV-positive people in a population than the number of people with AIDS. It takes an average of 10 years from the time people are infected with HIV until they develop clinical symptoms, although some may develop it much sooner (Irwin et al, 2003). As the immune system becomes weaker opportunistic infections in the environment cause debilitating illnesses and symptoms such as TB or pneumonia during the last two years of life (Nattrass 2002).

HIV is spread through having unprotected sex with an infected person, sharing needles or other drug injection equipment previously used by an infected person, or receiving a transfusion of blood contaminated with HIV. The virus can also be passed from a mother to her infant before or during birth or through breastfeeding (Cadre 2002).

HIV prevalence rates amongst the unemployed are typically 30-50% higher than among the employed (BER 2001) and are higher in the lower job grades compared to higher job grades (Dorrington 2001).

According to the South African Institute of Race Relations, AIDS accounted for 47% of all deaths in the country in 2006 (Nicholson 2007).

CHAPTER THREE

RESEARCH METHODOLOGY

3.1 INTRODUCTION

In this chapter the methodological process is described and issues pertaining to the research conducted are discussed. The reasons for having selected particular methodological choices are provided, potential sources of bias are identified and attempts at minimising these are described in the relevant sections.

The study comprised three components:

3.1.1 Becoming familiar with the families and living environments of the participants under investigation. This was of particular relevance to the majority of participants who came from low-income areas in the WC. Data were gathered by means of socio-political books and the government census (Bank et al 2003; Goldenberg 2003; Statistics SA 2001; Thompson 2001) in order to reflect the environment from where they came and to develop an appropriate and accurate interview schedule to achieve the aims of the study (Pope and Mays, 2000; Chamie, 1989).

3.1.2 Data gathering was by means of in-depth face-to-face interviews according to a semi-structured schedule. Data were tape recorded and the needs of the PT participants, patient group and their carers were explored and analysed.

3.1.3 Recommendations to improve the counselling services of the PT programme and neurogenetic clinic were relayed to the genetic counsellors and staff of the neurogenetic clinic and, where possible, were implemented immediately after all data had been analysed in order to improve the services offered.

3.2 RESEARCH DESIGN

The research was a phenomenological cross-sectional design using a 'multi-method' approach of both qualitative and quantitative methods. Qualitative research is very useful in providing a different aspect on the same phenomenon (Pope and Mays, 2000; Brink, 1991). For example, a great deal of research has been done on HD from the molecular and clinical perspectives, but there is a dearth of international and national literature on the psychosocial aspects of the daily lives of the individuals undergoing PT or patients with HD and their families. One of the main aims of phenomenological research is to understand and describe the multiple realities that portray the richness of real-life events that occur in natural settings from the perspective of the participants (Mason 2002; McMillan and Schumacher 2001; Smith et al 1995; Payton, 1994). As this study included an evaluation of the predictive testing and neurogenetic clinic genetic counselling services from the users' perspectives, a phenomenological design was the most appropriate method of studying this aspect (McMillan and Schumacher 2001).

3.3 STUDY POPULATION AND SETTING

In order to avoid the bias and incorrect inferences which can occur when using only a sample of the population being studied (Sacket, 1979), this research study comprised all the participants who underwent PT; all of the patients attending the neurogenetic clinic for the past three years; and all of the caregivers. This provided variation in demographic and socioeconomic backgrounds which is in keeping with the qualitative convention of broad diverse sampling in order to investigate the aims of the study comprehensively (Minichiello et al 1995; Sarantakos 1993). The three groups of participants comprised:

- all individuals (36) who had undergone the PT programme (PT group) at the Division of Human Genetics, UCT from 20.04.1995 to 19.04.2005;
- all patients (patient group) who had attended GSH Neurogenetic Clinic over the same period (41). This included data collected from 3 individuals who died during this time; and
- all main caregivers involved in the care of their affected relatives (38). Three patients were in the early stages of HD and did not require caregivers.

The study was conducted in the homes of the PT participants and the patients, as the real meaning of their quality of life is experienced in their home setting and is, therefore, best investigated within this social structure frame of reference (Katzenellenbogen et al 1997; Pinder 1993; Brink 1991; Bergner 1989). The home environment also provided more information and insight than is obtained in a formal clinical setting (Smith et al 1995) as it allowed the researcher the opportunity of observing and taking detailed notes of the living environment; relationships and interactions between family members; support or otherwise provided by the family; and critical events that were important and influenced the lives of the participants. Individuals were also most likely to be more comfortable in their home environment when responding to some of the questions which they might have found sensitive by increasing their sense of control over the nature and content of the interview (Giacomini and Cook 2000; Williams et al 1998). For those living further away it meant less stress and anxiety with regards to transport in reaching the clinic for an interview. It allowed contact with those that are disadvantaged and had less education and were least likely to request specific advice, although they were the most in need. It gave the researcher the opportunity of identifying issues that were difficult for them to raise in a hospital setting or might have resulted in their non-attendance. The researcher attempted to avoid the interview taking the form of a medical consultation and to create a situation of trust in which discourse could develop (Rapley 2004; Van Teijingen and Forrest 2004). The home visits thus enabled support for problems quite unrelated to the genetic aspects of HD such as social grants, housing and referral to appropriate health workers for other medical conditions when necessary.

3.3.1 INCLUSION CRITERIA

- PT participants, patients with HD and their caregivers who consented to the interviews; and
- PT participants who had received their results at least 12 months prior to the initial interview of the study. This was to allow the participants sufficient experience of the service to make a reasonable judgement about the quality of its processes (Clarke and Williams 1996) and once the individuals' psychological status would have adjusted and stabilised (Almqvist et al 2003; Tibben et al 1997a; Decruyenaere et al 1996).

3.3.2 EXCLUSION CRITERIA

- patients who were in the late stages and resided in nursing centres; and
- patients whose names appeared on the clinic attendance records, but had died before the study commenced.

3.4 METHODS AND MEASUREMENT INSTRUMENTS

3.4.1 INTERVIEW SCHEDULE (Appendix iii)

A semi-structured interview schedule was developed as the measuring tool to gather data as it was the most appropriate means for obtaining the quantitative information, guiding the discussion topics and ensuring that all the necessary information was included (Pope and Mays, 2000). Closed-ended questions were used to obtain sociodemographic data and for scoring the various measurement scales which included a choice of responses (Veale 1998). These were straightforward to code and analyse. The scaled responses sought a ranking that was nominal for information on age group, level of education, sex or occupation or 'yes', 'no' and 'not applicable' responses. Ordinal responses were required where there was a degree to which a problem affected them, for example, 'no problem', 'mild problem', 'moderate problem', 'severe problem' or 'complete problem'. Open-ended questions were used to encourage free responses without the limitation of preset categories, allowing the participants to express themselves more precisely and in greater depth (Arnold and David 2005; Smith et al 2005). Neutral prompt questions were used to guide the discussion to ensure that the required information was obtained.

The use of both open-ended and closed-ended question allowed increased objectivity and standardisation, but still allowed for probing and clarification (McMillan and Schumacher 2001).

The language used was simple to ensure that it could easily be understood by the respondents. Although parts of the schedule could have been completed as a self-report survey by some participants, the researcher completed them all personally for consistency as the majority of patients and their caregivers were not functionally literate. The interview time provided the researcher with an opportunity of getting to know the participants well and allowed pertinent questions to be asked which gave a deeper insight into their lives within their families and within their communities. That insight would not have been gained by simply processing the data obtained by a self-report method. It also provided the participants with the opportunity of asking questions and being provided with information about HD; PT; the management of their affected relatives; social services and grants to which they were entitled; and other matters of concern to the participants and researcher.

Items that were basic and simple to answer appeared at the beginning of the interview schedule. These included demographic information, knowledge of HD and family information. Questions that might have been sensitive or embarrassing occurred towards the end of the interview (Britten, 2000; Currier, 1990) once the researcher had established a rapport with the participants and they were comfortable about discussing their personal issues.

The interview schedule was constructed to gain information on the following aspects of their lives:

a) Section A: demographic details of both PT and patient groups.
b) Section B: level of knowledge of HD and previous contact with affected relatives of both groups.
c) Sections C, D and E: PT programme, satisfaction with the PT programme and health profile of the PT group.
d) Section F: disability profile of patient group.
e) Section G: social problems experienced by both groups.
f) Section H: social grants of patient group.
g) Section J: access to transport of both groups.
h) Section K: housing situation of both groups.
i) Section M: amount of social support received by the patient group.
j) Section N: satisfaction with GSH Neurogenetic Clinic of patient group.
k) Section R: abuse experienced by both groups.
l) Section U: profile of caregivers.
m) Section W: general needs of families of PT and patient groups.

n) Section X: knowledge of transmission and prevention of HIV/AIDS. The intention was not to obtain information on the participants' HIV/AIDS status, but merely to increase their awareness of the disease and its transmission. It was inappropriate to inquire about their status as the interviews were done in participants' homes and the patient/caregiver/family members were always present not allowing for any privacy.

o) Sections P, S, T were included in the above sections after the pilot study as some of the contents were found to be unnecessarily recursive. The letters I, L, O, Q and V were not included to avoid confusion in the data capturing on the spreadsheets.

3.4.2 Knowledge of HD

Evaluating the level of knowledge of HD was considered to be an important aspect as knowledge gain is one of the aims of genetic counselling and the delivery of information is the most frequent interaction between genetic counsellor and PT participants and patients (Davey et al 2005). Fifteen items were included in this scale on the level of knowledge of HD. These basic aspects of the disease and its genetic pattern of inheritance were covered by the genetic counsellors during the PT programme and by the neurologists and genetic counsellors repeatedly when the patients and their caregivers attended clinic visits. As one of the goals of the PT programme and neurogenetic clinic is to provide participants and patients with genetic and medical information regarding transmission of the disease and inheritance patterns it was important to evaluate the level of recall of information and understanding of risk status as a guide to the effectiveness of the counselling services provided at the two sites.

Satisfaction with the PT programme and neurogenetic clinic genetic counselling services

Satisfaction of service is important either as a single measure of genetic counselling or as an adjunct to information recall and informed decisions. The scale measuring the satisfaction with genetic counselling developed by Shiloh et al (1990) was used with the PT group in order to evaluate the individual processes of the genetic counselling service and neurogenetic clinic rather than a global level of satisfaction only. Evaluation of the individual components enabled the determinants of satisfaction to be determined. The same scale was used with the patient group to evaluate the neurogenetic service with three items altered to make it relevant to the clinic situation. The changes made were to item 6 'the counselling helped you in making your decision about being tested' was substituted for 'privacy when seeing the doctor'; item 11 'the manner in which the test results were presented to you' was changed to 'the manner in which the diagnosis was presented to you'; and item 16 'the extent to which team members met your expectations' substituted for 'decisions are made with you with full explanations provided to you'. The scale was developed to cover the three health care aspects considered relevant to satisfaction (Ben-Sira 1976). These included, firstly, the extent to which the participants/patients evaluate that the staff has the required skills, and gives the required treatment and reassurance; secondly it evaluates the staff's behaviour towards the PT participant/patient as a person rather than a 'case'; and thirdly the satisfaction with the administrative processes. There was one item evaluating the overall feeling of satisfaction with the clinic (Shiloh et al 1990). The multidimensionality of this satisfaction scale is very useful since global levels of satisfaction are not beneficial in identifying the strengths and weaknesses of a programme and as specific aspects of counsellor performance should be evaluated as participants might experience more satisfaction with certain aspects compared to others (Clarke et al 1996).

3.4.4 The Stanford Health Assessment Questionnaire Scale (HAQ)

The HAQ was used to measure the level of difficulty in performing activities of daily living. The researcher selected this scale as it had been widely used in clinical and research studies involving disability (Milligan et al, 1993; Wolfe et al 1988; Fries et al, 1980) matching well with disabilities of the patients with HD. Its thoroughness and results were rated the highest of 16 most frequently used activities of daily living scales (McDowell and Newell, 1996; Liang et al, 1990). It is broad in scope and can be completed in five to eight minutes as either a self-report or researcher-administered scale. It is a good descriptive instrument and although it is not suitable for measuring clinical change in outcome studies, that was not the intention of this study. The aim was to establish what activities of daily living the participants had difficulty in performing. The Unified Huntington's Disease Rating Scale was not selected as it did not provide for a range of ability. For example, there was a 'Yes' or 'No' response required for "Could the subject walk without falling?". Some individuals could walk around the house independently where the distances were limited and there were plenty of visual cues provided by the environment, but the same person was not able to walk outside where the terrain was rough, there were moving objects such as cars which cause distraction and the distances were much greater (Huntington Study Group, 1996).

The disability profile was positioned early in the interview schedule as it provided the researcher with a greater understanding of the patients' disabilities and allowed the questioning to be more appropriate and in-depth to gain a greater insight into their lives and problems experienced.

3.4.5 The Hospital Anxiety and Depression Scale (HAD)

As depression is such a significant feature in HD the HAD was used to measure the levels of anxiety and depression the patients experienced over the past week (Zigmond and Snaith 1983). The scale focuses on generalised anxiety and loss of pleasure response. A score of 11 or higher indicates a probable presence of the mood disorder; a score of 8 to 10 being suggestive of the presence of the respective state; and 7 or less for non-cases. It was selected as a measuring scale as it takes only 2 to 5 minutes to complete and it allows for the items and possible responses to be read to the participants.

3.4.6 Level of social problems

The scale on the level of social problems experienced in the lives of the participants was developed by focus groups of people with permanent disabilities living in low-income suburbs in Cape Town (Futter 2004). These suburbs included or were in close proximity to the residential areas of the study population. A later study conducted in Lotus River confirmed the frequencies and extent of these social problems (Futter 2004). There appears to be no universally acceptable definition of quality of life in medicine and there is no distinction between quality of life measures and methods (McDowell and Newell, 1996; Jette, 1994). Thus some of them are clinical scales, some involve measures of primary care and others are indicators of well-being (Bergner 1989; Kottke 1982; Jones 1977). In the social sciences, however, quality of life refers to the adequacy of people's material circumstances, and to their feelings regarding those circumstances (McDowell and Newell, 1996; Brink 1991) rather than in general categories of mobility, self-care or function as measured in the EuroQuol Quality of Life Scale or the Quality of Well-Being Scale (EuroQuol Group, 1990; Kaplan et al, 1989). Therefore the scale on the level of social problems was developed as an indicator of the quality of life specific to people with chronic disabilities living in the immediate environment and sharing similar histories and political influences.

3.4.7 **Social support profile** (MOS)

Social health was measured by means of The Medical Outcomes Study Social Support Survey scale (MOS). It was developed as a brief self-administered indicator of social support and was intended for use in general surveys in chronically ill patients matching the study population who were permanently disabled. It is quick to complete and demonstrates that functional support is distinct from the structural aspects of support, a distinction similar to the availability and adequacy of support (McDowell and Newell 1996).

3.4.8 **Caregiver Burden Scale** (CBS)

The CBS was selected as the tool for measuring caregiver burden as it was developed for caregivers who had been caring for chronically disabled relatives who had both physical and cognitive disorders, whereas other scales focus on dementia, Alzheimer's Disease and elderly people only (Elmstahl et al, 1996; Grafstrom et al, 1992; Vitaliano et al, 1991). It also matched the study population to a greater extent than other scales as it was developed for caregivers of people who had suffered from strokes for at least three years and the shortest time of onset of symptoms of the patients in the study population was two years. At the time the research was conducted the Huntington disease quality-of-life battery for carers (HDQoL-C), which would have been the scale of preference, had not yet been tested for reliability and validity (Aubeeluck and Buchanan 2007).

3.4.9 Audio recordings

Audio recordings were made of each interview so that the researcher could engage with the participants and patients without having to interrupt the flow of conversation to make notes. Audio-taping allows a more complete objective record than hand-written notes taken during the interview and it enables the researcher to concentrate on how the interview is proceeding (McMillan and Schumacher 2001; Smith et al 1995). The amount of data were also too large to capture by taking notes on the discussions that took place. The audio recordings were transcribed by an official transcriber employed by the university who was fluent in both English and Afrikaans. She merely captured the recorded discussions and did not make any attempt to interpret or translate them.

3.4.10 INTERVIEWS

PT participants

Interviews were conducted in the homes or venue of choice of the participants. The researcher explained that she was independent of the PT programme, explained the aims of the study and how important their contribution would be to obtain feedback about their perceptions and experiences of the programme at the time they underwent testing, and to be informed of any recommendations to improve the programme to benefit individuals undergoing testing in the future. Each interview took approximately two hours and was conducted in English or Afrikaans depending on the home language of the participants as the researcher was fluent in both languages. Participants were alerted to the fact that they might find the content of the discussion stressful as it might evoke strong emotions about themselves and their families and that a second follow-up interview would be arranged at their convenience to discuss these aspects and to refer them to the relevant counsellors, if necessary. The researcher also monitored the effect of the interview on the participants by observing non-verbal behaviour and the way in which participants replied to the questions (Smith et al 1995). The interview aimed to be like a guided conversation with a purpose and, although the interview schedule was semi-structured the order in which the content was covered differed with each participant to enable certain topics to be pursued in greater depth and to allow a free flow of conversation (Van Teijingen and Forrest 2004; Rapley 2004; McMillan and Schumacher 2001).

Patients and carers

In situations where the patients were unable to respond accurately due to cognitive problems or dysarthria, a proxy was used in the presence of the patient. Each interview took approximately two hours for the patients and one hour for the carer and was conducted in the language of choice of the patients and their carers. In the majority of cases this entailed a second home visit to complete. The researcher attempted to create an informal and friendly atmosphere so that the participants could feel at ease. She introduced herself to the participants as a physiotherapist and they appeared to perceive her role to be that of a 'helping' nature which made interaction easy. Once a rapport had been established they appeared to be comfortable with discussing the more personal aspects of the schedule and provided far more in-depth discussion in these areas than what was required of them. Comprehensive field notes of the home circumstances, interpersonal relationships and interactions of the family and any critical events that occurred and were an important influence on the lives of the participants were taken during the interviews and valuable data regarding the physical and social environment were gathered during this time. Although reliability can be a problem when using observation on its own as a method, it can be an invaluable approach in conjunction with other data collection techniques as events can be observed in the way in which they normally occur (Pope and Mays, 2000; Hicks, 1995). The impressions and feelings gained in this way allowed a far greater understanding of the patients' and carers' lives and environment than would have been obtained by a self-report survey.

Follow-up interviews

These were provided for PT participants if they wished to discuss emotional aspects that had been evoked by the first interview or wished to discuss other aspects of HD regarding their family members and their care.

Follow-up home visits were conducted to the patients and their carers to complete the interview schedule, if found necessary, or to confirm that their applications for social grants or further medical appointments had been processed. Further non-pharmacological management was also provided when the carers discussed aspects of caring that were problematic or needed reassurance about the care they were providing.

Obsequious bias was minimised by asking all questions in a neutral manner and probing the individual's responses when the information did not correspond with previous responses. A probe is a neutral request to clarify an ambiguous answer or to complete an incomplete answer (Neuman 1999). This also minimised the possibility of under- or over-reporting (Sacket 1979).

Rumination bias could have been introduced throughout the interview as most of the participants were confronted with questions to which they had previously not given much consideration.

3.5 TRUSTWORTHINESS/VALIDITY

In quantitative research "A valid measure is one which measures what it is intended to measure" (Grinnell, 1985: 161). Part of the reason for selecting the various scales used in the HD interview schedule was that they had already been tested for validity and reliability. The HAQ has been shown to have good validity and reliability correlations with other scores in several international studies (Hakala et al, 1994; Ramey et al, 1992; Brown et al, 1984; Fries et al 1982; Fries et al, 1980). The HAD scale has been found to perform well assessing severity and 'caseness' of anxiety disorders and depression in both somatic and psychiatric cases in both primary care patients and the general population (Bjelland et al 2002; Hermann 1997). The criterion validity coefficients, test-retest and item-scale correlations with loneliness, marital and family functioning and mental health had been demonstrated in several studies of the MOS (McDowell and Newell, 1996; Sherbourne and Stewart, 1991). The CBS has been shown to have good construct validity and test-retest stability in other studies (Elmstahl et al, 1996).

In qualitative research trustworthiness is used to measure validity. Research is considered to be trustworthy when it accurately reflects the thoughts and reality of the participants (Holloway 1997). According to Lincoln and Guba (1985) this involves the following elements: credibility, transferability, dependability and confirmability:

- Credibility is established by ensuring that the research participants are identified and described accurately (Holloway and Wheeler 1996). In this study peer debriefing was used to improve the credibility of the study by having an independent social worker regularly check the data interpretation and analysis (Minichiello et al 1995; Sarantakos 1993).
- Transferability means that the findings in one context can be transferred to participants or situations that are similar (Holloway 1997). The researcher has reservations about the findings of this interview schedule being generalised to other communities as they will have different historical backgrounds, socioeconomic backgrounds, geographical area, housing and transport situations and it may not necessarily “fit” other communities.
- For a study to be dependable it must be consistent and accurate. This was demonstrated by providing detailed descriptions of the methodology so that the readers can follow the path of the results and by having the data interpretation and analysis critically evaluated by an independent social worker familiar with the communities in which the study was conducted (Holloway 1997).
- The last element of trustworthiness is confirmability. Confirmability means that the findings of the research are due to the results and not biases and subjectivity of the researcher (Holloway 1997). According to Guba and Lincoln (1989) the data should be linked to their sources

so that the reader can establish that the conclusions and interpretations arise directly from them. This was obtained by the follow-up visits and interviews to confirm that the participants agreed with the transcription and the researcher's interpretation of certain aspects of the conversation that might have been meant differently. These visits provided the opportunity for any corrections and alterations to be made, increasing the accuracy of the interview (McMillan and Schumacher 2001).

Qualitative research acknowledges that objective reality and subjective experiences possibly occur together in the research data (Holloway 1997). Trustworthiness is very dependent on the skill and competence of the evaluator because the evaluator is the instrument (Patton, 1987) and thus the researcher has to ensure throughout the research that the process ensures that the results are trustworthy (Krueger and Casey 2000).

"Content validity refers to comprehensiveness, or to how adequately the sampling of questions reflects the aims of the index that were specified in the conceptual definition of its scope" (McDowell and Newell, 1996: 30). This was achieved with the HD interview schedule by having an impartial social worker who was familiar with qualitative methods and two experts on HD critically review the content of the schedule before the pilot study was conducted. This is a common procedure to establish content validity in qualitative research (Aubeeluck and Buchanan 2007; McDowell and Newell, 1996).

3.6 RELIABILITY

"A reliable measurement obtains the same result on repeated occasions and by independent individual's administering the instrument" (Grinnell, 1985: 171). Reliability is concerned with error in measurement and requires consistency or stability of the measurement process across time, patients, or observers (Feinstein, 1987). In other word the measurement has to be stable, repeatable and dependable.

Reliability of the interview schedule was established by the researcher and an independent genetic nursing sister each interviewing six individuals on the various closed-ended scales. Spearman's correlation coefficient revealed a correlation of 0.99. There was also agreement on the classification of most of the responses to the open-ended questions demonstrating inter-rater reliability of the interview schedule and that the data collection was conducted in a reliable manner (Hoffman, 2001; McMillan and Schumacher 2001; Katzenellenbogen et al, 1997).

The HAQ has been shown to have inter-rater reliability of 0.85 and test-retest reliability after six months of 0.98 (Ramey et al, 1992; Fries et al, 1980). The reliability for the MOS has been found to be 0.9 (internal consistency) and 0.78 for test-retest after one year (McDowell and Newell, 1996; Sherbourne and Stewart, 1991). The CBS has been found to have internal consistency of 0.70 and test-retest reliability of 0.98 (Elmstahl et al, 1996). The Satisfaction with Genetic Counselling Scale by Shiloh et al (1990) was found to have good reliability (Cronbach alpha = 0.78) with each of the 3 dimensions: instrumental reliability 0.68; affective reliability 0.59; and procedural reliability 0.59.

3.7 PROCEDURE

3.7.1 PILOT STUDY

After the validity of the HD interview schedule had been established a pilot study was conducted with two people with other permanent physical disabilities living in one of the study suburbs to avoid including the study population. This was done to ensure questions were easily understood and unambiguous providing a measure of face validity (Aubeeluck and Buchanan 2007). Small alterations were made which removed sections P, S and T and included them in the remaining sections as some of the questions were found to be unnecessarily repetitive.

At this stage it became clear that it was not necessary to translate the interview schedule into Afrikaans as the researcher was fluent in English and Afrikaans and capable of translating the items should the participants have any difficulty in understanding them.

Reliability tests were performed on the interview schedule (Chapter Three: paragraph 3.6).

Individuals from the PT programme were not included in the pilot test as the sample size was so limited it would have exposed them to the questions and may have altered their responses for the actual study interview, introducing a 'pretesting' error as can occur in pretest sensitising in quantitative research designs (McMillan and Schumacher 2001).

3.7.2 RECRUITMENT OF STUDY POPULATION

PT programme

Because of the confidential nature of PT the researcher was unaware of the individuals who had undergone testing or their test results. The genetic nursing sisters who administered the programme contacted the participants telephonically and informed them of the research the Division wished to conduct (Appendix iv). They were invited to participate and informed of the researcher's previous occupation and prior lack of contact with the Division and its activities, and the envisaged time an interview would require. If the individuals agreed the researcher was provided with their contact details and she telephoned them to arrange a suitable time and venue of their choice for the interview. If they did not wish to participate the genetic nursing sister would ask them to provide the reasons for not participating.

Patients from the Neurogenetic Clinic

The majority of patients attending the neurogenetic clinic did not have telephones at home and many of their addresses in the hospital file had not been updated. The researcher attended the monthly clinic and spoke to the patients and their caregivers in the waiting room before they were seen by the neurologist. They were informed of the research aims and that it involved a home visit by the researcher and an interview of approximately two hours. If they agreed the researcher updated their contact details and arranged a time which suited them for a visit.

Patients who did not have clinic appointments for six months and had no telephone were visited by the researcher on an ad hoc basis and arrangements were made for another visit at a time that suited them.

Carers

The caregiver, who was often the proxy as well as the carer, was interviewed after the patient interview. If more time was required, a further meeting was arranged for her/his interview.

3.7.3 Interviews

PT programme

The researcher informed the participants of the format that the interview would take and that they were free to interrupt discussions to ask questions or make statements. In order to ensure confidentiality between family members the researcher made it clear that there would be no discussion about family members other than their family pedigree and their own children's knowledge about their at-risk status and the availability of PT. The consent forms were given to the participants to read, explanations were provided for further understanding and their signatures were obtained. They were also informed that the interview might evoke disturbing emotions and that the researcher would arrange an additional interview or referral to a counsellor at the time of the follow-up interview if they so wished. At the second interview the participants were able to read the transcript of the first interview to ensure that the information that was captured was correct and what they had intended to portray. If necessary, alterations and additions to the first discussion were made then and a further discussion took place regarding their emotional state. They were reassured that if they ever needed more information or support with regards to their affected family members they could contact the genetic nursing sisters at any time in the future for appointments with the appropriate health workers. Their contact numbers were provided.

Patients/caregivers

A similar description of the intended interview as was given to the PT participants was provided to the patients and their caregivers before their written consent was obtained. After the interviews were completed and the researcher had become familiar with their social grant deficits and non-pharmacological problems with managing the symptoms of HD, they were advised about applying for their entitled social grants and assisted with feeding, dressing, walking problems and other difficulties such as housing and Dial-a-ride applications. Further home visits to patients and caregivers were conducted to confirm their responses to the questions at the first interview and to investigate whether recommendations for social grants had been processed and whether the patients and caregivers were finding the suggestions for the management of their different problems helpful and how they were implementing them.

After three interviews with the PT group and four with the patient group had been completed it became obvious that there were general needs of families with HD that the participants had discussed as essential means of support and which they were not receiving. Section W was thus added to the end of the interview schedule as closed-ended questions and was completed by the remaining participants. Those who had already completed the first interview completed this section at the follow-up sessions.

Once the questions regarding HIV/AIDS in the interview schedule had been completed the researcher was able to explain the magnitude of the problem in South Africa and to discuss any aspects of concern or interest which they themselves raised. They were provided with various appropriate leaflets on HIV/AIDS provided by the Department of Social Services and Poverty Alleviation (DSSPA) for education of the general public (Appendix v provides an example). They were also given the telephone numbers of the AIDS Helpline and advice on the nearest Community Health Centre where they could have a free HIV blood test and receive a free supply of condoms .

3.8 DATA MANAGEMENT

Quantitative data gathered by means of the interview were captured on Excel spreadsheets. They consisted of the demographic data, Knowledge of HD, Satisfaction with the PT Programme, HAQ, HAD, Social Problems, MOS, Satisfaction with the Neurogenetic Clinic and CBS scales of measurement.

The analysis of the quantitative data was conducted in two main steps. As the measurement scales use ratings from 1 to 5 the scale is ordinal and it would be too strong an assumption to attribute higher numerical properties to the responses and treat them, initially, at interval or ratio level of measurement. Therefore, in the first step each construct was analysed by examining the mutual relationships between the items within the set. This was done by various interdependence and scaling techniques such as Categorical Principal Component Analysis (CAT-PCA)[1] and Cronbach's Alpha test. CAT-PCA belongs to the group of nonlinear multivariate (or

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[1] CAT-PCA belongs to the group of non-linear multivariate (or optimal scaling) techniques. It can be used as an alternative to log-linear models. These scaling techniques have the advantage of remaining stable when sample sizes are small or large avoiding the difficulty of interpreting the large number of parameter estimates required by log-linear models. CAT-PCA performs non-linear analyses on a set of items that may have different levels of measurement to reveal limited, but major dimensions of variation. Thus, the original set is replaced by a smaller one with as little loss of information as possible. CAT-PCA was developed by the Data Theory Scaling System Group (DTSS), Psychology Department, Faculty of Social and Behavioural Sciences, Leiden University, The Netherlands. It is part of the procedures option in SPSS, a software package for data management and statistical analysis.

optimal scaling) techniques. CAT-PCA, in contrast to classical PCA, can perform nonlinear analysis on variables with different measurement levels: nominal, ordinal, interval or ratio. CAT-PCA projects the items, which were treated as either or both single nominal or ordinal levels, as vectors into a two or more dimensional space (SPSSA, 2001). The length of the vector represents its relative contribution to a dimension. Dimensions are characteristics which are grouped together by means of clusters of responses. The angle between the vectors represents the level of association between the vectors. Although the variables are quantified in nonlinear principal components the technique has a strong visual ability to identify possible relationships among variables and objectscores (respondents) in a multidimensional space. When the set of items appeared one-dimensional, Cronbach's Alpha test was used as a reliability coefficient to express the internal consistency between the items.

In the second step, where required, items were rescaled by transformations invariant to the level of measurement of the items if this level was not violated. Thus all subsets were transformed into one or a few scales depending on the underlying dimensions[2] of the constructs. Eigenvalues[3] are given to indicate how successful the maximalisation and minimalisation criteria were. The component loadings[4] and mutual relationships of these scales were subsequently analysed by appropriate multivariate techniques.

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[2] Dimension: a reduced number of variables which represents the information of a large number of variables without the loss of information. The original variables are weighted such that they correlate maximally with one dimension, but minimally with another thus creating dimensions that are not correlated. Therefore, conceptually, a dimension is labelled by the common characteristic of the variables that determined the particular dimension.

[3] Eigenvalue: "a special set of scalars associated with a linear system of equations that are sometimes also known as characteristic roots" (Marcus and Minc, 1988:144). It indicates how successful the maximisation and minimalisation criteria were.

[4] Component loadings: are the factor loadings in a factor analysis. They are weights contributing to and can be considered as the correlation with the dimension. Thus a high component loading is important in the definition of a dimension and variables with relative high component loadings on a dimension are strongly related.

Open-ended questions were analysed by means of a process of constant comparative analysis. This is a technique in which the researcher simultaneously collects information through interviews, reads and re-reads the transcriptions as individual cases, dissembles interviews through coding, rearranging coding categories into patterns, and reintegrating the patterned categories into a conceptualisation that encompasses the experience of all participants while identifying the deviations and exceptions to these trends. It thus involves analysing the content of the actual words used by the participants and patients and not the researcher's preconceived hypotheses (Larsson et al 2006; Van Riper 2005; Roberts and Sarangi 2002; Smith et al 1995). Simple frequency tables were used to categorise the responses into descriptive statistics such as percentages, means and modes to provide a summary of the categories of some aspects of the interviews (Pope et al 2000).

3.9 ASSUMPTIONS

This research was based on the assumption that

- the participants would respond to questions and provide unsolicited information both accurately and honestly; and
- at the end of the research fieldwork period of eighteen months, the total population of patients and caregivers had been reached as there were no further new referrals to the neurogenetic clinic. There were three family members who the researcher met during the home visits who appeared to have extrapyramidal symptoms, but as they had not yet attended the clinic for diagnosis and did not mention their obvious movement and balance difficulties, they were not included in the study group. Their families were, however, provided with telephone numbers should they need to contact the clinic in the future.

3.10 ETHICAL CONSIDERATIONS

3.10.1 The study was granted approval without reservations by the Medical Research Ethics Committee of the University of Cape Town (Rec Ref 362/2003) (Appendix vi).

3.10.2 PT programme individuals were contacted by the genetic nursing sisters inviting them to participate in the research and only then with their consent were their names and contact details provided to the researcher. No form of persuasion was used to encourage the individuals to participate and they were assured that if they did not wish to participate it would not have any influence on their or the family members' future medical management at either the neurogenetic clinic or genetic clinic.

3.10.3 Audio-taped recordings were transcribed immediately after the interviews and these records were kept in a locked filing cabinet in the Division of Human Genetics and were destroyed once the study was written up. The participants received numerical codes and their names did not appear on the interview schedules, audio tapes, transcriptions or spreadsheet.

3.10.4 The risk to the participants in this study was the discussion of sensitive information and stressful experiences. The researcher ensured that confidentiality and anonymity was maintained. No discussions took place with the clinic staff or genetic counselling staff before or after the interviews. The researcher was sensitive to the emotional state of the participants throughout the interviews. Participants had the opportunity of a second interview when any emotional issues evoked during the initial interview were dealt with or any questions that the participant had were answered.

3.10.5 The long-term benefit of this study will be to use the information to, if necessary, improve the genetic counselling process to support the individuals and their families undergoing the PT programme; and to improve the clinical counselling service at GSH neurogenetic clinic.

3.10.6 Consent forms (Appendix vii) were signed prior to the interviews for consent to the interview, audio recordings to be taped and photographs to be taken. If the cognitive level of the patients precluded informed consent, it was provided by their carers in their presence.

3.10.7 Feedback of the results was provided to the genetic counselling team after the data had been analysed and to the participants of the PT programme on completion of the research write-up.

3.11 WEAKNESSES OF THE STUDY

- The paucity of information, literature and research data concerning living conditions of individuals with HD and their families in disadvantaged communities. Where relevant, data from international studies were used. However, caution has to be exercised in generalising from the communities of one country to another and from one national region to another.
- Researcher bias with the structured questionnaire had to be recognised. The subjective responses of the participants might have been what they thought was appropriate, rather than their true circumstances. However, subtle signs and intimations were followed up by further in-depth questioning by the researcher which might have, to a certain extent, overcome this bias.
- Data were only gathered at one point in time. This is a limitation of descriptive studies as changes in the population cannot be measured (Taylor 2005a; Neuman 1999). However, major changes in population characteristics of this cohort, for example, education level, income and housing were unlikely to have changed; and only minor changes in the

nature and extent of the problems associated with HD were likely to have occurred over the research period. For this reason it was thus also considered to be unnecessary to repeat measurements over time.

- The findings are not statistically or conceptually generalisable because of the small sample limited to one geographical area (Larsson et al 2006; Taylor 2005a). Because of the diverse nature of the study, had the number of participants been larger it may have been possible to detect other similarities and differences across outcome measures.
- Data were based on self-reports and might not have been accurate or honest.
- PT participants who reported levels of depression severe enough to warrant medication or counselling by their medical practitioners had not had the depression diagnosed on any structured psychiatric interview or conventional depression scales and, therefore, it might be considered a subjective bias.
- As the research design was ex-post facto some of the participants who had been symptomatic for a number of years had difficulty in recalling facts concerning their disease.

3.12 STRENGTHS OF THE STUDY

- The researcher had not worked in the Division of Human Genetics prior to the study and, therefore, did not know the participants involved or their family histories. This enabled the evaluation to be more objective as she had no vested interest or private agenda for personal gains in the outcome of the appraisal and the participants felt free to criticise certain aspects without thinking that they were jeopardising their or their families' future management through the Division or clinic. It also allowed the participants the opportunity of discussing negative aspects of the programme and clinic more freely than with people they were accustomed to seeing at these clinics.

CHAPTER FOUR

ANALYSIS, FINDINGS AND DISCUSSION

4.1 INTRODUCTION

The findings of the research are reported in this chapter. Mutual relationships between the items within each section are presented in tables or graphs followed by a discussion. Because this research is concerned with the evaluation of a programme, where possible, reference is made to literature showing similarities and differences to other HD research findings. It was also essential for the researcher to be familiar with the population in general and the environments in which the majority of the patients lived in order to comprehend the psychosocial problems with which they were confronted, therefore, population statistics are included to enhance the context of the data.

In view of the vast amount of data that were collected the findings have been presented separately.

- data gathered from the individuals who underwent PT (PT group) are presented in section 4.3 (page 145). The results of the predictive testing programme evaluation are presented in two parts. Part 1 (page 172) deals with the analysis of relationships between and within the demographic variables and satisfaction scale items. Part 2 (page 182) provides an evaluation of the actual level of satisfaction with the predictive testing programme.

- data from the patients (patient group) are presented in section 4.10 (page 192) and that of the caregivers in section 4.14 (page 248). The headings have been presented in the same format as the Interview Schedule (Appendix iii) and should be read in conjunction with the than that of Australia, Canada, USA and France where the uptake rate has been between 5% to 20% (Trembath et al 2006; Harper et al 2004; Taylor 2004; Creighton et al 2003; Evers-Kiebooms et al 2002; Meiser and Dunn 2000). There appears to be a wide range of uptake at different centres within one country (Creighton et al 2003). One of the reasons for this might be in the nature of genetic counselling provided, where one centre might be neutral and another more positive to the outcomes of PT. There was no decline in the rate of requests for testing during the period under review in the WC cohort, but subsequent to this study there has been a decline in the requests as has been found in the Canadian population (Creighton et al 2003). Another reason for the low uptake might be the low level of formal education of the general population of families with HD in the WC. This is confirmed by Soliz-Perez et al (2001) who found that the uptake rate in their cohort was determined by the access to information of the population at-risk. The drop-out rate of individuals who had attended at least one counselling session, but had not completed the programme, was 8.3% (3/36). This is low compared to international figures of a drop-out rate of up to 40% after the first visit (Bradley et al 2004; Myers 2004; Hayden 2003), but similar to that found in Australia, namely, 14.3% (Trembath et al 2006). The 3 drop-out individuals of this cohort were interviewed to investigate the reasons for them dropping out to ensure that they were not of special concern to the quality of the genetic counselling process. The reason given by 1 individual was that she did not want to know the test result in case it compromised her plans to emigrate. The second individual stated,

"The programme is too long and I don't understand the purpose for me to have to wait that long and go through the whole programme when I come from a family with many affected relatives and I am fully aware of the facts of the disease".

His opinion of the length of the protocol is confirmed by Van t'Spijker and Ten Kroode (1997) as one of the reasons for people not undergoing the PT programme. The third individual obtained employment overseas and was unable to complete the required 4 sessions of the pretest protocol in the prescribed time framework.

Patients from the Neurogenetic Clinic

The majority of patients attending the neurogenetic clinic did not have telephones at home and many of their addresses in the hospital file had not been updated. The researcher attended the monthly clinic and spoke to the patients and their caregivers in the waiting room before they were seen by the neurologist. They were informed of the research aims and that it involved a home visit by the researcher and an interview of approximately two hours. If they agreed the researcher updated their contact details and arranged a time which suited them for a visit.

Patients who did not have clinic appointments for six months and had no telephone were visited by the researcher on an ad hoc basis and arrangements were made for another visit at a time that suited them.

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The caregiver, who was often the proxy as well as the carer, was interviewed after the patient interview. If more time was required, a further meeting was arranged for her/his interview.

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- Data were only gathered at one point in time. This is a limitation of descriptive studies as changes in the population cannot be measured (Taylor 2005a; Neuman 1999). However, major changes in population characteristics of this cohort, for example, education level, income and housing were unlikely to have changed; and only minor changes in the

nature and extent of the problems associated with HD were likely to have occurred over the research period. For this reason it was thus also considered to be unnecessary to repeat measurements over time.

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- The researcher had not worked in the Division of Human Genetics prior to the study and, therefore, did not know the participants involved or their family histories. This enabled the evaluation to be more objective as she had no vested interest or private agenda for personal gains in the outcome of the appraisal and the participants felt free to criticise certain aspects without thinking that they were jeopardising their or their families' future management through the Division or clinic. It also allowed the participants the opportunity of discussing negative aspects of the programme and clinic more freely than with people they were accustomed to seeing at these clinics.

CHAPTER FOUR

ANALYSIS, FINDINGS AND DISCUSSION

4.1 INTRODUCTION

The findings of the research are reported in this chapter. Mutual relationships between the items within each section are presented in tables or graphs followed by a discussion. Because this research is concerned with the evaluation of a programme, where possible, reference is made to literature showing similarities and differences to other HD research findings. It was also essential for the researcher to be familiar with the population in general and the environments in which the majority of the patients lived in order to comprehend the psychosocial problems with which they were confronted, therefore, population statistics are included to enhance the context of the data.

In view of the vast amount of data that were collected the findings have been presented separately.

- data gathered from the individuals who underwent PT (PT group) are presented in section 4.3 (page 145). The results of the predictive testing programme evaluation are presented in two parts. Part 1 (page 172) deals with the analysis of relationships between and within the demographic variables and satisfaction scale items. Part 2 (page 182) provides an evaluation of the actual level of satisfaction with the predictive testing programme.

- data from the patients (patient group) are presented in section 4.10 (page 192) and that of the caregivers in section 4.14 (page 248). The headings have been presented in the same format as the Interview Schedule (Appendix iii) and should be read in conjunction with the than that of Australia, Canada, USA and France where the uptake rate has been between 5% to 20% (Trembath et al 2006; Harper et al 2004; Taylor 2004; Creighton et al 2003; Evers-Kiebooms et al 2002; Meiser and Dunn 2000). There appears to be a wide range of uptake at different centres within one country (Creighton et al 2003). One of the reasons for this might be in the nature of genetic counselling provided, where one centre might be neutral and another more positive to the outcomes of PT. There was no decline in the rate of requests for testing during the period under review in the WC cohort, but subsequent to this study there has been a decline in the requests as has been found in the Canadian population (Creighton et al 2003). Another reason for the low uptake might be the low level of formal education of the general population of families with HD in the WC. This is confirmed by Soliz-Perez et al (2001) who found that the uptake rate in their cohort was determined by the access to information of the population at-risk. The drop-out rate of individuals who had attended at least one counselling session, but had not completed the programme, was 8.3% (3/36). This is low compared to international figures of a drop-out rate of up to 40% after the first visit (Bradley et al 2004; Myers 2004; Hayden 2003), but similar to that found in Australia, namely, 14.3% (Trembath et al 2006). The 3 drop-out individuals of this cohort were interviewed to investigate the reasons for them dropping out to ensure that they were not of special concern to the quality of the genetic counselling process. The reason given by 1 individual was that she did not want to know the test result in case it compromised her plans to emigrate. The second individual stated,

> *"The programme is too long and I don't understand the purpose for me to have to wait that long and go through the whole programme when I come from a family with many affected relatives and I am fully aware of the facts of the disease".*

His opinion of the length of the protocol is confirmed by Van t'Spijker and Ten Kroode (1997) as one of the reasons for people not undergoing the PT programme. The third individual obtained employment overseas and was unable to complete the required 4 sessions of the pretest protocol in the prescribed time framework.

4.3 SOCIODEMOGRAPHIC DATA

Age, gender, ethnicity and first home language

Age	Gender	Ethnicity		Total
		White	Mixed	
20 - <30	Male	1	0	1
30 - <40	Male	4	0	4
	Female	6	3	9
40 - <50	Male	3	0	3
	Female	3	2	5
50 - <60	Male	1	1	2
	Female	2	0	2
>59	Male	1	0	1
	Total	**21**	**6**	**27**

Table 1: Participants per age group, gender and ethnicity (mean age 41.1 years)

The study cohort comprised 59.2% (16/27) females and 40.7% (11/27) males (Table 1) (Figure 3). The predominance of females presenting for PT is similar to that of other studies (Bombard et al 2007; Trembath et al 2006; Velakoulis et al 2005; Shannon 2004; Creighton et al 2003, Hayden 2003; Goizet et al 2002; Mueller and Young 2001; Harper et al 2000). Six (5 females, 1 male) (22.2%) were Mixed Ancestry and 21 (11 females, 10 males) (78%) were White according to self-perception and self-classification and not based on a legal definition. The population in the WC comprises 61.1% Mixed Ancestry and 19.4% White, clearly showing that their were more individuals from the minority White group requesting PT compared to the majority Mixed Ancestry group. The home language of 66.6% (18/27) was English and 33.3% (9/27) Afrikaans; whereas the first home language of the general population in the WC is 26% English and 68.2% Afrikaans (Statistics SA 2001).

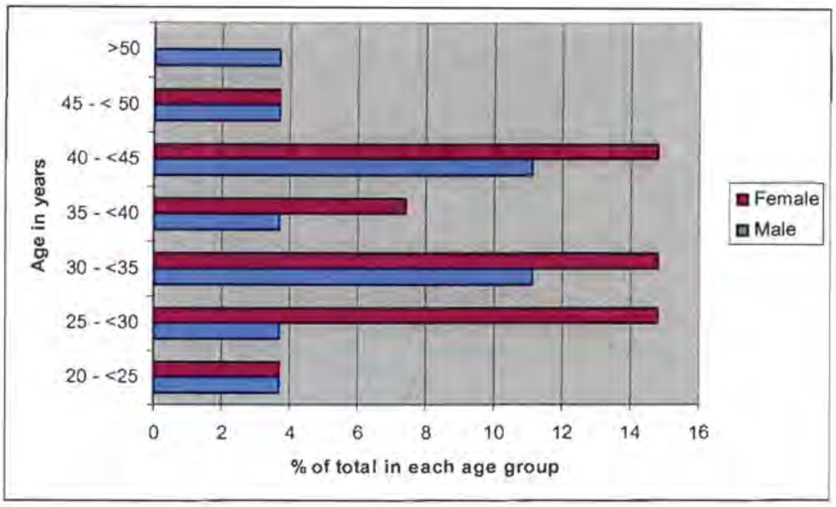

Figure 3: Age distribution of males (n=11) and females (n=16) at the time of the first visit to the HD PT programme

Occupation

Category	Participants	Western Cape population
Managerial, professional, semi-professional	77.8% (21/27)	22%
Clerical, sales, service	18.5% (5/27)	22%
Skilled agricultural, craft, operators	3.7% (1/27)	7%
Elementary occupations	0% (0/27)	49%

Table 2: Categories of employment (n=27)

One hundred percent of the participants had been in full-time permanent employment up to recently, but 11.1% (3/27) had become symptomatic and could no longer work. In the WC the overall employment rate is 48.5%. A comparison of the categories in which the participants and general population were employed in 2001 is provided in Table 2 where it can be seen that 77.8% (21/27) of the participants were employed in managerial or professional positions, as defined by the census (Statistics SA 2001).

Level of Income

No. of Participants	Monthly income R	Total R
3	1 200	3 600
1	2 400	2 400
4	4 800	19 200
13	19 200	249 600
4	38 400	153 600
2	75 000	150 000
	Total	578 400
n=27	**Average**	**21 422**

Table 3: Level of income of PT participants or, in the case of single individuals, per individual

According to Statistics SA (2001) the mean annual income of working adults aged 15 - 65 in the WC is R18, 703 (R1 559 per month); Whites R64 968 (R5 414 per month) and Mixed Ancestry R42 823 (R3 569 per month) (Table 3). The participants in this cohort earned an average annual income of income of R257 064 (R21 422 per month) confirming the findings of Bloch et al (1989) that individuals undergoing PT are from a higher socio-economic background.

Level of education

In the general population of the WC only 11.2% have more than one year of tertiary education, whereas 77.8% (21/27) of this cohort had an average of 2.7 years of tertiary education, which is similar to the findings of Craufurd et al (1989) and Paulsen et al (2006) that people who undergo testing are more likely to have higher educational levels than the general population.

Marital status and number of children

Four (15%) of the participants were single and 85% (23/27) were married at the time of testing. Prior to the first PT visit, 66.6% (18/30) of the participants had 1 or more children (Table 4). These results are similar to those of Trembath et al (2006); Velakoulis et al (2005) and Creighton et al (2003).

Number of children	0	1	2	3	4	5	Total
Frequency	9	6	6	5	1	0	37
Number of grandchildren	23	1	0	0	2	1	14

Table 4: Number of children and grandchildren per participant (n=27); **(mean number of children 1.4, grandchildren 0.5)**

There were no participants who were pregnant at the first visit, but subsequent to the test disclosure, 7 participants reconsidered having children/more children (Table 5).

P	PT result	Decision	Outcome
1	Gene-positive	Not to have offspring	Adopted a child
1	Gene-positive	To have another child	Unable to have vasectomy reversed
2	Gene-positive	To have own child	Trying to fall pregnant
1	Gene-negative	To have own children	On infertility treatment
1	Gene-negative	To have another child	Third child
1	Gene-positive	Not to have more children	No further children

Table 5: Child-bearing decisions following PT

Distance to the test centre

Eight-five percent (23/27) of the participants lived within a radius of 40 kms of the test centre and owned a vehicle, so the distance to the test centre did not impact on their attendance at the clinic. However, 14.8% (4/27) lived 769 kms and 1079 kms away and found the traveling time, costs and accommodation expenses a major consideration in completing the full PT programme requirement of 4 visits. Their protocol was, therefore, reduced and they attended only 2 and 3 visits respectively. None of the 11.1% (3/27) drop-outs had any difficulties with the distance or transport as they lived within 20 km of the centre and owned their own vehicles.

General social problems experienced

	Items	No problem 1	Mild problem 2	Moderate problem 3	Severe problem 4	Complete problem 5
G1	Lack of money	16	1	7	0	3
G2	Transport	24	1	1	1	0
G3	Housing	24	0	1	0	2
G4	Safety	27	0	0	0	0
G5	Unemployment	26	0	0	0	1
G6	Isolation and loneliness	24	0	1	1	1
G7	Privacy	27	0	0	0	0
G8	Health Services	24	0	1	0	2
G9	Social Services	25	0	1	0	1
G10	Domestic assistance	26	0	1	0	0
G11	Attitude of the community	24	0	1	1	0
G13	Abuse	20	0	1	1	5
G14	Disability	24	0	0	0	3
G15	General health	27	0	0	0	0

Table 6: General social problems experienced by PT participants (n=27)

As the majority of the participants of the PT programme were from a high socio-economic group they did not experience many general social problems (Table 6). The participants 11.1% (3/27) who rated 'lack of money' as a 'complete problem' were the individuals who had developed symptoms and only received a DG. They also complained of isolation and loneliness when their spouses were at work and they were left alone at home during the day. Housing was a problem for 2 of those who were affected as 1 had become divorced and was renting a backyard apartment and the other required a larger apartment as she needed a carer 24 hours per day and she and her husband had a one-bedroom flat. The remaining 88.8% (24/27) owned their homes. The houses of all of the participants in this part of the study comprised a kitchen, at least one bathroom and sitting room, and at least one bedroom per member of the family. All participants owned a vehicle or, in the case of one who had become affected, a car was available in the family. Most of the participants (88.8%, 24/27) had private medical insurance and utilised private medical facilities. The 11.1% (3/27) individuals who were symptomatic found their disability to be a 'complete problem' as they were becoming increasingly dependent on a carer. The 11 individuals who reported abuse as being a problem as a child had had a mother who was affected with HD and they were not able to provide the role of protector to the children, leading to abuse by other children, family members and people from the community (Table 7). This finding confirms that of other studies (Dubinsky 2006; Paulsen 2006; Rosenblatt et al 1999). The abuse experienced as adults included psychological and/or physical abuse by their husbands.

	Perpetrator	Psychological	Physical	Sexual	Frequency	%
Childhood	Children	3	2	0	5	45
	Family	2	2	1	5	45
	Community	1	0	0	1	10
	Total	6	4	1	11	100
Adulthood	Family	10	3	0	13	100
	Total	**10**	**3**	**0**	**13**	**100**

Table 7: Frequency and type of abuse experienced by PT participants

4.4 GENERAL PROFILE OF INDIVIDUALS UNDERGOING PT

Family history

At the first visit there were only 3.7% (1/27) of the individuals who did not know of a family history of the disease. This person's parents had died in their late forties and early fifties respectively of cancer and had not shown any symptoms, although a grandparent had had some undiagnosed neurological condition. Ninety-six percent (26/27) of the participants were able to identify an affected parent who had been diagnosed genetically and/or clinically as having HD and, therefore had a prior genetic risk of 50%. These findings are similar to Tassicker et al (2006), Trembath et al (2006) and Creighton et al (2003) and Harper et al (2000) where 94%, 84%, 89.4% and 91% respectively of their cohorts had a prior risk of 50%.

There was a significantly large number of participants who had a maternal family history of HD compared to a paternal one (81% vs. 18%, p<.001) (Table 8) confirming the findings of other studies (Trembath et al 2006; Goizet et al 2002; Decruyenaere et al 1999; McCusker et al 1996). These authors suggest that a possible reason for this is that paternally transmitted HD shows greater anticipation than maternal transmission and therefore, a

Psychosocial effects of HD family members 'taking care' of affected family members

	Items	No problem 1	Mild problem 2	Moderate problem 3	Severe problem 4	Complete problem 5
C14	Emotional strain	2	3	0	2	17
C15	Physical strain	10	1	1	4	8
C16	Financial strain	6	1	6	2	9
C17	Coping with normal family duties	7	0	5	3	9
C18	Marital discord	12	4	1	0	7
C19	Family discord	11	2	2	1	8
C20	Physical caring of affected person	4	2	4	1	13
C21	Lack of respite	5	2	4	2	11
C22	Lack of personal time	7	0	6	0	11
C23	Deterioration of personal health	15	0	1	0	8
C24	Isolation	8	1	4	2	9
C25	Feelings of resentment	12	0	1	2	9

Table 9: Psychosocial effects of caring for affected family members (n=24)

The psychosocial effects of the 35.2% (24/68) individuals from both the PT group and patients group who were involved in 'taking care' of affected family members while they were growing up and as adults are combined in Table 9. There were more PT individuals 48.1% (13/27) involved in the caring of affected relatives than those from the patient group 21.6% (9/41). This might account for part of the reason for these individuals requesting PT as they were more familiar with the disease and its implications.

The relationship between the items measuring psycho-social effects of taking care of HD members by the participants was investigated by a CAT-PCA on the 12 items (C14 to C25) and number of years of taking care. A two-dimensional solution explained almost 73% of the total variance. Figure 4 reveals five clusters.

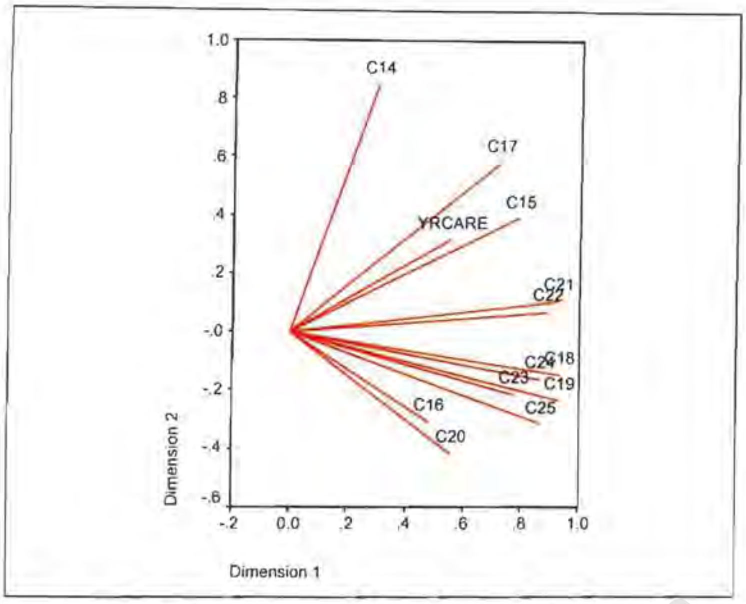

Figure 4: A plot of component loadings of the 12 items

The vector 'emotional strain' C14 appears somewhat on its own, but dominates the second dimension. This suggests that clusters that load relatively high on dimension 2 are perceived as having strong emotional aspects.

The second cluster (Figure 5) suggests that the strain associated with the number of years is mainly 'physical strain' and 'coping with normal family duties' (C17, C15).

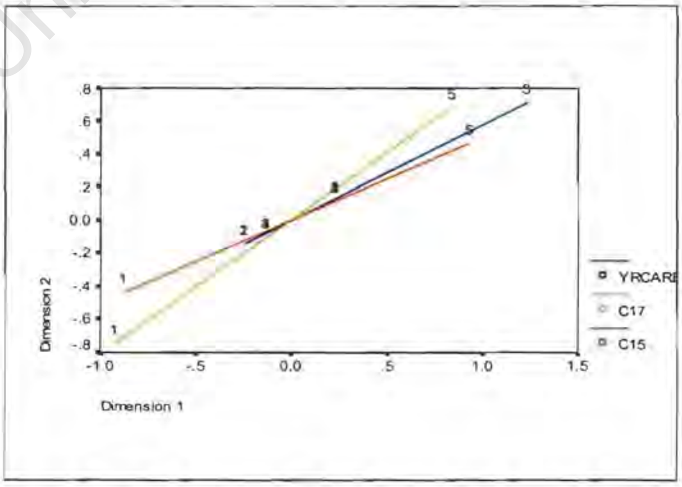

Figure 5: Category points of years of care and items C15 and C17

The next cluster is comprised of C21 and C22; 'lack of respite' is strongly associated with the 'lack of personal time'. Further, the relationship between 'financial strain' and of 'physical caring' for the affected person (C16,C20) suggests the need for external support to meet the specific demands of the affected person.

Items C18, C19, C23, C24, and C25 are interrelated and form a cluster that refers to psycho-social interactions within a family clearly related to the presence of an affected HD member of the family.

Availability of testing

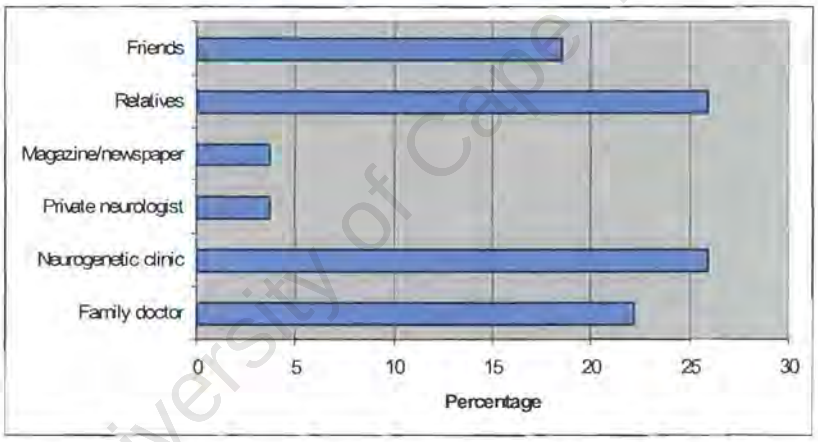

Figure 6: Knowledge of availability of testing programme

Participants discovered about the testing programme in a variety of ways, but mostly through a family doctor or the neurogenetic clinic when they were accompanying an affected relative to a doctor's or clinic appointment, or through family members (Figure 6).

Period of awareness before testing

The mean period of awareness before testing was 8.3 years (SD=9.9). Those whose prior exposure was categorised as 'none' waited 1 year; those who had limited contact 11 years; and those who had lived with affected schedule to comprehend the statements to which the item numbers refer in the scoring blocks.

- data referring to the general population of the WC have been obtained from the Statistics South Africa Central Statistical Services Official Population Census (2001).

Because the cohort is small, for ease of reading numbers less than 10 will be displayed as numerals rather than the conventional form of being presented in text.

4.2 PREDICTIVE TESTING PROGRAMME

The protocol for PT for HD (Appendix i) had been developed in 1994 (Division records) and had not recently been reviewed. As present practice had changed considerably from the document, this required revision to formalise the changes to ensure that these were captured in the information sheet distributed to potential PT participants and to ensure the protocol conformed to international practice.

A total of 36 individuals underwent the PT programme offered by the Division of Human Genetics, University of Cape Town (UCT) and Division of Neurology, Groote Schuur Hospital (GSH) from April 1995 to April 2005. There were no pre-natal tests or pre-implantation genetic diagnoses conducted during this period. All tests were by direct mutation analysis and all individuals lived in the WC. For this research, 83% (30/36) of the participants were interviewed. Three percent (1/36) was too severely affected to be interviewed and the spouse declined the request to be included in the programme and 14% (5/36) had emigrated and were not able to be located for a telephonic interview.

The uptake of PT was approximately 4.5% of the estimated at-risk population in the WC with a mean of 3 individuals tested per year which is slightly lower

- The majority of PT participants, patients and caregivers received at least two home visits, or interviews in the case of the PT individuals, by the researcher which allowed her to gain greater insight into the complexities of having HD or being part of a family with the disorder.
- By having time to establish a good rapport with the participants and their families there was sufficient time to use the opportunity available to discuss their day-to-day problems in coping with their test result or impact of the disease on the individual and family members.
- The researcher conducted all the interviews personally and, therefore, this limited the possibility of inter-rater reliability problems.
- Certain sections of the schedule contained scales used in similar international research increasing the reliability/trustworthiness of the study.
- All interviews were audio recorded to allow a more complete record than handwritten notes by the researcher and allowed the researcher to engage with the patients/participants without the necessary interruptions when taking notes.
- The recommendations of the participants and patients have all been addressed and the relevant changes and additions to the respective predictive testing programme and neurogenetic clinic have been or are in the process of being implemented.

longer latency of being asymptomatic and more time to present for predictive testing. In the case of this cohort, however, it appeared that the participants had "lost" their mothers to the disease during their childhood or adolescence. Most of them were very emotional when they described how she was there physically, but was unable to care for them, protect them or demonstrate any love for them. They expressed great love for their mothers and bore no resentment towards them for neglecting them during their childhood or the onerous caregiving responsibilities they had to undertake during their young lives. Twelve of these participants had not had any children and their main reason for testing was to ensure that they did not have children if they were going to develop HD and would not be able to care for them adequately.

	Participant		
Affected parent	**Male**	**Female**	**Total**
Paternal	1 (4%)	4 (14%)	5 (18%)
Maternal	9 (33%)	12 (45%)	21 (81%)
Unknown	1(4%)	0(0%)	1(4)
Total	**11 (41%)**	**16 (59%)**	**27 (100%)**

Table 8: Maternal and paternal family history

Prior exposure to Huntington disease

The participants' prior exposure to HD was categorised as none 14.8% (4/27) if they had been adopted or had left home before the parent had been diagnosed; having observed affected relatives living elsewhere 22.2% (6/27); having lived with an affected family member and only observed the symptoms 14.8% (4/27); or having lived with an affected family member and cared for or assisted with their care 48.2% (13/27). These findings are similar to that of Trembath et al (2006). The median age at which the PT participants in the WC cohort became aware of their personal at-risk status is 31 years and that of the patient group 34.5 years.

relatives waited the longest time of 13.3 years (p<.001). These findings confirm that of Trembath et al (2006) where those who waited the longest had prior exposure to severe HD. The mean length of time that the WC cohort waited before requesting PT was 8.3 years compared to 9.7 years in Australia (Trembath et al 2006). All of the individuals 11/27 (40.7%) who became aware of their 50% at-risk status for the first time when they were adults due to an apparent new mutation or because of adoption, presented for testing within 1 year. The female:male ratio of this group was 60:40 which was similar to that of the general cohort (59:41). The mean age of the participants who presented for testing within 1 year of learning of their at-risk status and who had little or no previous exposure to HD was 32.5 years compared to 35.1 years for those who had been raised in a family with affected individuals. This differs somewhat from the findings of Trembath et al (2006) of 23.8 years and 36.2 years respectively, where the mean age of those with little or no previous exposure to HD was much younger than the cohort in the WC study.

Life events triggering request for PT

Triggering event	Frequency	Percentage
Relative recently diagnosed with HD	16	59.2
Recent PT of relative	1	3.7
Family planning	3	11.1
Awareness of symptoms	2	7.4
Request/pressure by significant other	3	11.1
Development of meaningful relationship	1	3.7
Recent knowledge of availability of PT	1	3.7
Total	**27**	**100**

Table 10: Life event triggering request for PT

The significant life events that triggered the participants to request PT are presented in Table 10. They are similar to the findings of other international studies (Taylor 2005a; Wexler et al 2004; Evers-Kiebooms et al 2002; Rosenblatt et al 2001; Decruyenaere et al 1995b).

Reason for requesting PT

Although Decruyenaere et al (2003) found a subgroup of individuals whose reasons for testing were simply to relieve the uncertainty of not knowing without any further specific reason, this was not found to be the case in the WC cohort. Uncertainty was undoubtedly something that remained in the background for all of them, but was not the main reason for testing, apart from one individual. In this case he was attending counselling for substance abuse independently and was persuaded by the psychologist to have the genetic test as the psychologist felt that it would reduce the individual's anxiety which he perceived to be caused by the uncertainty of the individual's genetic status. Few participants had a single reason for requesting testing. Most of them cited two or three reasons. Although 62.9% (17/27) reported that it was to reduce uncertainty; 59.2% (16/27) that it was for planning for the future, and 33.3% (9/27) felt it was their responsibility, these were secondary to the prime purpose of informing their children of their status, guilt of passing on the gene to their offspring and family planning (Table 11).

	Frequency	Percentage
Reduce anxiety and uncertainty	17	62.9
Planning for the future	16	59.2
Responsibility	9	33.3
Informing children of their status	7	25.9
Guilt of passing on gene	7	25.9
Informing children of personal status	6	22.2
Family planning	5	18.5
Financial decision-making	3	11.1
Insurance purposes	3	11.1
Decision for relationship or marriage	2	7.4
Coercion/persuasion by doctor/family	2	7.4
Employment purposes	1	3.7

Table 11: Reasons for deciding to undergo PT

Emotional and psychological state at first contact

The emotional and psychological state of the participants at the first visit is presented in Figure 7.

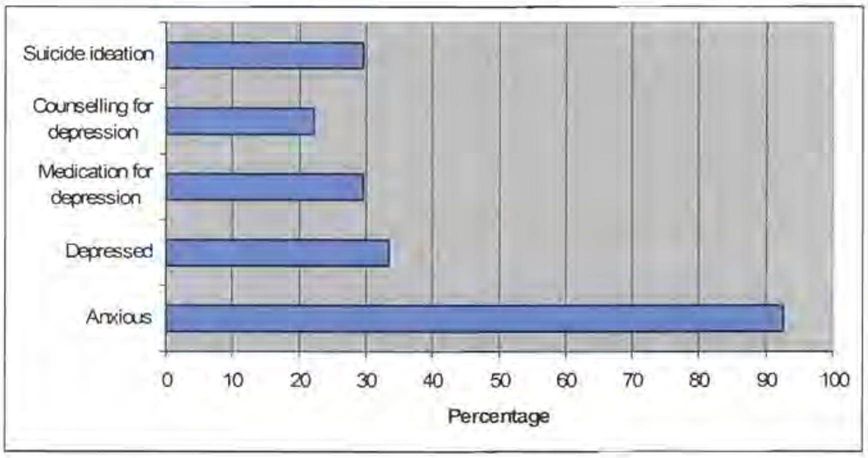

Figure 7: Emotional and psychological state at first counselling session

Many of them reported feeling more than one psychological state at the time. Ninety-three percent (25/27) reported being anxious at the first counselling session and 51.8% (14/27) had been receiving medication or psychological counselling for episodes of depression immediately prior to their PT programme. Of the 81.4% (22/27) participants reporting suicide ideation should they test gene-positive or being on medication or receiving counselling for depression, 27.2% (6/22) had been the main caregivers of their affected mothers during their teenage and adolescent years. Pilnick and Dingwell (2001) and Robins et al (2000) confirm that genetic information has the potential to significantly impact the psychological wellbeing and cause stress, anxiety and suicide ideation.

4.5 PREDICTIVE TESTING PROTOCOL

There are centres throughout the world where PT for HD is performed without providing counselling before, to prepare individuals for a positive or negative result, and after the test disclosure to support them if needed. The participants' responses to why they thought an extensive protocol was necessary for PT are presented in Table 12.

	Frequency	Percentage
Ensure understanding of HD	18	66.6
Ensure understanding of seriousness of decision	16	59.2
Allow sufficient time to reconsider decision	10	37
To prepare participant for the worst	5	18.5
Punishment	1	3.7

Table 12: Understanding importance of undergoing full test protocol

The majority of them provided more than one reason and the 18.5% (5/27) who responded that it was to prepare them for the worst were individuals who had tested gene-positive. One (1/27) individual was negative about the whole programme and considered it to be a 'punishment' even though he only attended 3 of the genetic counselling sessions because he lived 1 069 kms from the testing centre and the programme was curtailed for this purpose. He regarded the protocol as a waste of time since he considered that he had obtained all the information that he required from the internet and had made up his mind that he definitely wanted to know his test result. Three of the remaining 6 participants, who attended fewer than 4 sessions of protocol, maintained that since completing the PT protocol and having had time to reflect on the preparation they required to deal with the disclosure, they believed it to be essential and suggested that it should be even more lengthy and rigorous. These participants had grown up with an affected mother and felt that they needed further psychological counselling to deal with these specific problems before being confronted with the repercussions of either a positive or negative test result.

Attitude to termination of pregnancy

Participants' responses to their attitude to termination of pregnancy if the foetus were found to be gene-positive are presented in Figure 8. Although 74% stated that they did not agree with abortion as such they thought it was a personal choice when it came to a foetus who was gene-positive. Other studies have found that the most important reasons for couples not wanting antenatal testing was objection to termination of pregnancy and the hope that a cure would be found before their children developed symptoms (Downing

2005; Lea et al 2005; Evers-Kiebooms et al 2002; Hayden and Kremer 1995).

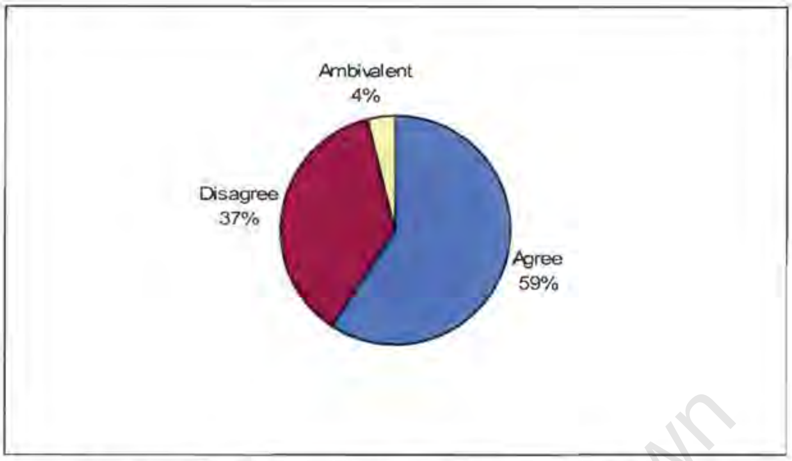

Figure 8: Attitude to termination of pregnancy if the foetus is gene-positive

Self-perception of gene status prior to PT

	Gene-positive	Gene-negative	Reduced penetrance	Ambivalent
Before	33.3% (9/27)	40.7% (11/27)	0% (0/27)	26% (7/27)
Test result	40.7% (11/27)	55.6% (15/27)	3.7% (1/27)	0

Table 13: Self-perception of gene status prior to PT (n=27)

Seventy-four percent (20/27) of the participants had definite perceptions about what their gene status was prior to testing (Table 13). Only 1/27 (3.7%) had been 'preselected' by her family as being the one to inherit the disorder as she resembled her affected mother and her family believed that females were more likely to develop symptoms of HD since her grandmother had also been affected. The rest of the cohort had no specific explanations as to why they believed that they were gene-positive or gene-negative. There was no relationship between their perceptions before the test and the test results.

Outcome of PT

The outcome of the testing is presented in Table 14. Results have been given to all participants in the cohort. Many studies of at-risk samples have found that those testing gene-positive are significantly younger than those testing gene-negative (Codori et al 2004); while others have found that the gene-positive group were 38 years compared to the gene-negative group of 40.2 years (Creighton et al 2003); and in the WC cohort the average age of the 40.7% (11/27) individuals testing gene-positive was 35.5 years and that of the 55.6% (15/27) testing gene-negative was 35.1 years. The results of those found to have expanded CAGn repeats (40.7%) is similar to the findings of other studies (Tassicker et al 2006; Trembath et al 2006; Taylor 2005a; Creighton et al 2003; Harper et al 2000; Craufurd et al 1992; Conneally 1984).

Completed tests	Gene-positive CAG>39 n(%)	Reduced penetrance CAG 36-39 n(%)	Gene-negative CAG<36 n(%)	Total
Female	6 (22.2)	1 (3.7)	9 (33.3)	16 (59.2)
Male	5 (18.5)	0 (0)	6 (22.2)	11 (40.7)
Total	**11 (40.7)**	**1 (3.7)**	**15 (55.6)**	**27 (100)**

Table 14: Outcome of PT by gender

The CAGn repeat lengths of the participants are presented in Table 15.

CAGn repeat length	Frequency	Percentage
<27	15	55.5
39	1	3.7
40	1	3.7
41	2	7.4
42	2	7.4
44	2	7.4
45	2	7.4
47	1	3.7
53	1	3.8
Total	**27**	**100**

Table 15: CAGn repeat lengths

Disclosure of test results

Disclosure of test results was very much of a personal choice and there was no specific trend as to who the participants selected to divulge the information and the reason they had been selected. Only 18.5% (5/27) of the participants disclosed their genetic test result to 'everybody'. Seven percent (2/27) had tested gene-positive and came from a family where HD had only recently been diagnosed and the other 11.1% (3/27) had tested gene-negative and had lived with and cared for their affected mothers during their teenage and adolescent years. The remainder of the cohort disclosed their results selectively. All of them had taken their spouses/partners to the counselling sessions as support partners and, therefore, they knew the results. Some individuals did not divulge the information to their parents because the parents had not wanted their children to undergo testing. Others who tested gene-positive did not want to cause their parents guilt and anxiety about them developing symptoms later in life confirming the findings of Arnold et al (2005) and Knebel and Hudgings (2002). Individuals who tested gene-negative did not see the necessity of informing their children of their status while some of those who tested gene-positive did not tell their children if they thought they were too young. Some individuals informed all of their siblings of their status whether they had tested gene-positive or gene-negative while others who had tested gene-negative sometimes chose not to inform affected siblings of their status. Few elected to make the information known to relatives outside of the immediate family. Individuals who did not have a spouse or children disclosed their positive status to a few close friends. Several participants recommended that genetic counsellors caution the individuals undergoing pretest counselling against divulging their gene-status to friends or relatives who may not heed their request of keeping it confidential if they do not wish the information to become public. P7 voiced this as,

"I told my best friend about my test and told her it was very confidential. A few weeks later my father challenged me about it as he had heard the information from a close family friend who was also a friend of my friend. My family did not want me to go for testing and now have detached themselves from me and treat me as though I have betrayed them".

Four (14.8%) individuals disclosed the information to their employers as they had had to ask for time off to attend genetic counselling sessions and felt they had to explain the reason for taking so much leave when they were medically well. Fifty-two percent (14/27) cited 'moral duty' as the main reason for disclosing their gene status to their selected family or friends.

Reasons participants gave for not disclosing their gene status to everybody is presented in Figure 9.

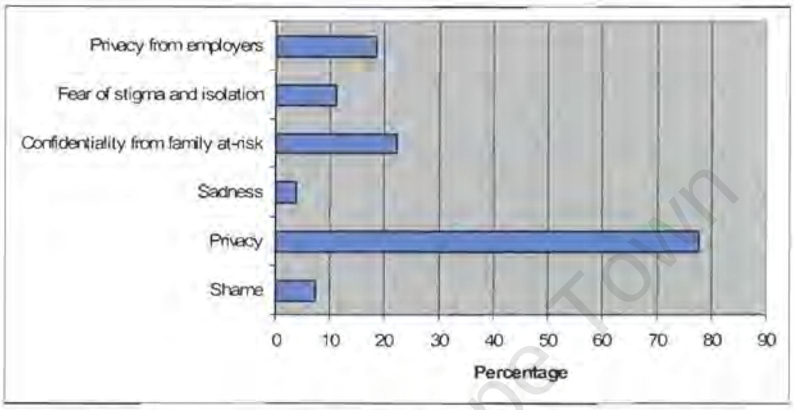

Figure 9: Reasons for disclosing predictive test results to selected people only

4.6 EMOTIONAL AND PSYCHOLOGICAL REACTIONS TO TEST RESULT

Within first seven days after test disclosure

Two (7.4%) individuals developed major adverse reactions within the first 7 days of hearing their test result; one was hospitalised for major depression and the other for suicide ideation. The individual who developed severe depression had tested gene-negative and the one who was at-risk of attempting suicide was found to be gene-positive. The worldwide risk of major catastrophic events of suicide, suicide attempt and psychiatric hospitalisation is 0.97% (Timman et al 2004; Almqvist et al 1999). Furthermore, 4 (14.8%) participants were so traumatised by their test results that they needed to consult their private medical practitioners who provided them with medication and/or referred them for counselling. One (3.7%) had a result of reduced penetrance and 3 (11.1%) had tested gene-positive.

Emotional and psychological state one year after PT

	Before PT	After PT	Total
Gene-positive	5 (18.5%)	9 (33.3%)	14 (51.8%)
Gene-negative	7 (25.9%)	9 (33.3%)	18 (66.6%
Total	**12 (44.4%)**	**18 (66.6%)**	

Table 16: Frequency of depression before and after predictive testing

The self-reported frequency of episodes of depression severe enough to warrant medication or counselling before and after PT is presented in Table 16. There is no relationship between depression before and after PT or whether the test result was gene-positive or negative which is similar to the findings of Velakoulis et al (2005), but unlike other studies where a history of depression before testing increased the risk of an adverse outcome regardless of whether the result was positive or negative (Blain and Brooks 2007; Pakenham et al 2004; Shannon 2004; Decruyenaere et al 2003; Brouwer-DudokdeWit et al 1998; Evers-Kiebooms and Decruyenaere 1998). Eleven (40.7%) had depression before and after the test disclosure, 6 (22.2%) had tested gene-positive and 5 (18.5%) gene-negative. One participant who tested gene-negative had depression before, but not after the result. Seven (25.9%) participants had depression after the test result, but not before; 3/27 (11.1%) of had a positive test result and 4/27 (14.8%) a negative result. These findings differ significantly from that of other studies (Marshall et al 2007; Codori et al 2004; Tibben et al 1997b). In the study of Codori et al (2004) of major depression 1-year after PT they found that the prevalence of depression in those who had a gene-positive (6%) result was twice that of those with a gene-negative result (3%) while Tibben et al (1997b) found that approximately 10% to 20% of both mutation-positive and mutation-negative individuals develop psychiatric symptoms after PT; whereas in this cohort 66.6% of the individuals developed depression. Because the individuals testing positive and negative in the present cohort showed no difference in levels of depression before and after PT it would not be possible to predict which individuals are likely to experience more distress. The frequency of depression among individuals who underwent testing within one year of first learning of their at-risk status and those who waited longer are presented in Table 17.

	Frequency	Test result
< 1 year	36.3% (4/11)	Gene-positive
	27.2% (3/11)	Gene-negative
Total	63.5% (7/11)	
> 1 year	31.2% (5/16)	Gene-positive
	37.5% (6/16)	Gene-negative
Total	68.7% (11/16)	
Total	**100.00 (18/27)**	

Table 17: Frequency of depression and waiting period before genetic testing

This is possibly the reason so many authors caution against PT before a substantial period of time has lapsed (Ensenauer et al 2005; Lea et al 2005; Sarangi et al 2005; Myers 2004; Taylor 2005a; Craufurd and Tyler 1992). This is confirmed by the following extracts of individuals who had been tested within the first year of becoming aware of a relative being diagnosed with HD:

"I think I tried to do too much in that time. My mother had been living with us for a few years and was becoming worse and more difficult to care for. Then finally when she was diagnosed I thought I had to know as soon as possible for my children's sake and it was just too much to come to terms with so soon. I think I would have been much more stable in myself after a longer time to adjust to her diagnosis and sorting out her future care". P18 who had a gene-negative result.

"It was all so terrible. My mother had just been diagnosed and it was the first time we had even heard about a disease like this. Then I was told I was also at-risk. I panicked and thought that I just had to know whether I was going to get this awful disease and thought it was merely going to reassure me that I was negative, but instead I was told I was positive and nearly went mad. I think that if I had had more time to consider the options and once I knew more about the disease I would have been better able to bear with the result". P11 who had tested gene-positive.

"Even though I tested negative when I think of it now I realise that I should have the four full counselling sessions or even many more before the test and that it would have been much better to wait a much longer time to know the result. To be told suddenly that I had this threat above my head and for me to hear my husband say that he would not stay with me if I developed the disease was such a shock to me. But I think if I had had more time to think about my future and come to terms with my husband's attitude I would have been able to cope much better". P18.

The 15 participants who had received a negative-gene result described their emotional and psychological state at the end of the first year after PT as presented in Table 18.

	Frequency	%
No difference	4	14.8
Relief of negative result	5	18.5
Relief of uncertainty	8	29.6
Sadness	9	33.3
Preoccupied with thoughts and memories of present and deceased affected family members	6	22.2
Moderate depression	9	33.3
Breakdown of important relationships	10	37
Substance abuse	3	11.1
Worry about future caregiving of affected family members	4	14.8
Concern about accuracy of test	4	14.8

Table 18: Emotional and psychological state of gene-negative participants one year after receiving test results (categories are not mutually exclusive)

The frequency of depression reported (33.3%, 9/27) in this study is much higher than that found by Codori et al (2004) whose cohort of gene-negative individuals experienced depression identical to that of the general population, namely, 3.0%. None of the individuals in the WC study described 'survival guilt' as one of the reactions to their test result. They differentiated this from 'sadness' for their relatives as they did not wish to have the symptoms instead of their affected relatives, but rather felt deep sorrow and despondency for them. None of the participants in this group reported any difficulty in adapting to their test result in spite of 18.5% (5/27) of them considering themselves to be gene-positive before the test. This is contrary to the findings of Tassicker et al (2006) who found that some individuals who learned of their HD risk status during adolescence and who tested gene-negative also have adjustment difficulties sufficiently severe to warrant post-test psychological support. There was a breakdown in marriage in 7.4% (2/27) of the couples where the participant had tested gene-negative. Both of these had been psychologically abused by their husbands prior to testing and the participants had tolerated it as they believed that they were dependent on them to care for them should they develop symptoms of HD in the future. Richards and Williams (2004) found that the stress in marriage had been previously attributed to their HD risk, but once this was removed the stress remained and they resorted to divorce. A further 29.6% (8/27) experienced a breakdown in the bonds between themselves and their relatives when they tested negatively, a finding corroborated by Duncan and Delatyicki (2006) and Meiser and Dunn (2000).

	Frequency	%
Relief of uncertainty	1	3.7
Difficulty in adapting to test results	6	22.2
Numbness	10	37
Guilty of passing HD gene down to offspring	6	22.2
Pessimistic about the future	5	18.5
Angry with family for passing down HD	2	7.4
Concerned about onset of symptoms	8	29.6
Moderate depression	9	33.3
Suicide ideation	6	22.2
Concern about future care of children	6	22.2

Table 19: Emotional and psychological state of gene-positive participants one year after receiving test results (categories not mutually exclusive)

The emotional and psychological state of the 11 participants receiving a gene-positive result is described in Table 19. The most reported reaction was that of 'numbness' as the participants felt that they had lost control of their lives and that they would not be able to fulfill their original life plans. They stated that they had difficulty in adapting to their test results; 22.2% (6/27) had thought they were gene-negative before the test. The prevalence of self-reported depression requiring medication or counselling was 33.3% (9/27) which was also much higher than that of the cohort of Codori et al (2004) where the prevalence of depression was found to be 6% in those testing gene-positive and 3% in those testing gene-negative.

In spite of the negative reactions the participants attributed to the predictive test results 100% of them maintained that they had no regrets about having undergone the test, that they found it beneficial and that they would recommend other at-risk individuals to undergo the test. This confirms the findings of Almqvist et al 2003; Brower-DudokdeWit et al 2002; Horowitz et al 2001; Meiser and Dunn 2000). Forty-eight percent (13/27) believed that underage people should be allowed to be tested provided that they requested it themselves and it was ascertained by the genetic team that there had been no coercion in their decision and confirmed their "readiness" for the test. Reasons given were that early testing would reduce their level of anxiety and that they would be able to plan for their future appropriately. The 52% (14/27) who disagreed with children being tested claimed that they should be allowed to enjoy their childhood without the concerns of a possible gene-positive status being confirmed and that they were too emotionally immature to deal with the consequences of a gene-positive result.

Present strategies of coping with gene-positive result

Individuals with gene-positive results used a variety of strategies to cope with the knowledge that they would develop symptoms sometime during their lives and that their children were at-risk of inheriting the mutant gene. Family support was cited as the strategy most utilised for coping (Table 20). Individuals who had a religious conviction found that their personal faith and church members provided them with a great deal of support supporting the findings of French and Sims (2004) and Kolt and Anderson (2004) that religious values provide a sense of meaning to life and enhance the individual's experience of social and emotional support during threats of serious illness.

	Frequency	Percentage
Planning for future	3	11.1
Comforting ideas	3	11.1
Thinking positively	1	3.7
Constructive avoidance thoughts and behaviour	2	7.4
Live day by day	4	14.8
Make the most of life	2	7.4
Family support	7	25.9
Support of church	2	7.4
Personal faith	6	22.2
Counselling	5	18.5
Medication	4	14.8

Table 20: Present strategies of coping with gene-positive result

Concerns since receiving gene-positive or -negative test result

There was only 1/27 (3.7%) individual who did not have any concerns after the test result. This individual did not appear to have a family history of HD as his parents had both died relatively early from cancer and his grandmother had had some undiagnosed neurological condition. Otherwise all gene-positive and gene-negative individuals experienced difficulties as presented in Table 21.

	Frequency	Percentage
Worry about future care of spouse and children	6	22.2
Worry about risk status of children	6	22.2
Worry about untested relatives	7	25.9
Symptom watching	6	22.2
Sadness for affected relatives or those who tested gene-positive	10	37
Distancing by relatives	4	14.8

Table 21: Concerns since receiving predictive test result

The participants described how these concerns were constantly with them and hung like a shadow over their lives every day either in the form of concern for affected relatives or those who might develop the disease, or about their own inevitable symptoms and the form they would take and when they might appear. In the words of P3:

"I've had this heavy feeling for most of my life. I remember when I was young and my mother was well we all had so much fun. Then she became ill and became less interested in me and I started with this heavy feeling. Later I helped to care for her until she died. The heaviness became worse and I went to a psychologist for a while to get over her death. But much later I was still feeling this heaviness and thought it must be associated with the uncertainty of whether I was going to develop the disease someday. For a few years it didn't get any better so I thought by having a test and definitely knowing I would be able to get on with my life. The test was negative, but that heaviness has not left. Now I am depressed about my sibling who has tested positive and who has a beautiful little child and I still feel this heaviness when thinking about my mother and what my sister and her child still have to face".

P3 described her emotional experience since testing as, *"When I became aware that my family had HD I rushed off to be tested as I thought that I could not live with the uncertainty and would be able to plan for my future and that of my children if I knew what my status was. However, now I think that it may have been better in the long run to live with the uncertainty as I am constantly wondering when I am going to develop symptoms and everyday I bump into furniture or drop something I query whether the disease has got to me. And now I have the added worry that my children are definitely at-risk now whereas before it was only a possibility".*

P18 found that, "*Since I received my negative result my relatives do no want to discuss HD or PT with me and they don't come and visit or invite us to visit them anymore and we used to be a close family. They treat me as though I am a criminal or a leper. It is very depressing, but I am very pleased that I have been able to reassure my children that they and their children are clear of the disease*".

These findings are all similar to other studies of psychological reactions to PT of HD (Semaka et al 2006; Sobel and Cowan 2003; Maat-Kievit et al 2001; Van t' Spijker and Ten Kroode 1997; Fox et al 1989).

4.7 EVALUATION OF THE PT PROGRAMME

Knowledge of HD

Table 22 presents the participants' knowledge about HD as this was the content covered during the counselling sessions.

Item	Knows	Does not know
B1 How do people get HD?	25	2
B2 What part of the body does it affect?	27	0
What signs can you see that a person has HD?	27	
B3 unwanted movements/slow movements/unable to move	27	0
B4 loss of memory/decreased intellectual capacity/dementia	22	5
B5 anxiety/irritability/depression/personality changes	18	9
B6 How old are people when they first start to show the signs of HD?	25	2
B7 Is there a cure for HD?	27	0
B8 What is the treatment for HD?	27	0
What are the risks of your family having HD?		
B9 brothers and sisters	25	0
B10 parents	25	0
B11 children	26	1
B12 yourself	24	3
What kind of genetic disorder is HD?		
B13 autosomal	1	26
B14 dominant	3	24
B15 Are males and females equally affected with HD?	27	0

Table 22: Participants knowledge of HD (n=27)

The majority had a very good understanding of it even though their contact with the PT programme had been from 1-10 years prior to the study, achieving an average score of 81.3% (12.2/15) which is contrary to other research findings where 40-80% of medical information provided by healthcare professionals was forgotten immediately, the more content provided the less was correctly remembered and that nearly 50% of the information was incorrect (Kessels 2003; McGuire 1996; Anderson et al 1979). However, the genetic information provided by the genetic team in this study was repeated several times during the programme and the participants themselves used the internet to search for additional information which also allowed repetition of information whereby it could be consolidated. The participants were from a higher income group and educational level and Eggers et al (1999) found that in this group understanding and recall was significantly higher. However, there were 3 participants who had misunderstood their test results. Two of them thought they were gene-positive as they were told that although their CAG repeat length was normal there was still a small possibility of them developing HD although their children were gene-negative and would never develop symptoms. Several authors caution counsellors to refrain from dwelling on the uncertainties and to focus on information based on certainty to avoid confusing the individuals (Semaka et al 2006; Van Den Boer-van den Berg and Maat-Kievit 2001). The selective nature of remembering information at a time of anxiety has been studied by Michie et al (1997b) who found that aspects related to family matters were recalled 100%, but there was only 68%-78% recall of issues related to genetic or medical information. P14 had been under this misperception for 9 years and P19 for 10 years. When P11 was told that her test result was not gene-negative two years previously she reacted very emotionally and rushed off without taking her test result report with her and before the counsellor was able to explain, in detail, that she had a reduced penetrance result and the implications thereof. She had only recently become aware of the diagnosis in the family and had immediately requested testing to reduce the uncertainty. Semaka et al (2006) found that with an intermediate result individuals may experience adverse psychological consequences because the very reason for them wanting to have the test, namely, to remove the uncertainty has not been reduced. During the research interview P11 was advised to contact the genetic counsellor for another appointment to have the results explained in depth and to receive the written report of her CAG repeat length.

Only 14.8% (4/27) were able to respond correctly to the item on the type of genetic disorder to which HD belonged. When explanations were provided by the researcher the participants claimed not to have heard of the terms previously in spite of it having been provided during the PT counselling sessions and fifty-nine percent (16/27) of them having reported that they had used the internet regularly for more information about the disease at the time of testing and subsequently for updates on new research published. It may have been due to the time lapse between testing and the evaluation (De Pina-Neto and Petean 1999; McGuire 1996) or due to the fact that the majority of participants were anxious at the time of their PT and that they did not pay the same amount of attention to that particular information (Kessels 2003; Hammond 1998).

Satisfaction with the PT programme

The results are presented in two parts. The first section deals with the analysis of relationships between and within demographic variables and interview schedule variables (items). As Table 25 is ordinal and it would be too strong an assumption to attribute higher numerical properties to the responses and treat them, initially at interval or ratio level of measurement, CAT-PCA was used to analyse the interdependency between demographic and responses to the satisfaction scale. The second section non-parametric statistics were used to evaluate the actual level of satisfaction with the programme in the context of the explorative insights.

Part 1

Firstly, certain demographic variables are analysed. Table 23 presents a number of demographic variables that are considered to be of relevance to the differentiation in responses to the level of satisfaction scale.

Item	Category		
	1	2	
Gender (A1)	11 (Male)	16 (Female)	
Symptoms (A19)	1 (Yes)	26 (No)	
Positive test result (A22)	12 (Yes)	15 (No)	
Hospitalisation (E1)	2 (Yes)	25 (No)	
Counselling (E4)	7 (Yes)	20 (No)	
Affected Parent (A14)	5 (Mother)	22 (Father)	
Marital status (A10)	4 (Single)	18 (Partner)	5 (Divorced)
Educational level	6 (H School)	21 (Tertiary)	

Table 23: The occurrence of categories of a subset of demographic variables (n=27)

To explore a possible relationship between these variables and the responses to the satisfaction questionnaire, only items with sufficient variance (category differentiation) were selected. Thus the variables 'symptoms' (A19) and 'hospitalisation' (E1) were not included in the analysis.

A two-dimensional CAT-PCA revealed that only gender appears to have a relevant association with some items of the satisfaction scale. Variables 'affected parent' and 'positive test result' explained only their own variance. This will be discussed further at the end of part 1.

The analysis also revealed two outliers as indicated by Figure 10. P17 is located in the extreme left bottom. This suggests a response pattern to the satisfaction scale that deviates significantly from those of the other participants. P18 also appears to have responded differently, although less extreme, from the other participants.

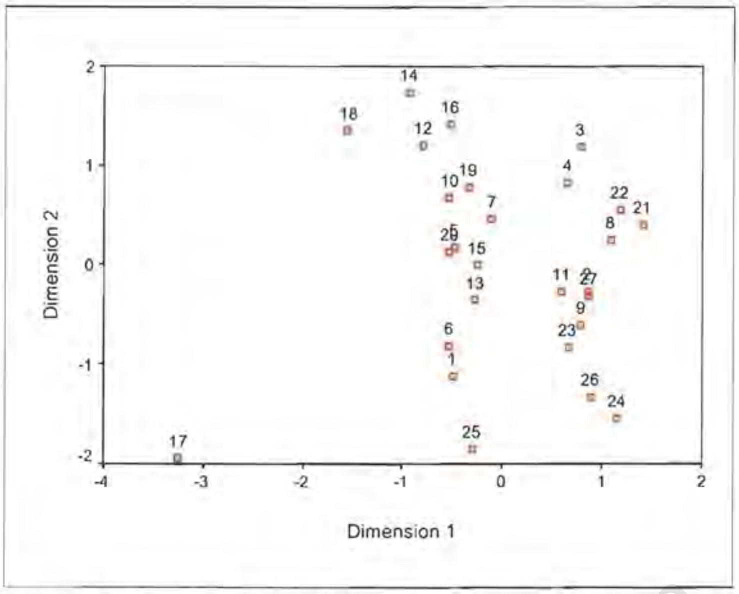

Figure 10: Plot of the participants (objectscores)

Table 24 shows that P17 rated all items of the satisfaction scale as '1' apart from item 18. P18 almost dichotomised the rating scale into either 'very unsatisfactory' or 'very' and 'highly satisfactory'. Participant 17 is a single male. Participant 18 is a female with a partner (or married). Both individuals studied at tertiary institutions. P17 was removed from further analysis.

Participant	Item number									
	D1	D2	D3	D4	D5	D6	D7	D8	D9	D10
17	1	1	1	1	1	1	1	1	1	1
18	5	1	1	1	1	1	1	4	4	4

Participant	Item number								
	D11	D12	D13	D14	D15	D16	D17	D18	D19
17	1	1	1	1	1	1	1	5	1
18	1	1	5	5	4	1	1	5	1

Table 24: Ratings of participants 17 and 18 per item of the satisfaction scale

Before exploring, in a more dimensional space, associations between the response patterns of the items, including the gender variable, the frequency distribution of the ratings was investigated.

Items		Very unsatisfactory	Unsatisfactory	Satisfactory	Very Satisfactory	Highly satisfactory
		1	2	3	4	5
D1	The manner in which HD was explained to you	0	0	4	8	14
D2	The doctor/counsellor/sister listened to what you had to say	1	0	4	11	10
D3	The doctor/counsellor/sister understood what was worrying you	3	6	3	7	7
D4	The doctor/counsellor/genetic sister answered all your questions	2	3	4	9	8
D5	The doctor/counsellor/genetic sister showed you a lot of caring	1	1	8	7	9
D6	The counselling helped you in making your decision about being tested	3	1	4	7	11
D7	The counselling helped you to cope better with your problems	9	6	6	1	4
D8	There was enough time given to you at each session	2	1	4	6	13
D9	The clinic environment	7	4	4	6	5
D10	The doctor/counsellor/genetic sister reassured you	0	0	3	10	13
D11	The manner in which the test results were presented to you	3	5	5	7	6
D12	There was time in the visits to have your questions answered	6	1	2	4	13
D13	The time you waited to get an appointment for your first visit	0	0	2	5	19
D14	The time you waited to see the counselling staff at each visit	2	0	1	4	19
D15	How satisfied were you with the information you received during the counselling sessions?	1	3	6	10	6
D16	The extent to which the team met your expectations of them	2	0	11	8	5
D17	Follow-up clinics/visits provided you with a lot of support	23	2	0	0	1
D18	General administration and concern shown by genetic sisters	0	0	0	1	25
D19	Overall, how do you rate the HD PT programme	1	0	10	10	5

Table 25: The rating frequencies per item on the satisfaction scale (n=26)

Table 25 shows that items D17 and D18 failed to generate variance in the responses. All respondents rated the 'general administration and concern shown by genetic sisters' (D18) as 'very/highly satisfactory'. A ceiling response such as this justifies its removal from further analysis. The same applies to item D17 with the difference that only 1 participant considered the 'Follow-up clinics/visits provided me with a lot of support' as 'highly satisfactory' as radically opposed to the other 25 who considered this item to be 'unsatisfactory' and 'very unsatisfactory'. Item D19, the overall rating of the HD PT programme, was not included as this item conceptually reflects a combination of all the items. It was designed as a 'direct' measurement of satisfaction that allows the evaluation of the contribution of clusters of items to the construct of satisfaction. It is dealt with in Figure 14 (page 183).

Shiloh et al (1990) found that there were three dimensions, namely, affective (A), instrumental (I) and procedural (P), which formed the basis of their satisfaction scale. The component loadings of this cohort are presented in Table 26. The distribution of component loadings in do not support three dimensions and the assumed three factors suggested by Shiloh et al (1990), namely, 'A' affective, 'I' instrumental, 'P' procedural.

Item	Dimension		
	1	2	3
D1 (I) The manner in which HD was explained to you	.792	.313	-.039
D2 (I) The doctor/counsellor/sister listened to what you had to say	.896	-.111	-.269
D3 (A) The doctor/counsellor/sister understood what was worrying you	.777	-.182	-.281
D4 (I) The doctor/counsellor/genetic sister answered all your questions	.824	-.247	.109
D5 (A) The doctor/counsellor/genetic sister showed you a lot of caring	.824	-.314	.201
D6 (I) The counselling helped you in making your decision about being tested	.823	.140	-.349
D7 (I) The counselling helped you to cope better with your problems	.822	-.050	-.013
D8 (P) There was enough time given to you at each session	.904	.014	.053
D9 (P) The clinic environment	-.046	-.564	.740
D10 (I) The doctor/counsellor/genetic sister reassured you	.793	.323	.112
D11 (I) The manner in which the test results were presented to you	.408	-.650	.118
D12 (P) There was time in the visits to have your questions answered	.814	.143	-.213
D13 (P) The time you waited to get an appointment for your first visit	.466	.726	.436
D14 (I) The time you waited to see the counselling staff at each visit	.465	.726	.437
D15 (I) How satisfied were you with the information you received during the counselling sessions?	.429	-.142	.240
D16 (I) The extent to which the team met your expectations of them	.652	-.583	.073

Table 26: Component loadings of a three-dimensional CAT-PCA

The analysis did reveal that item D9 dominates the determination of the third dimension although it contributes considerably to the second dimension (Table 26). Item D9 refers to how the respondents perceived the clinical environment. This item explained almost 4% of variance and with residue variance by the remaining 15 items, the contribution of a third dimension to the total fit was only 8.7%. These results suggest that the present sample of participants perceived or experienced aspects of items differently as the clusters of items could refer to different psychological constructs.

The size of the contribution of the third dimension and the identification of its main source do not outweigh the insights of a two-dimensional analysis. The results of a two-dimensional CAT-PCA are summarised in Table 27.

Dimension	Cronbach's Alpha	Variance Accounted For	
		Total (Eigenvalue)	% of Variance
1	.935	8.108	50.678
2	.659	2.620	16.375
Total	**.967(a)**	**10.728**	**67.053**

(a) Total Cronbach's Alpha is based on the total Eigenvalue

Table 27: Summary of a two-dimensional analysis

The component loadings of the two-dimensional CAT-PCA are presented in Table 28. The component loadings are represented as vectors in the two-dimensional space in Figure 11 and help to identify the possible clustering of the items.

Satisfaction items	Dimension	
	1	2
D1 The manner in which HD was explained to you	.794	.313
D2 The doctor/counsellor/sister listened to what you had to say	.904	-.102
D3 The doctor/counsellor/sister understood what was worrying you	.780	-.183
D4 The doctor/counsellor/genetic sister answered all your questions	.822	-.245
D5 The doctor/counsellor/genetic sister showed you a lot of caring	.805	-.351
D6 The counselling helped you in making your decision about being tested	.825	.143
D7 The counselling helped you to cope better with your problems	.827	-.040
D8 There was enough time given to you at each session	.907	.002
D9 The clinic environment	-.140	-.580
D10 The doctor/counsellor/genetic sister reassured you	.794	.301
D11 The manner in which the test results were presented to you	.405	-.671
D12 There was time in the visits to have your questions answered	.820	.129
D13 The time you waited to get an appointment for your first visit	.476	.707
D14 The time you waited to see the counselling staff at each visit	.489	.705
D15 How satisfied were you with the information you received during the counselling sessions?	.431	-.166
D16 The extent to which the team met your expectations of them	.637	-.596

Table 28: Component loadings per item and dimension of a two-dimensional CAT-PCA

Figure 11 shows the mutual association between the items in relation to each dimension after normalisation.

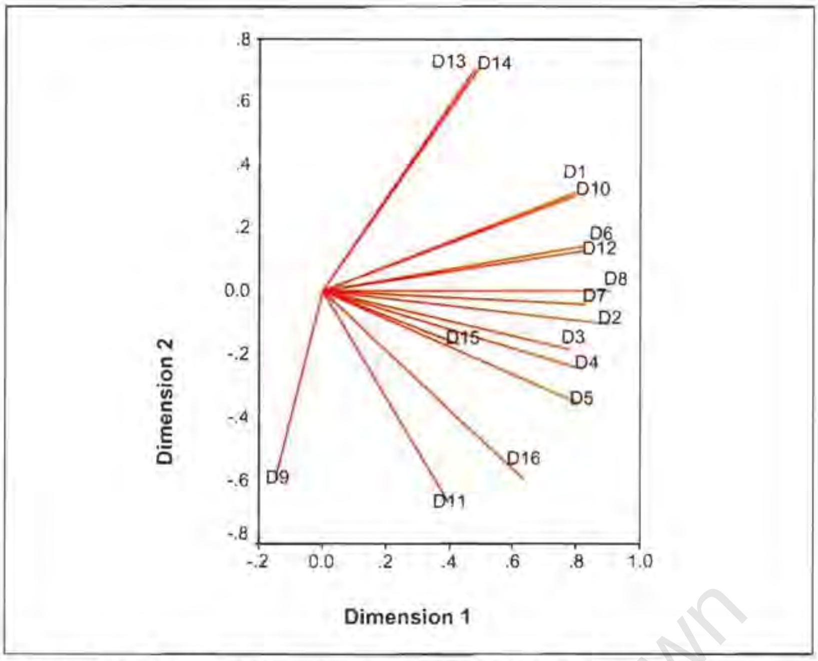

Figure 11: A plot of the component loadings of the 16 items

The figure reveals a set of local clusters. For example, items D13 and D14 are strongly related and contribute relatively strongly to dimension 2. Both D13 and D14 refer to the time to 'wait before one gets an appointment' and 'sees the counselling staff'. D9 also contributes to dimension 2. It refers to how the clinical environment is perceived. D9 is projected opposite D13 and D14, but under an angle, suggesting that the 'time to wait' might influence the perception of the 'clinical environment'. The plot (Figure 12) of the category points suggests, however, a very weak relationship. This is consistent with the initial finding that suggested that the 'clinical environment' refers to a different construct as do the other items.

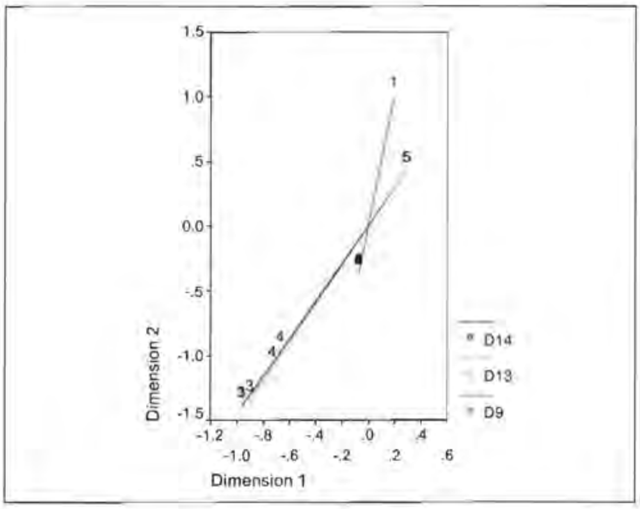

Figure 12: Category points of items D9 'clinic environment', D13 'waited for first appointment' and D14 'wait to see Counselling staff'

Moving clockwise (Figure 11) the next small cluster is formed by items D1 and D10. The cluster indicates that participants feel particularly reassured by the manner in which HD was explained to them.

The close proximity of items D6 and D12 indicates that satisfaction with the 'support in making a decision about the testing' is associated with the 'time spent to have the individual's questions answered'. The rather close projection of these items to D1 and D12 suggests that these also affect 'reassurance'.

The next cluster is formed by D2 and D7. This indicates that the individuals' perception of being 'listened to by the doctor/counsellor/sister' is strongly associated with the perception 'to be able to cope better with ones problems'. Item D8 is projected in between D2 and D12 indicating that the perception 'to be listened to' and the 'time spend to answer questions' are linked by being satisfied with the 'amount of time given at each session'. Furthermore, item D8 has the highest and an almost unique contribution to dimension 1.

The next cluster is less pronounced and is formed by items D3, D4 and D5. These items indicate that the level of 'understanding of what the participants are worried about' is associated with the 'feeling that all questions were addressed by the staff' and to the 'level of exposure of care by that staff'. The less pronounced presence of Item D15 suggests that the latter aspects are not strongly related to the extent to which participants were satisfied with the information during counselling.

Items D16 and D11 refer to the 'manner in which the results are presented' to the participants and the participants' 'expectations of the team'.

In summary, the third and fourth clusters, items D8, D2, D7; and D6, D12 refer, in particular, to aspects associated with the 'quality of information regarding the disease'. This group of items dominates the first dimension. The second group consists of the second and the fifth clusters. Items D1, D10 and D3, D4, D5, appear to refer to 'how much empathy the staff displays'. These items do contribute to dimension 1, but also, to a lesser extent, are important in determining dimension 2. The first and sixth cluster, items D13, D14, D9 and D11 dominate dimension 2, but are relatively strong contributors to dimension 1. Item D16 ambiguously contributes to both dimensions. These items refer to procedural conditions that, because of the contributions of the second group-items, appear to be experienced rather in the context of how much 'understanding' these conditions have for the dependency of the individuals. Thus, the participants in this cohort seem to perceive and experience the programme predominantly from two angles: in the context of the quality and accessibility of the HD information provided by the staff (dimension 1) and, the psychological context based on how much empathy the staff and the environment displayed (dimension 2). Although aspects of the original triad of affective, procedural and instrumental dimensions are present, they appear in this study, to represent two psychological constructs: the *quality* of the information provided by the counselling staff in open interaction and the amount of *empathy* the counselling staff displays. This confirms the findings of Green and Murton (1996).

Furthermore, as mentioned at the beginning of Part 1 regarding the relationship between the responses to the satisfaction scale and the demographic variables, only gender appeared of relevance. Figure 13 shows a plot of the category points of variable D15, the only variable gender seemed to affect, and gender. The plot suggests that in particular female respondents were satisfied with the information received during counselling sessions.

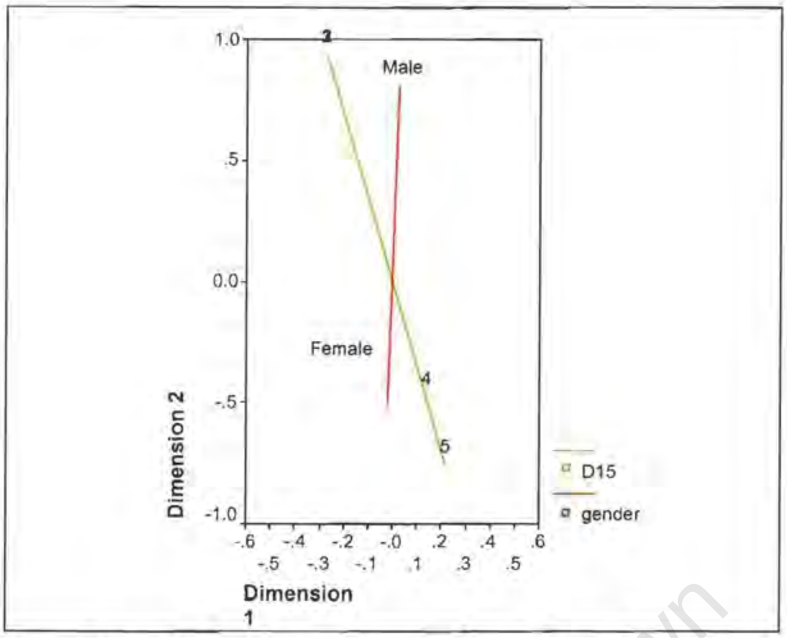

Figure 13: Category points of items 'gender' and D15 'information received during counselling sessions'

The relationship between 'gender' and D15 was tested post hoc. Table 29 shows the frequency distribution of the responses to this item by males and females. Since the five rating categories would generate low expectancy values that are too low the categories were recoded into two categories considering the frequency distribution of males and females on this item.

Gender	Rating D15					Total
	1	**2**	**3**	**4**	**5**	
Male	1	3	4	0	2	10
Female	0	0	2	10	4	16
Total	**1**	**3**	**6**	**10**	**6**	**26**

Table 29: Cross tabulation of gender with item D15 (information received during counselling sessions)

Table 30 shows the relationship between categories per gender after recoding. Although the relationship is statistically significant Chisq$_{(4)}$= 11.85; $p<.01$ one cell had an expectancy of 3.85. (The total is 26 as, stated on p34, Participant 17 was removed from further analysis).

Gender	Rating D15		Total
	Unsatisfactory	**Satisfactory**	
Male	8	2	10

Female	2	14	16
Total	**10**	**16**	**26**

Table 30: Cross tabulation of gender with recoded rating of item D15 (information received during counselling sessions)

Following the procedure suggested by Shiloh et al (1990) to establish a level of construct validity, the objectscores of dimension 1 and the objectscores of dimension 2 were correlated with item D19 'the overall rating of the satisfaction with the HD PT program'. The correlations are presented in Table 31.

		D19	Objectscores dimension 1	Objectscores dimension 2
D19	Correlation Coefficient	1.000	.718(**)	-.197
	Sig. (2-tailed)	.	.000	.335
	N	26	26	26
Object-scores dimension 1	Correlation Coefficient	.718(**)	1.000	.112
	Sig. (2-tailed)	.000	.	.584
	N	26	26	26
Object-scores dimension 2	Correlation Coefficient	-.197	.112	1.000
	Sig. (2-tailed)	.335	.584	.
	N	26	26	26

** Correlation is significant at the 0.01 level (2-tailed)

Table 31: Spearman Rank correlations between the objectscores of dimensions 1 and 2 and item D19 (overall satisfaction with HD PT programme)

Although the correlation between D19 (overall satisfaction with HD PT programme) and the objectscores on the first dimension are considerably higher than reported by Shiloh at al (1990), the second dimension is considerably lower and even reciprocal. This, however, is inherent to the restriction of a normal Principal Component analysis and CAT-PCA models: dimensions are supposed to be unrelated and the more successful the fit the more explicit the construct of the dimension seems to indicate. Thus, in general, most variance will be found in dimension 1. Hence this analysis does neither add to the achieved insights nor attributes more or decreases weight to the construct validity of the scale. The above correlations merely indicate that the overall rating of the programme is rather determined by dimension 1: the quality of the information provided by the counselling staff.

Part 2

This part evaluates the actual satisfaction with the PT programme as measured by the satisfaction scale. As indicated in the introduction, the level of measurement of the rating scores of the scale is ordinal. Assuming higher measurement levels would imply inappropriate attributions of properties to the responses and ignorance of the skew distribution of the rating scores. Rather than representing the responses as averages of rating scores, which violates model assumptions, the medians of the responses per item were calculated. Figure 14 shows the median per item. The figure shows that the majority of the participants considered the programme as 'very satisfactory'. The green line represents the median of item D19 'the overall satisfaction with the testing program'. Furthermore, the majority of the respondents were satisfied with most of the aspects of the programme to which the various items referred. Items D17 'follow-up visits provided a great deal of support' and D7 ' the counselling helped you to cope better with your problems' were considered as 'very unsatisfactory' (1/5) and 'unsatisfactory' (2/5) respectively. Items D9 'the clinic environment' and D11 'the manner in which the test results were presented to you' were considered as fairly neutral (3/5) and (3.5/5) respectively. It is interesting to note that 50% (6/12) of those individuals who tested positive rated D11 'the manner in which the test results were presented to you' as 'very satisfactory' confirming the findings of Green and Murton (1996) that it is possible to deliver negative news in a satisfactory manner.

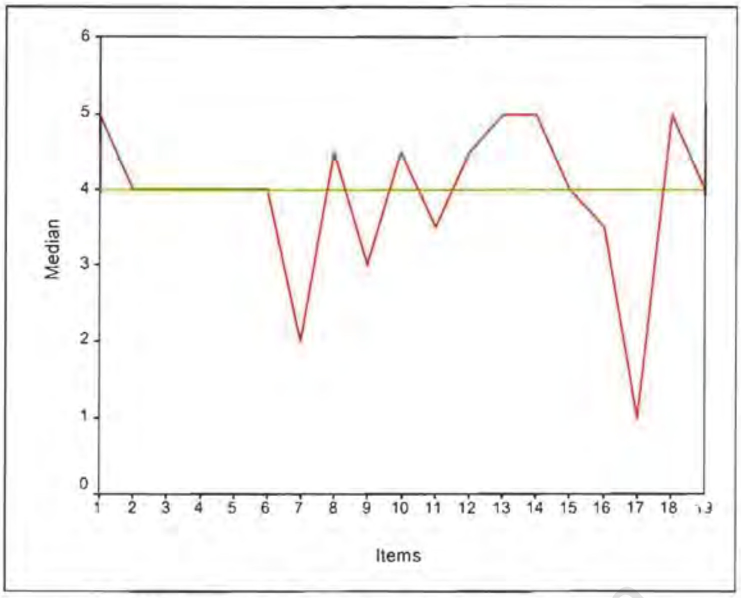

Figure 14: Median of the rating scores per item

The median and mode statistics and the 75th percentile of the nineteen items of the satisfaction scale are presented in Table 32.

	D1	D2	D3	D4	D5	D6	D7	D8	D9	D10
N	26	26	26	26	26	26	26	26	26	26
Median	5.00	4.00	4.00	4.00	4.00	4.00	2.00	4.50	3.00	4.50
Mode	5	4	4*	4	5	5	1	5	1	5
75th Percentile	5.00	5.00	5.00	5.00	5.00	5.00	3.00	5.00	4.00	5.00

	D11	D12	D13	D14	D15	D16	D17	D18	D19
N	26	26	26	26	26	26	26	26	26
Median	3.50	4.50	5.00	5.00	4.00	3.50	1.00	5.00	4.00
Mode	4	5	5	5	4	3	1	5	3*
75th Percentile	4.25	5.00	5.00	5.00	4.25	4.00	1.00	5.00	4.00

* Bimodal (smallest value is shown)

Table 32: Non parametric statistics per item (n=27)

When participants rated an item as less than 'satisfactory' (3/5) the researcher would pursue the content with in-depth questioning to investigate their personal reason for scoring the particular aspect of the PT programme process. A random sample of their verbatim comments for items D7, D9, D11 and D17, which were rated as ‘very unsatisfactory’ (1/5), ‘unsatisfactory’ (2/5) or only just ‘satisfactory’ (3/5), are provided in Table 33.

Item	Verbatim quotes by participants
D7	**The counselling helped you to cope better with your problems** *"There was not enough of it (psychological counselling). It had only started to scratch the surface. I actually needed a great deal more as I had suffered so much as a child and young person".* *"The biggest problem for me was my insurance and legal problems about my minor child and what would happen to him if I tested positive. Nobody could answer my questions and advised me to go to an insurance broker. Well, this would immediately have alerted them to the fact that I had some genetic condition so I could not risk this and have just done nothing about it."* *"No, the counselling that I received was very good, but so much was missing. My brother went for testing overseas and had a session with a legal consultant before anything else so that he knew exactly where he stood with this job and insurance".* *"Nobody ever asked me what it was like to live in a family where the mother is affected and the children have to see her getting worse and worse and also help to look after her. I think I needed several sessions before the test to deal with those past problems because I'm not sure how ready I actually was to go through the test with all those problems that had not been sorted out. Luckily my test result was negative otherwise I don't know what I would have done".* *"I feel that there was too much time spent on repetition of the genetics and not enough time on the psychological aspects of my past. I knew all about the genetics and the disease from the internet, but needed a lot more psychological counselling dealing with my past family aspects before having the test done".*
D9	**The clinic environment** *"It was very distressing for me to go back to the same clinic I used to take my mother when she needed appointments. It brought back very painful childhood memories".* *"I did not like to see the patients in the waiting room. It really upset me and made me very anxious about what I was letting myself in for. I thought it might be better to live and not know about your future than to see what you were actually in for. If I had had to go back to the same clinic for the rest of the sessions I would never have gone through with it".* *"It was a shocking experience".* *"I was very upset that first morning seeing the state which the patients had reached and how difficult it was for their family who were with them".* *"It made me very depressed to see the patients in the clinic. I wondered if the staff didn't realise what a painful experience it is for people asking for the test to be done and if they couldn't have thought about reducing the stress a bit for us".* *"It was a traumatic experience for me. I wouldn't advise anybody to go there for a test, but rather to go to a private clinic where you can just give your blood and they send you the results. Then you don't have to get so shocked".*
D11	**The manner in which the test results were presented to you** *"It was given very clinically and professionally, but with little personal interest. I don't know how I would have coped with a positive result. I was simply given the result and that was the end of that. There was no further comment or contact".* *"It was not very good. I felt completely abandoned. The result was read out from the envelope and I was asked what I was going to do ..... and that was the end of that. I was completely shocked as I didn't expect a positive result and really needed counselling immediately .... but I just had to get up and go. My wife was in the same state as I was and it was very difficult to try and support each other. We should have had some support by the staff who had been dealing with us all the time. They didn't seem interested in that part of one's life. Only up to the time of the test and then get on with it on your own."*

	"Very cold and impersonal and disinterested. It was just something they had to do and they got over with it as quickly as possible. I needed some time to think about it on my own with my partner as we were so shocked. But then I really needed someone of the team to talk to about it, but they were not available for that sort of stuff". *"I was absolutely numb with shock after the result had been read out. They asked me what I thought about it, but there was no way I could talk about it then. I was too busy holding myself together for my husband's sake. And then we just had to get up and leave. We felt as though we had been dumped because of the positive result".* *"It was a great disappointment just being told that I was negative and that was the end. It was just as much of a shock as it would have been with a positive result as I had always thought I would test positively. This part of the counselling really needs a lot more attention about the person's feelings and not to just leave them in the lurch".* *"They just discarded me and I felt my husband and I were on our own. They did talk about what we might feel like during the counselling, but nobody can prepare you for the reality of the situation. And then to be left to cope on your own was awful".*
D17	**Follow-up clinics/visits provided you with a lot of support** *"There were no follow-up sessions. This was a big omission in the programme. I needed a lot of support afterwards and it would have been better from the team who knew what I had just gone through instead of having to go to a psychiatrist who I had never seen before".* *"My wife and I desperately needed follow-up counselling which was just not available. It only occurred to me afterwards that the genetic programme was actually missing the most important part .... to help us to get on with our lives and to get used to the idea that I was going to get this terrible disease at some stage .... and how to deal with the children and what to tell them at this stage and when to tell then that they are also at-risk of getting the disease".* *"There were no suggestions of further visits and I had no further contact with them until they asked me to be part of the research project. Until the result was given the counselling sessions were very good, but as far as I am concerned the most important part was completely missing. What I needed most of all were those and I had to go to my doctor and he referred me to a psychologist. Otherwise I don't know how we would have coped".* *"There was no mention of any follow-up sessions. I felt extremely let down and was very depressed once I got over the shock even though I had a negative result and should have been feeling on top of the world. It was almost worse than getting a positive result and I just didn't seem to be able to pull myself out of it until my doctor gave me some medication. I'm sure a bit of counselling afterwards would have avoided all the trauma I went through".* *"There was absolutely no support provided after the results. I had a telephone call from one of the sisters to ask me how everything was going, but there were relatives in the room and I really couldn't say any thing more than I was coping OK. What I actually needed was to know that there was another appointment that I had to attend and that I would be able to deal with my queries then and have somebody who knows about the horrors of the disease just sit and talk to*

Table 33: Participants' quotes on 'unsatisfactory' components of PT programme

In conclusion of the analysis of the satisfaction scale, in exploring the underlying constructs that determined the patterns in the responses, the CAT-PCA revealed two dimensions explaining 67% of the total variance within the responses of 26 respondents to 16 items about the satisfaction with the PT program. The first dimension was identified as 'quality of information' referring to the quality of the information provided by the counselling staff. The second dimension was identified as 'empathy' referring to the amount of empathy the counselling staff displays. The program, in general, appears to be experienced as 'very satisfactory' (4/5). Overall, patients evaluated their satisfaction by weighting, on one hand, the extent and manner the staff provided insight into HD, its testing procedure and follow up and on the other hand, the amount of empathy they experienced from the staff and the hospital environment.

Participants' recommendations for improvements to the programme

Item	Recommendations	N=27	%
D7	More psychological counselling sessions during testing programme	19	70
	Legal consultant to be included in team	18	67
D9	Change of venue	18	67
	No contact with patients	15	56
D11	Test result – short recess – immediate counselling session	9	33
D17	Prearranged routine follow-up sessions 1 week and 1-monthly for one year	21	78
	Post-test counselling for partner/spouse/children	18	67

Table 34: Participants' recommendations for improvements to the predictive testing programme

After rating the satisfaction scale participants were requested to recommend changes they felt would improve the programme. These responses were unsolicited and simply the outcomes to the open-ended question, "What suggestions can you make to improve the present PT programme?" The responses were grouped according to items D7, D9, D11 and D17 which had not received a rating of at least 'very satisfactory' (4/5) or 'highly satisfactory' (5/5) and are listed in Table 34.

With regards to item D7 70% (19/27) considered that the programme should include more psychological counselling sessions prior to the testing. Those participants whose mothers had been affected as they were growing up felt that they had not come to terms with the loss of a person who was supposed to protect and care for them; the trauma experienced by their temper outbursts and marital discord between the parents; and the loss of childhood due to caring responsibilities often not appropriate for their age. Sixty-seven percent (18/27) felt that a legal consultant was an essential component of the team to advise them on insurance and employment implications for themselves as well as their children.

The venue (D9) was problematical to the majority of participants (67%, 18/27) who suggested that it be changed from the hospital environment and 56% (15/27) who requested that they have no contact with HD patients during the PT programme. Those who had not had prior contact with affected family members found the experience of sharing the waiting room with HD patients to be traumatic as they could imagine themselves going down the same route should they test gene-positive. Others who had lived in families with an affected person found the experience traumatic as it evoked painful memories of close relatives who were now deceased.

Thirty-three percent (9/27) requested that at the time of the test disclosure (D11) the participant and support partner be given a short recess of approximately 30 minutes alone followed immediately by a counselling session. Participants made it clear that the manner in which the counsellor presented the test result was not unsatisfactory, it was the session itself which was inadequate.

Recommendations for item D17 which was rated as 'very unsatisfactory' by 96.2% (26/27), were for routine regular follow-up sessions for themselves (78%, 21/27) and for their partner/spouse/children (67%, 18/27). They suggested that had they been offered this service as part of the PT programme they would have had more psychological support to cope with the implications of their test result and might have experienced less depression. The importance of follow-up counselling sessions are endorsed by Tassicker et al (2006), Codori et al (2004), Richards and Williams (2004), Almqvist et al (2003), Decruyenaere et al (2003), SuttonBrown and

Suchowersky (2003), Evers-Kiebooms et al (2002), and Hayden and Kremer (2001), and Craufurd and Tyler (1992).

Contacted by member of genetic team

The protocol information with which each participant is provided at the beginning of the programme clearly states that if they require an appointment with a psychologist for follow-up counselling they should contact the genetic nursing sisters who would make the necessary appointments. Although the medical files of the participants indicated that one of the team members had made or had tried to make telephonic contact with 51.8% (14/27) of the participants a week or two after the test disclosure only 25.9% (7/27) remembered this contact and the discussion that took place at the time. Those who perceived that there had not been any contact expressed their disappointment in the programme with regard to this aspect. When the author queried why they had not contacted the genetic nurses themselves the responses given are presented in Table 35.

	Frequency	Percentage
Nothing was specifically arranged	26	96.2
Too depressed to make contact personally	12	44.4
"They were not really concerned about what happened after the result"	14	51.8

Table 35: Reasons for participants not contacting genetic nurses after test disclosure

4.8 EVALUATION OF PRESENT LIFE SITUATION

In spite of the daily concerns experienced by 96.2% (26/27) participants in Table 18 (page 165) they rated their present life situation as presented in Figure 15.

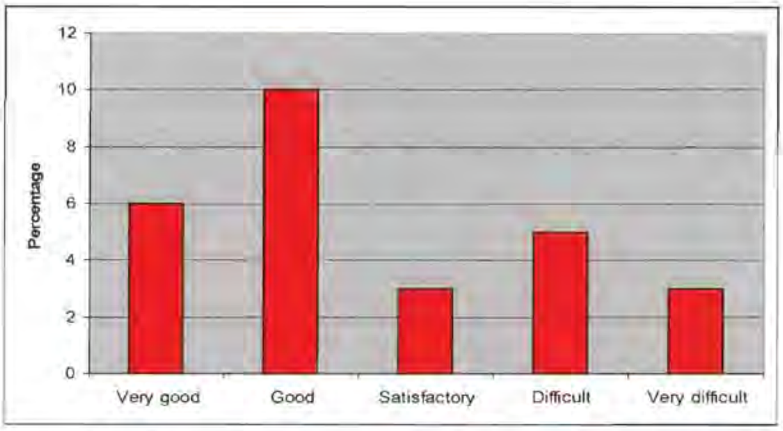

Figure 15: Evaluation of present life situation by participants

All individuals (100%) who had adult children had disclosed their test results to them and had informed them of the PT programme. None of the children have subsequently requested testing. Those who had young children had decided not to tell them, but were adamant that they would inform them once they were more mature. With regards to smoking habits before and after PT there was no change in habits reported by any of the participants. Only 3.7% (1/27) who reported his drinking habits before the test as being 'a few' had become a heavy drinker after his test disclosure of a gene-negative result.

4.9 SUMMARY OF THE PROFILE OF THE PT PARTICIPANTS

Data from this part of the research are generally in accordance with international findings with respect to demographic and socioeconomic data, age at presentation, 'at-risk' status, and reasons for undergoing PT and test results. The uptake of PT was 4.5% which is lower than the 5%-20% found internationally. This may be accounted for by the fact that individuals presenting for PT are generally from higher educational and socio-economic levels and the majority of the individuals with HD in the WC have low levels of education and come from a poor socio-economic background (Figure 16, (page 194), Figure 18 (page 195) and Table 39 (page 197). The ethical principle of justice (Ensenauer et al 2005) being violated does not appear to apply as all individuals in the WC at-risk for HD have equal access to the genetic testing service regardless of their socioeconomic and ethnic backgrounds and the geographical regions in which they live. However, the testing programme is not equally accessible to them as the transport costs for the pre- and post-counselling sessions add up to a substantial amount which the majority of people from the low socio-economic group are not able to afford. The most significant life event which triggered the request for testing was a relative recently diagnosed with HD. All of those who only became aware of their at-risk status as an adult presented for PT within 1 year of the knowledge. Those who had a limited exposure to the disease waited a mean of 11 years and those with a great deal of exposure to an affected relative waited the longest time, 13.3 years.

Disclosure of test results was found to be limited to selected immediate family members and close friends only.

The frequency of major adverse reactions immediately after the test result and frequency of depression 1 year after PT are much higher than other studies although no participants regretted having had the test done. One of the reasons for this increased incidence could be that the programme did not offer a period of counselling after test result disclosure. The research findings of several authors show that counselling is an effective process for assisting individuals to adjust to their status and maintain psychological wellbeing through the provision of emotional and informational support (Decruyenaere et al 2007; Davey et al 2005; Randall et al 2001; Michie et al 1997a) so that individuals perceive that they have control over the consequences such as emotional reactions (Berkenstadt et al 1999). A second reason could be the possibility of the lack of an HD support group in the WC (Figure 33) and thirdly that 11/27 (40.7%) had been tested within the first year of becoming aware of their at-risk status and this is advised against by several authors as the individuals have not had sufficient time to adjust to their at-risk status before undergoing the test (Ensenauer et al 2005; Lea et al 2005; Sarangi et al 2005; Myers 2004; Taylor; Craufurd and Tyler 1992). Fifty-five percent 15/27 of the PT group had been involved in caring for affected relatives whereas only 9/41 (21.6%) of those from the patient group had cared for or assisted in the care of affected relatives. This might explain why these individuals requested PT as they were more familiar with the disease and its physical, emotional, cognitive, and financial implications.

In spite of the high levels of depression 1 year after testing 100% of the cohort did not regret having the test done and would advise other at-risk individuals to undergo the test.

The participants' level of knowledge of the genetics of HD was excellent with an average of 81.3% for recall and understanding. Their overall level of satisfaction with the PT programme was rated as 'very satisfactory' (4/5) and is determined by the 'quality of the information' provided and the amount 'empathy' displayed by the counselling staff. This overall rating was negatively affected by 2/16 items, D7 and D17, which were considered to be 'unsatisfactory' (2/5) and 'very unsatisfactory' (1/5) respectively. Recommendations for easily implementable improvements to these items were made by the participants.

4.10 PROFILE OF THE AFFECTED PATIENTS RECRUITED FROM THE NEUROGENETIC CLINIC

Forty-one patients who were attending the neurogenetic clinic were included in the study. No patients refused the opportunity of participating in the research. All patients received 2 or 3 home visits to complete the interviews and to follow up on their needs. The initial interviews lasted approximately 2 hours and follow-up visits approximately 1 hour. The majority of patients (95.1%, 39/41) lived within a radius of 42kms and only 4.8% (2/41) came from a rural area 95kms away from the clinic. At the time of diagnosis 12.2% (5/41) did not know of a family history of the disease. Almqvist et al (2001) found that 25% of their cohort of newly diagnosed patients did not have a family history of HD.

	Frequency
Self	5 (12.2%)
Partner/spouse	14 (34.2%)
Child/child-in-law	8 (19.6%)
Relative	11 (26.8%)
Parent	3 (7.3%)
Total	41 (100%)

Table 36: Relationship of proxy to patient

As the patients had been symptomatic for several years (Figure 20, page 200) the majority required their carer to be present during the interview (Table 36). In some situations the patients responded themselves and the researcher was able to understand them. Their responses were corrected or confirmed by the proxy with the patients' agreement. When they were dysarthric the proxy responded and the patients nodded in agreement or indicated that they disagreed and the researcher would repeat or rephrase the questions until there was agreement on a response.

4.10.1 SOCIODEMOGRAPHIC DATA

Age, gender, ethnicity and first home language

Age	Gender	Ethnicity		Total
		White	Mixed	
<30	Male	0	1	1
30 - <40	Male	1	8	9
	Female	0	1	1
40 - <50	Male	1	2	3
	Female	1	7	8
50 - <60	Male	2	3	5
	Female	2	6	8
>59	Male	1	1	2
	Female	2	2	4
	Total	**10**	**31**	**41**

Table 37: Patients per age group, gender and ethnicity

The cohort comprised 48.8% (20/41) males and 51.2% (21/41) females of which 24.4% (10/41) were classified as White and 75.6% (31/41) as Mixed Ancestry. The first home language of 36.6% (15/41) was English and 63.4% (26/41) Afrikaans whereas the in the general population of the WC it was

26% and 68.2% respectively (Statistics SA 2001) . The mean age at the first interview was 48.6 years with the mean of females being 52.7 years and that of males 44.9 years (Table 37).

Income

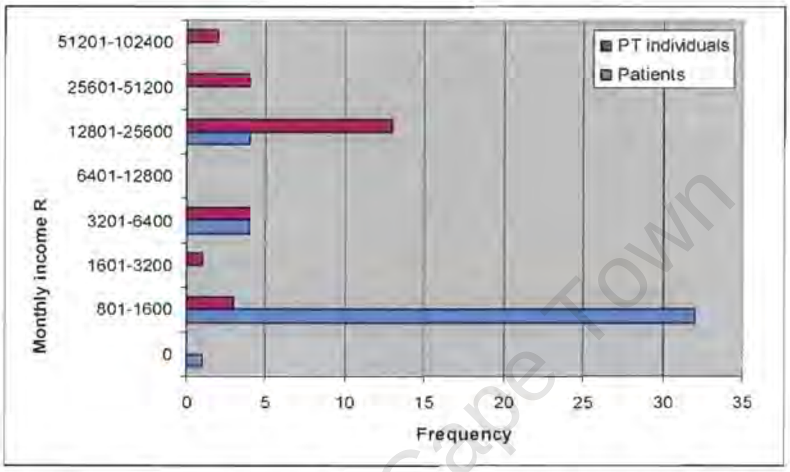

Figure 16: Comparison of monthly income of patients and PT participants

As can be seen in Figure 16 there is a vast discrepancy between the PT group and patient group of the cohort with regards to the monthly income. The PT group earned a mean of R21,422 per month and the patient group R2,825 per month. Seventy-eight percent (32/41) of the patients received an income from the DG only which was R820 per month. The DG supported a mean of 3 persons per patient recipient. Seven percent (2/41) were from the White population group and 93% (27/41) from that of the Mixed Ancestry group.

Figure 17 shows the frequency of social grants to which the families were entitled and those to which they were entitled, but were not receiving because of lack of knowledge of their rights to financial assistance.

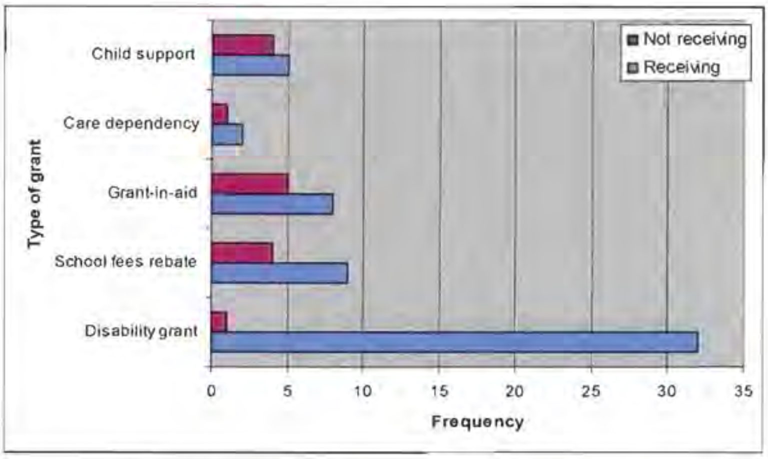

Figure 17: Social grants of patients and their family members

The 65% (27/41) of patients who had never been employed or had been employed on a casual basis only considered that HD had not been a financial burden on the family as they were better off with a regular monthly income of R820 rather than no income at all or only periodic income. However, those (34.2%, 14/41) who had been employed prior to the onset of symptoms considered their disability to be a major financial burden on the family.

Level of education

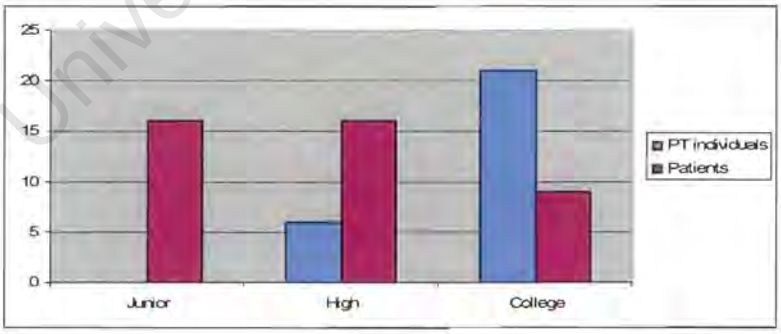

Figure 18: Comparison of the levels of education between the two groups

Figure 18 depicts a comparison of the level of education of the two study groups. The majority from the PT group had a tertiary education with a minority having only a high school qualification; whereas the majority from the patient group had only a junior or high school level of education. The mean years of education of the patients group was 9.4 years compared to that of 14.7 years of the PT group.

The reasons for not continuing with school or tertiary education are presented in Table 38. The categories are not mutually exclusive. For example, patients discontinued school because of 'lack of money' and to 'help mother with the domestic chores' or 'lack of interest' and to 'help support family'. The 11.1% (3/27) PT participants who indicated 'other' cited choice of wrong career, pregnancy and death of a parent as the reason for leaving school or college. Sixty-seven percent (18/27) of the PT group had completed their tertiary education as opposed to 21.6% (9/41) of the patient group and only 11.1% (3/27) of the PT group had to leave school because of lack of money whereas this was the main reason for 36.6% (15/41) of the patient group doing so.

	PT group (n=27)	Patients group (n=41)
Lack of money	3 (11.1%)	15 (36.6%)
Lack of interest	4 (14.8	6 (14.6%)
No accessible school	0 (0%)	1 (2,4%)
To help mother with domestic chores	0 (0%)	3 (7,3%)
To help support family	2 (7.4%)	3 (7,3%)
To care for affected parent	0 (0%)	4 (9.8%)
Joined a gang	0 (0%)	5 (12.2%)
Completed diploma/degree	18 (66.6%)	9 (21.6%)
Other	3 (33.3%)	0 (0%)

Table 38: Reasons for not completing or continuing with education

Occupation

The majority of PT participants were employed in managerial, professional or semi-professional categories as defined by the Census (2001) and the majority of the patient group had been employed in elementary occupations before they became symptomatic (Table 39) (Statistics SA, 2001). The percentage of patients in elementary occupations was significantly higher than that of the general population in the WC (53,6% vs. 29%) indicating that their families were of the lowest income families in low-income areas. Twelve percent 12.2% (5/41) of the patient group had never been employed in either the formal or informal sector, but had earned their living by unlawful means such as robbery, armed robbery, illicit sales of alcohol and banned substances.

Category	PT group (n=27)	Patient group (n=41)	Western Cape population
Managerial, professional, semi-professional	77.8% (21/27)	21.5% (9/41)	22%
Clerical, sales, service	18.5% (5/27)	4.8% (2/41)	22%
Skilled agricultural, craft, operators	3.7% (1/27)	7.3% (3/41)	20%
Elementary occupations	0% (0/27)	53.6% (22/41)	29%
Undetermined	0% (0/27)	12.2% (5/41)	7%

Table 39: Comparison of occupations of the two groups compared to the general population in the Western Cape

Marital status and number of children

At the time of the first interview 12.2% (5/41) of the patients were single, 14.6% (6/41) were widowed, 43.9% (18/41) married and 29.3% (12/41) divorced. The number of children and grandchildren is presented in Table 40. The mean number of children and grandchildren per patient was 2.7 and 2.3 respectively compared to that of the PT group which was 1.4 and 0.5 respectively (Table 4, page 148). However, this does not give a true reflection of their families in general. It was not unusual for the patient to have had six or seven siblings and each of those siblings to have had five or six children and the children to have had four to six children themselves. None of them had undergone PT and all were at 50% risk.

Number of children and grandchildren													
Children	**0**	**1**	**2**	**3**	**4**	**5**	**6**	**7**	**9**	**10**	**12**	**14**	**Total**
Single	5	1	0	0	0	0	0	0	0	0	0	0	1
Married	0	3	5	4	4	1	0	0	0	0	0	0	46
Divorced	0	1	3	4	1	1	1	0	0	0	0	0	34
Widowed	0	0	2	1	1	1	1	1	0	0	0	0	29
Total	0	5	20	27	24	15	12	7	0	0	0	0	**110**
Grandchildren	25	4	1	1	1	1	1	1	3	1	1	1	**94**

Table 40: Marital status, number of children and grandchildren per patient (n=41)

Only 2.4% (1/41) of the patients were unable to identify an affected parent who had been diagnosed genetically and/or clinically as having HD as their paternity was unknown. The remainder (97.6%, 40/41) had a prior genetic risk of 50% (Table 41). Seventy-eight percent (32/41) had either been raised in or had cared for an affected family member so were well-acquainted with the symptoms.

	Participant		
Affected parent	**Male**	**Female**	**Total**
Paternal	7 (17.1%)	10 (24.4%)	17 (41.5%)
Maternal	12 (29.3%)	10 (24.4%)	23 (56.1%)
Unknown	1 (2.4%)	1 (2.4%)	1 (2.4%)
Total	**20 (48.8%)**	**21 (51.2%)**	**41 (100%)**

Table 41: Maternal and paternal family history

4.10.2 DISABILITY PROFILE

Age at first symptoms of HD

As found in previous studies (Quarrell et al 2007; Quarrell and Cook 2004; Hankey and Wardlaw 2002; Fletcher 2001), the age at which the patients first started with symptoms was difficult for them to identify due to the insidious nature of the disease. The majority could not pinpoint a specific time that they noticed any cognitive, behavioural or motor changes. However, in retrospect they could often determine the number of years before being diagnosed that there had been subtle signs and symptoms developing. Often the patients had not noticed any changes, but their relatives had, although not all of them had associated the changes with the onset of the disease at the time they first observed them. The ages at which these occurred are presented in Figure 19. The average AOO was 39.6 years which compares with the findings of international studies (Tassicker et al 2006; Hayden and Kremer 2001).

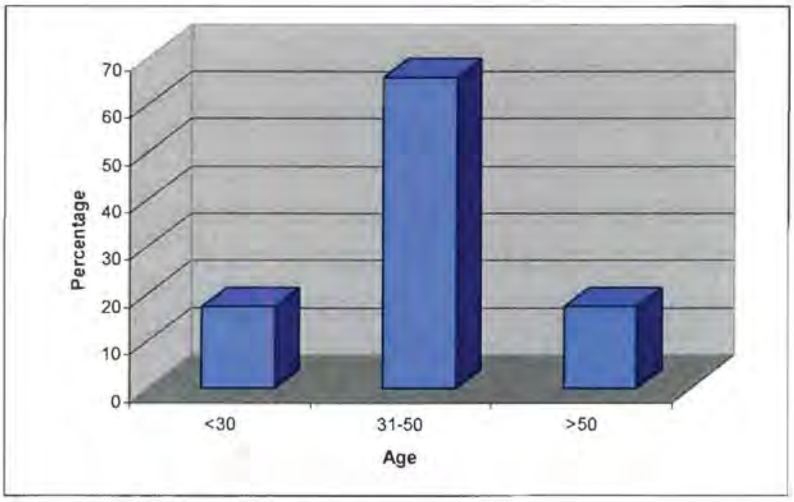

Figure 19: Age at first symptoms of HD

Nature of the first symptoms

Category	Symptoms	Frequency	Percentage
Motor	Restlessness	18	43.9
	Chorea	15	36.6
	Involuntary movements	7	17.1
	Weakness	5	12.2
	Dysarthria	16	39.2
	Unable to balance	12	29.3
	Falling	9	21.6
	Gait	14	34.2
	Total	**96**	**Mean = 29.9%**
Cognitive	Concentration	10	24.4
	Forgetfulness	23	56.1
	Ability to think clearly	18	43.9
	Lack of insight	9	21.6
	Total	**60**	**Mean = 18.7%**
Affective	Irritability	28	68.4
	Anxiety	23	56.1
	Disinhibition	15	36.6
	Depression	27	65.9
	Aggression	29	70.7
	Anger	37	90.2
	Compulsive behaviour	6	14.6
	Total	**165**	**Mean = 51.4%**

Table 42: Nature of first symptoms

The nature of the first symptoms that were identified by the patient or family members are presented in Table 42. The categories are not mutually exclusive as many developed a variety of symptoms at onset. In this cohort of patients 70.1% presented with cognitive and affective disturbances which confirms the findings of (Askin-Edgar et al 2004; Quinn 2003, Squitieri et al

2003) where up to 79% of their study sample presented with behavioural changes as early as 20 years before the onset of chorea (Paulsen and Nehl 2004; Quarrell and Cook 2004; Shannon 2004). This refutes the findings of Harper (2004), Jacobs et al (2003), Creighton et al (2003) and Li et al (2003) who found that individuals presented with motor disturbances followed many years later by psychiatric disorders (Leroi et al 2002; Folstein 1989).

The number of years of being symptomatic is shown in Figure 20. Most authors have found that the average time from onset to death is 17 to 20 years (Myers 2004; Taylor 2004; Almqvist et al 2003; Quinn 2003; Brouwer-DudokdeWit et al 2002; Evers-Kiebooms et al 2002; Donaghy 2001; Harper 1996) so it is interesting to note the sudden decrease in the percentage of patients in the 10 to 14 years group in the WC cohort. However, it may be accounted for partly by the big difference in general life expectancy figures of South Africa and those of the countries from which the majority of research on HD occurs. Figures given by the World Factbook (2006) give the life expectancy rates as Australia 80.5 years, Canada 80.22 years, UK 78.54 years, USA 77.5 years and South Africa 50.7 years (Knight 2006).

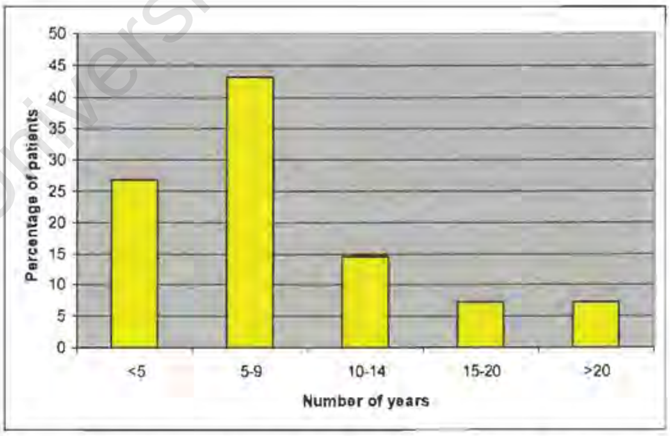

Figure 20: Number of years since onset of symptoms

Level of functional ability

Table 43 represents the frequency of the ratings of the items of the Stanford Health Assessment Questionnaire (HAQ) regarding the level of functional ability of the patients.

	No difficulty	Some difficulty	Much difficulty	Unable to do
Dressing yourself	8	18	5	10
Shampoo your hair	9	14	6	12
Stand up from chair	10	17	4	10
Get in and out of bed	11	17	3	10
Cut your own meat	5	14	4	18
Lift a glass to your mouth	6	13	10	12
Open a new milk carton	6	14	4	17
Walk outdoors on flat ground	6	19	3	13
Climb up and down five stairs	7	18	3	13
Wash and dry your body	10	11	8	12
Bath in a proper bath/shower	9	12	6	14
Get on and off the toilet	8	16	5	12
Reach for object above your head	6	12	3	20
Pick up clothing from the floor	11	15	3	12
Run errands and shop	6	13	3	19
Get in and out of a car	7	18	5	11
Do housework or gardening	6	14	4	17

Table 43: Frequency of rating items of activities of daily living (n=41)

The items were rescaled to one additive scale (Table 44) representing the overall level of difficulty that the participants had with their functional activities. Thirty-four percent (14/41) of the participants had 'some' level of difficulty with function and 46% (19/41) experienced 'much' difficulty or 'total' inability with performing their basic activities of daily living.

	Frequency	Percentage	Cumulative Percentage
No problem	140	20	20
Some	246	35	55
Much	79	11	66
Unable to do	231	34	100
Total	**696**	**100**	

Table 44: The ability of the patients to perform functional activities

Certain items of the HAQ were selected to demonstrate the percentage of disabled who had 'much difficulty' or were 'unable to do' the basic activities of daily living to show more clearly how disabled they were (Figure 21).

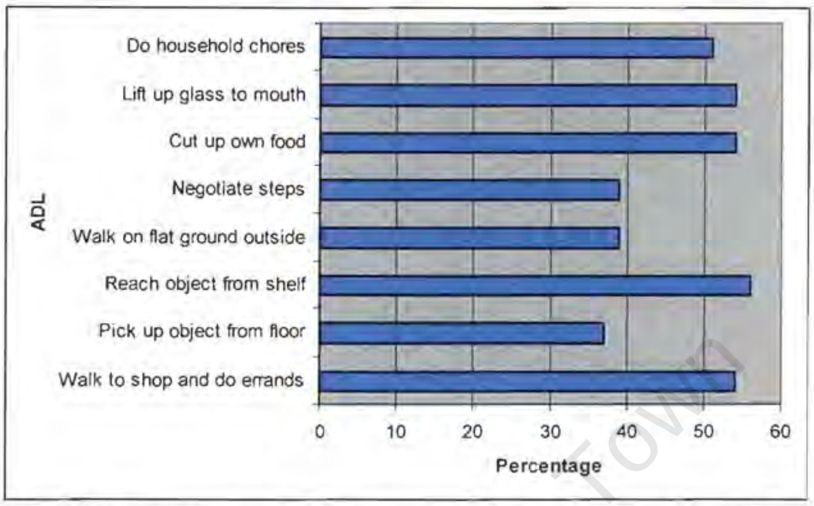

Figure 21: Percentage of patients unable to perform certain activities of daily living

The Functional Disability Index of the HAQ was determined by adding the highest score in each of the seven components to form a total (range 0 – 21); this was divided by seven to provide a 0 – 3 continuous score. If either devices and/or assistance was required for a component the score was determined as 2 unless the score on any of the other component question was already 3.

Siegert et al (1984) suggested the following interpretation of the overall scores (Table 45).

Score	Description of functional disability
0.0 - 0.5	Completely self-sufficient
0.5 - 1.25	Reasonably self-sufficient; experiences some minor and even major difficulties in activities of daily living
1.25 - 2.0	Self-sufficient, but has many major problems with activities of daily living
2.0 – 3.0	Severely handicapped

Table 45: Categorisation of the different levels of disability

Figure 22 shows the number of patients who fitted into these different categories. As can be seen 4.8% (2/41) of the patients had major problems with activities of daily living (1.26 – 2.0) and 48.8% (20/41) were severely handicapped (2.0 – 3.0). This high level of disability was to be expected as the mean number of years since onset was 9.2 years and 68.4% (28/41) of the patients received permanent DGs. The functional capacity of the majority of patients was impaired by their cognitive decline and psychiatric symptoms rather than movement dysfunction, confirming the findings of SuttonBrown and Suchowersky (2003); Heindel et al (1991) and Knopman and Nissan (1991).

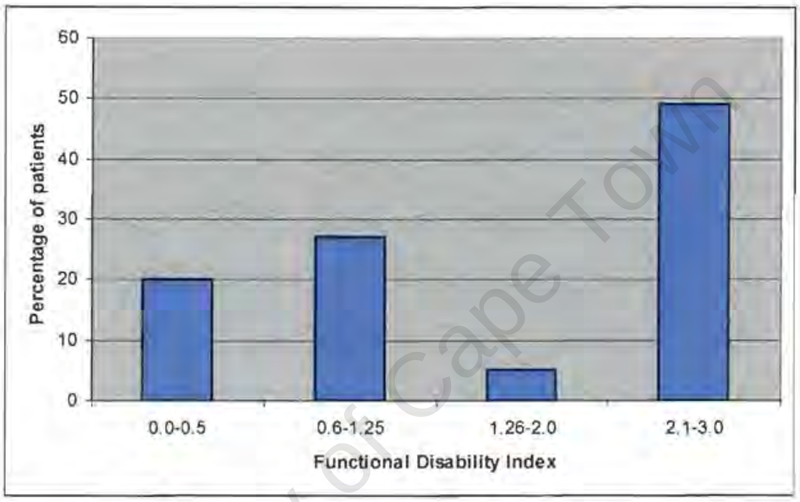

Figure 22: Functional disability scores of patients

Hospital anxiety and depression scale

Score	Description	Anxiety	Depression
11 – 21	Presence of mood disorder	25 (60.1%)	12 (29.3%)
8 – 10	Suggestive	0 (0%)	2 (4.8%)
0 - 7	Normal range	16 (39.9%)	27 (65.9%)
	Total	**41(100%)**	**41(100%)**

Table 46: Frequency (percentage) of anxiety and depression scores

As the patients were unable to complete the forms because of their impairments and many of the carers were functionally illiterate, the researcher read the items to them aloud which is acceptable practice under these circumstances according to Snaith (2003). Their depression and anxiety scores are presented in Table 46 indicating the presence of the mood disorders. According to SuttonBrown and Suchowersky (2003), Hankey and Wardlaw (2002) and Hayden and Kremer (2001) depression occurs in 9 - 70% of patients and this eventually can lead to anxiety.

Other medical conditions affecting patients

Conditions	Frequency	Percentage
Incontinence	10	24.4
Pressure sores	3	7.3
Diabetes	4	9.8
Pulmonary TB	15	36
Arthritis	2	4.8
Hypertension	12	29.3
Pain	9	14.6
Cancer	1	2.4

Table 47: Additional medical conditions affecting patients

As the concept of pain is subjective and can fluctuate over a period of time depending on the medical condition of the individual, 'pain over the past week' was considered to be a representative measure. In 14.6% (6/41) cases the pain was generalised and flitted around the body from day to day, 4.8% (2/41) had specific knee joint pain related to osteoarthritis and 2.4% (1/41) had acute back ache from metastasis from breast cancer.

In addition to those 36.6% (15/41) undergoing treatment for TB themselves, 9.8% (4/41) were sharing a room and/or bed with a family member currently undergoing treatment for TB. According to government sources the countrywide prevalence of TB is 511 per 100 000 population with an annual mortality incidence of 71 per 100 000 population is one of the highest in the world (WHO 2007; http://www.hst.org.za/healthstats/163/data). As the majority of patients were interviewed in the presence of their caregivers, due to the confidential nature of the disease, it was not appropriate to question them on their HIV/AIDS status. However, by the very nature of its prevalence of 15.7% in the WC (Dorrington et al 2006; http://www.info.gov.za/issues/hiv/framework.pdf), it is to be expected that there would have been 6 or 7 of the patients would be HIV-positive or affected with AIDS at the time of the interviews.

Wheelchairs and assistive devices

There was only one patient who had access to a wheelchair. This could only serve as a form of seating inside the house because of space limitation and with great difficulty outside due to the rough terrain and the fact that the wheelchair was broken (Photograph 1). However, it was most beneficial for the patient and her carer when she required being transported to the family planning clinic. The wheelchair had been donated by a neighbour whose spouse had died of a stroke.

Photograph 1: Broken wheelchair used for transport to clinic

Ten percent (4/41) had been issued with a walking stick or walking frame by the local clinic and had to be removed from them by the researcher for safety reasons as the patients were using them as weapons when they became aggressive and had temper outbursts. They were not appropriate walking aids as the walking frame could not be used inside because of the small size of the house (Photograph 2) and the patients who possessed walking sticks could not use them as their chorea and balance were too severely affected and the aids were not appropriate at their stage of symptoms. In all cases the walking aids had been issued to prevent falling, but served no purpose as the patients were falling due to motor impersistance rather than lack of balance or postural instability.

Photograph 2: inappropriate walking aid supplied

However, taking into consideration their housing situation, outside environment and level of function, the researcher considered that the following assistive devices would allow the patients increased independence and alleviate the burden of their carer for their basic activities of daily living (Figure 23).

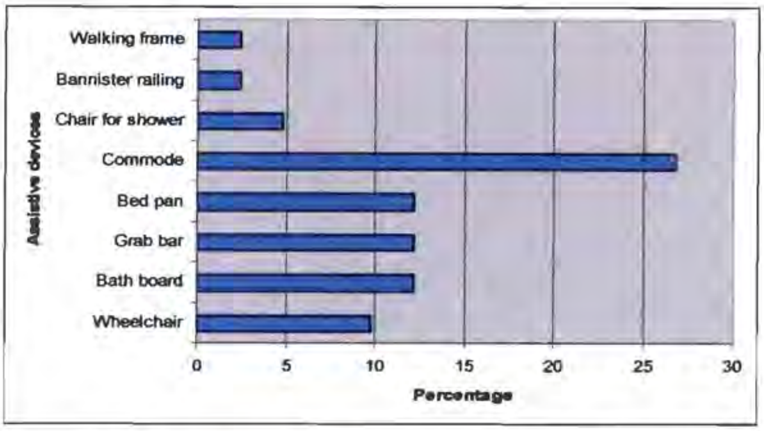

Figure 23: Percentage of patients requiring assistive devices required for increased independence of ADL

Not all the patients at the same functional level required the same assistive devices as their home environments varied to such an extent. One female required a walking frame to be able to walk independently as her house was large enough to accommodate it and she had adequate paths, sidewalks and road surfaces in the immediate environment. Another female required a bannister rail to allow her to ascend and descend the stairs from her bedroom which was downstairs to the bathroom upstairs. Without this she required to be carried up by her carer as the staircase was too narrow to accommodate the patient being assisted by her carer at her side. Two patients had access to showers, but could not use them independently as they were unable to balance in the shower. Shower chairs would enable them to be assisted to the shower cubicle and then to do their own ablutions. Eleven patients lived in backyard Wendy houses (colloquial term for self-constructed additional accommodation) with no bathroom facilities and urgently required commodes as they had no access to bathroom facilities at night as the residents in the main house had to lock their doors for security reasons. Those patients requiring bedpans were bedridden in the late stages of HD and had bathrooms upstairs only. Five patients requiring grab bars would have been able to shower and use the toilet independently with grab bars, but had to struggle to use these facilities as they required assistance from their carers and there was insufficient space in the rooms to provide this from the side. Simple bath boards would have made a great deal of difference to the quality of life of five patients who could not use a bath was unable to lift them in and out of the bath. Four patients were immobile and housebound and required a wheelchair to be able to go outside, to attend clinics or to visit neighbours. It is essential to stress that these were the requirements at the time of the interview. However, with the progress of the disease over a few months together with the scores of the Functional Disability Index it can be predicted that further equipment needs would increase significantly in the short-term.

4.10.3 GENERAL SOCIAL PROBLEMS EXPERIENCED

The extent to which the different social problems affected the patients is presented in Table 48.

Item	No problem 1	Mild problem 2	Moderate problem 3	Severe problem 4	Complete problem 5
G1 Lack of money	6 (15%)	3 (7%)	10 (24%)	2 (5%)	20 (49%)
G2 Transport	15 (37%)	0 (0%)	4 (10%)	1 (2%)	21 (51%)
G3 Housing	22 (54%)	1 (2%)	2 (5%)	1 (2%)	15 (37%)
G4 Safety	27 (66%)	1 (2%)	3 (8%)	0 (0%)	10 (25%)
G5 Unemployment	25 (61%)	0 (0%)	1 (2%)	2 (5%)	13 (32%)
G6 Isolation and loneliness	17 (41%)	5 (12%)	4 (10%)	2 (5%)	13 (32%)
G7 Privacy	30 (73%)	0 (0%)	2 (5%)	0 (0%)	9 (22%)
G8 Health services	21 (51%)	1 (2%)	4 (10%)	1 (2%)	14 (35%)
G9 Social services	20 (49%)	1 (2%)	3 (8%)	4 (10%)	13 (31%)
G10 Domestic assistance	34 (83%)	1 (2%)	0 (0%)	1 (2%)	5 (13%)
G11Attitude of the community	33 (81%)	2 (5%)	1 (2%)	1 (2%)	4 (10%)
G13 Abuse	14 (34%)	0 (0%)	10 (24%)	3 (8%)	14 (34%)
G14 Disability	3 (8%)	1 (2%)	9 (22%)	3 (8%)	25 (60%)

Table 48: General social problems experienced by patients (n=41)

Sixty percent or more of the patients experienced 'no problems' with 'safety', 'unemployment' and 'privacy' (Table 48). Eighty percent or more experienced 'no problem' with 'domestic assistance' and 'attitude of the community'. These categories had far lower frequencies than 'lack of money', 'transport', 'housing', 'social services', 'health services' and 'disability'. Apart from 'disability' these findings confirm that of the research done by Futter (2004) in similar low socio-economic suburbs of the WC.

Safety

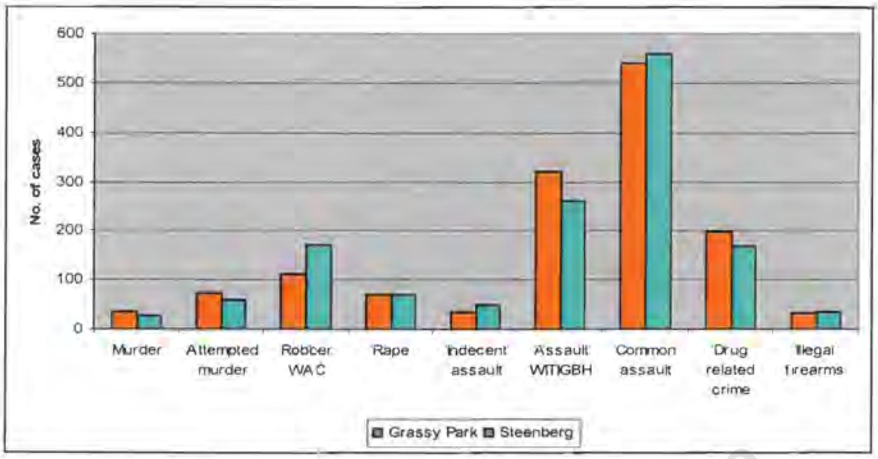

Figure 24: Crimes involving bodily harm in Grassy Park and Steenberg police precincts

Contrary to expectations more than 60% of the patients rated 'safety' as 'no problem'. This is in spite of official crime statistics showing that the WC has by far the worst overall crime problem in the country with the nation's highest rate of murder: 85 murders per 100,000 citizens with the majority of non-natural deaths due to murder (MRC 2001). The WC has the highest gang-related violence in South Africa and Lavender Hill and Hanover Park (Figure 24), where the majority of patients lived, have the highest incidence in the WC (Goss 2002). Murder rates in the mixed ancestry population are the highest because a higher proportion of this ethnic group is both urban and poor when compared to other ethnic groups (Leggett 2004a). This is probably due to the highest rate of illegal possession of firearms and ammunition. It also has the country's highest levels of indecent assault, robbery and property crime which are likely due to alcohol and drug abuse and gangsterism which are the highest in the country. It appears that addicts steal to pay for their habits impacting on property crime (Leggett 2004b).

Although the majority of patients came from low-income suburbs there are areas within each suburb itself where there is greater poverty than others. People living in the higher income areas with larger houses with no backyard Wendy houses believed that their areas had no 'safety' problems as there were no gangsters living in the immediate vicinity. Their windows and doors had burglar bars which prevented theft and they stated that they had no problems with the gangsters in the streets. The patients living in the poorer areas where there were smaller houses and additional backyard Wendy houses reported that they felt safe as they were well-acquainted with the gangsters. They had grown up in the same neighbourhood with each other and they believed that they would not harm the residents. Some of the druglords even provided the families with money to pay their food bills. Their only fear for personal safety was the cross-fire during the gang wars. Their biggest problem was not being able to leave their houses unattended as thieves stole their movable property although this theft was not by the gangsters. However, with the overcrowding, unemployment and nature of HD there were people in the houses at all times so that they did not find safety a significant social problem. Patients from this cohort all lived in the poorer areas of the suburbs. The outcome data of >60% of the patients rating 'safety' as 'no problem' is based on the average of the group responses, but does not adequately represent those patients who do find 'safety' an issues and the extent of the difficulties experienced as a result. Of the patients interviewed 7/41 (17.1%) reported a father, son or brother having been shot or stabbed to death by gangsters or as a result of violence in the area.

Unemployment

'Unemployment' was not rated as a problem of any significance as 78% (32/41) received a DG and, although minimal, it was some form of income compared to those individuals in the community who were unemployed and had no income at all (39.7%) and those who were employed in casual elementary occupations (36.1%). Driving through the various low-income suburbs in which the majority of patients lived and seeing the number of young male adults sitting on the pavements and chatting on the street corners gave the impression that the unemployment rate was significantly higher than the figures above provided by the census (Statistics SA 2001).

Privacy

It is difficult to comprehend that the patients did not find 'privacy' a substantial problem as 65.9% (27/41) of them who were single or married had to share a bedroom with at least one other person (Table 49). In many cases, where the patient or married couple did not have to share a bedroom, the sitting room and/or the kitchen served as bedrooms at night.

Number of patients	14	2	3	4	5	6	7
Number of additional people in bedroom	0	6	5	4	3	2	1

Table 49: Number of additional people sharing bedrooms with patients and their spouses/partners (n=41)

Domestic assistance

'Domestic assistance' did not appear to be a problem as there were many family members sharing the houses and household chores were distributed among those who stayed at home during the day.

Attitude of the community

Although 83.0% (34/41) of the patients reported that they had 'no problem' with the attitude of the community towards them 9.8% (4/41) complained bitterly about the fact that they had been repeatedly refused entrance to supermarkets or transport on taxis as the staff considered them to be intoxicated. They were also teased by members of the community who did not know them. It appears that this was a common problem to many of the patients when they first became symptomatic. Bearing in mind the high alcohol and substance abuse in these suburbs it is understandable that even their neighbours initially thought they were intoxicated as their past behaviour patterns frequently involved alcohol abuse. However, as people became familiar with them and were informed that they had a neurological disease they stopped being discriminated against and victimised. Patients claimed that the Medic Alert bracelets they wore were of no assistance as there was no explanation of HD provided and comments were made such as, "*I can* *also wear a bracelet like that and also drink and get drunk and then say I have a sickness".*

Lack of money

'Lack of money' was a significant problem to 78.2% (32/41) of the patients and their families. Most of those who had been employed at the time of onset could not continue as they were doing labouring jobs and could not be given a change of job description due to their low levels of education. Seventy-eight percent (32/41) received a DG of R820 per month which had to support a mean of 3 persons per family. In addition to this 24.4% (10/41) of the carers had to leave their jobs especially to care for their affected family member and thus the family income was reduced even further. Some of these carers (7.3%, 3/41) had not applied for a grant-in-aid to which they were entitled as they thought they may be able to find some casual job occasionally which would mean they were not entitled to the grant-in-aid and feared that this might jeopardise their affected family members' DG. Because of the families' financial difficulties a number of the children were forced to leave school before matriculating in order to work to supplement the family income. Care of the patient was often compromised as there was no money for food supplements when they had dysphagia; no money to purchase incontinence clothing and bedding for their special needs; and no consideration given to purchasing wheelchairs and/or aids to maximise their physical independence of ADL and decrease caregiver burden.

Transport

Access to transport was a major problem to 63.4% (26/41) of the patients. Table 50 shows the various forms of transport used for different purposes.

	No access 1	For general use 2	For medical purposes 3	For disability pay-point 4
J3 Own vehicle	26	0	0	0
J4 Vehicle in household	40	0	1	1
J5 Vehicle of family member or friend	38	0	3	3
J6 Train	10	1	4	4
J7 Bus	12	3	3	3
J8 Taxi	2	12	12	12
J9 Dial-a-ride	12	0	2	0
J10 Other	13	2	2	0

Table 50: Access to various forms of transport

The categories in Table 50 relate to the 41 patients and are not mutually exclusive and 'own vehicle' only relates to those patients who did not possess a vehicle. Twenty-four percent (10/41) of the patients never left their homes. Seven percent (3/41) of them could not leave their apartments as there were no elevators and they lived on the second or third storey of the apartment block and could not negotiate the steps. The other 17% (7/41) did not leave their houses because they were unable to walk independently and did not possess wheelchairs in which to be taken outside. Even if they had access to wheelchairs the sandy terrain would have made utilisation of a wheelchair extremely difficult (Photograph 3, page 214; photograph 9, page 220 and photograph 10, page 220). The difficulties they experienced with the public transport are presented in Figure 25.

Photograph 3: sandy terrain outside dwellings

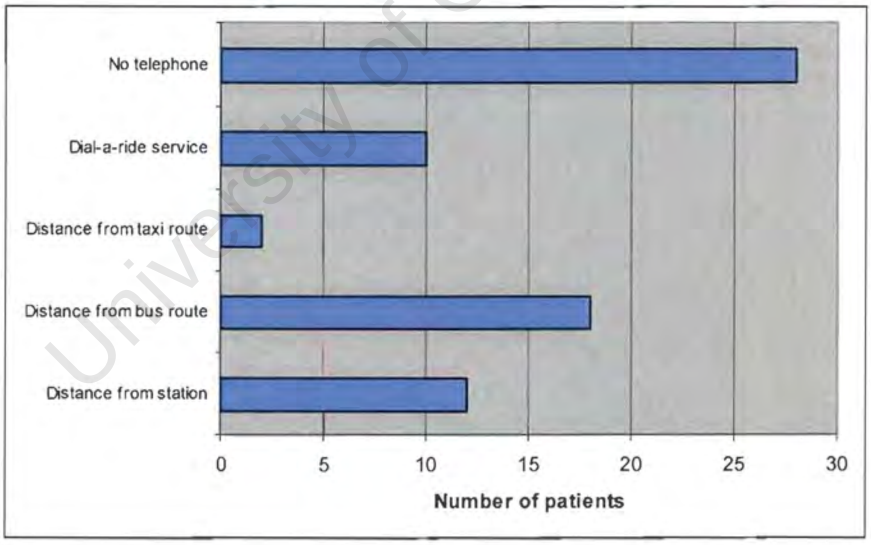

Figure 25: Difficulties experienced with public transport

The main problem with the public transport system was the distance from the train and bus routes. For these patients the Dial-a-ride service was essential to attend medical appointments, but was not accessible to the 12 individuals who required it. They found the service unreliable as the buses arrived too late for them to attend the relevant clinics or else they did not possess a telephone and it was an essential requirement for the use of this service. Those who did have telephones found that the receptionists were unhelpful, impolite and impatient with them if they had mild dysarthria. Patients also found the drivers were loathe to go to certain areas which they considered to be dangerous. As described by several patients and their carers:

"They do not like to come here to Hanover Park, but if your address is Wynberg or Athlone then they don't mind fetching you. They also prefer to fetch the people that go to work everyday. I would also like to work, but cannot because of my sickness. I need the transport to the clinic just as much as they do to go to work. They are supposed to help us, but they do not try at all".

"When you phone they are very rude and unhelpful and the costs are also very high if you need to go to the clinic or hospital regularly".

"I do not have a phone and you have got to have a phone for them to come and fetch you. Nobody in this area has a phone, we are all very poor people".

Social services

Table 48 (page 208) shows that 31.7% (13/41) of the patients reported having 'complete problems' with the social services. These patients had been regularly receiving a grant for two or three years when it was suddenly discontinued with no warning or explanation. When they approached the Wynberg offices of the Social Services Department and had their DGs reinstated it took between six months to one year to receive the monthly payments and there were no back payments made for the months in which they received no payments. These delays had a catastrophic effect on the beneficiaries and their dependents and merely increased their cycle of poverty. They had to borrow money from the local loan sharks and were never able to fully repay their loans due to the interest rates charged.

Housing

Ownership		House	Flat	Wendy house	Total
Owned	Self-constructed formal	13	0	0	13
	Council	9	0	0	9
Rented	Self-constructed	3	0	0	3
	Council	1	8	0	9
	Self-constructed informal	0	0	4	4
Homeless					3
	Total	**26**	**8**	**4**	**41**

Table 51: Type and ownership of dwellings (n-41)

Table 51 presents the type and ownership of housing in which the patients lived. Fifty-three percent of the dwellings were owned by a member of the family which is lower than the general population in those suburbs where ownership is between 63% and 79%. This confirms the findings in the UK and USA that the percentage of people with disabilities owning homes is lower than the general population (Little 2002; Statistics SA 2001; Locker 1983). Seven percent (3/41) of the patients were homeless people who had no permanent accommodation. They spent nights with various charitable neighbours who also provided them with food. One patient lived in a car on the property of a neighbour of her sister (Photograph 4). All 3 patients were aggressive and abused alcohol and drugs. Their relatives refused to have them in their houses as they were belligerent and physically violent. As they received a DG they were welcomed by people who shared their abusive lifestyle. One patient described the relationship with her hosts as,

"When the time comes for my disability grant I have lots of friends, but when the money runs out so does their friendship and I am back on the streets waiting for some kind person to offer me a place and food for the night".

Photograph 4: car which was 'home' of a patient

The housing problems listed below relate specifically to the 78% (32/41) patients who received a DG (Figure 26).

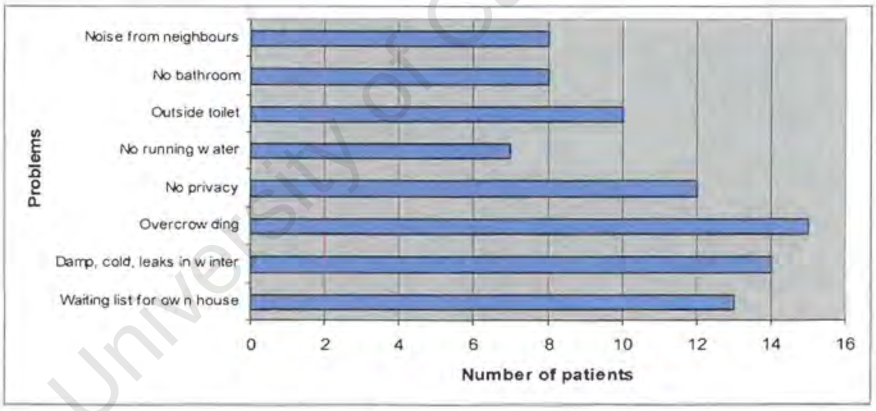

Figure 26: Housing problems experienced by patients receiving disability grant (n=32)

The housing in these low-economic suburbs comprises mostly of high-density semi-detached houses, maisonettes, flats or Wendy houses built in backyards (Photographs 5, 6, 7 and 8). Many of the houses do not have bathrooms and none of the Wendy houses have bathrooms or toilets. Eight patients did not have running water in their houses/flats as they had not been able to pay their water bills and had been put onto a 'drip' system where they could collect one litre of water per person per day from the tap which was set to 'trickle' water.

Photograph 5: Blocks of one and two bedroom flats rented from the council

Photograph 6: Backyard Wendy house with no bathroom or toilet

Photograph 7: Maisonette without front door rented from the council

Photograph 8: Self-owned house

Four who lived in informal houses did not have access to piped running water, but had to walk to a community stand for their needs (Photograph 9 and 10).

Photograph 9: Informal housing settlement with no running water or waterborne sewerage

Photograph 10: self-constructed dwelling with no form of water or sewerage

Residents in the flats complain a great deal of the noise from neighbours either from teenagers who play loud music throughout the day and night or from neighbours who abuse alcohol resulting in domestic arguments and physical confrontations. Thirteen of the patients had been on waiting lists for their own house for between 12 and 15 years. As their children had matured and had their own children and were unable to find houses they either moved in with their parents or built Wendy houses in the backyards. It was not unusual to see a maisonette with 3 or 4 Wendy houses accommodating 25 relatives from three generations on the property. Overcrowding was reported to be a major problem (Figure 26, page 217) as not only were there too many people per room, because of space limitation, it meant that two or three people also had to share a bed. The Wendy houses did not have toilets, but used the facilities of the main house. This was difficult for the patients as the houses were locked at night for safety reasons and they had to resort to a bucket system for their personal use.

Isolation and loneliness

As can be seen from Table 48 (page 208) 'isolation and loneliness' were not considered a major problem due to the level of overcrowding in the homes. Whenever the researcher was conducting interviews there was a constant stream of family members or neighbours coming in and out of the houses and there would be an exchange of greetings, a bit of banter and local news shared with the patients. Patients who did experience loneliness and isolation were from higher income families where there were only one or two people in the home and the neighbourhoods in which they lived tended to be more reserved with fewer spontaneous ad hoc visits between neighbours.

Abuse

Sixty-three percent (26/41) of the patients had experienced a problem with 'abuse'. Sometimes it had occurred over a long period of time in the case of domestic violence related to alcohol or drug abuse, but in other cases it could have been related to the fact that the patients' mothers were affected with HD while they were growing up (56.1%, 23/41) and were unable to care for their children adequately. However, most of the patients who experienced abuse as adults lived in suburbs which are regarded as having a high level of domestic violence. It is not possible therefore, to determine whether this was as a result of growing up in a dysfunctional family because of a parent having HD or the environment in which they lived. Figure 24 (page 209) gives an indication of the incidence of crime in the precincts of Grassy Park and Steenberg where the majority of patients lived. Presently police statistics on the incidence of domestic violence are not available because there is no crime categorised as 'domestic violence' so it is not possible to state whether the domestic violence amongst the patients with HD and their families compared to that of the specific community and precinct (Vetten and Bhana 2002) or was higher.

Table 52 presents the frequency and type of abuse to which the patients were subjected during their childhood and as adults.

	Perpetrator	Psychological	Physical	Sexual	Frequency	Percentage
Childhood	Children	3	3	0	6	30
	Family	3	3	6	12	60
	Community	1	1	0	2	10
	Total	7	7	6	20	100
Adulthood	Family	6	6	3	15	58
	Community	4	5	2	11	42
	Total	**10**	**11**	**5**	**26**	**100**

Table 52: Frequency and type of abuse experienced by patients (not mutually exclusive)

In spite of the patients' claims of family and community abuse there was only one occasion that the researcher observed bruising consistent with physical abuse and which was confirmed by the patient and her husband. Otherwise the patients were cared for very well under very difficult circumstances. However, it is generally accepted that the abuse of females by their spouses/partners in the WC is approximately 44% (Vetten and Bahna 2002). In four of the homes visited there were two patients who had served prison sentences for incest: one was for attempted rape of a step-daughter and the other for the rape of a friend. In the other two homes other family members had served sentences for attempts of rape of teenagers by adults living in the same house.

The families were compassionate about the needs of the patient and were tolerant of their individual idiosyncracies, likes and dislikes with regard to meal times, getting up, being put to bed and being bathed. Several families used humour as a coping strategy which Ross and Deverell (2004) claim is only an option when the patient enjoys a secure sense of self and receives strong social support. The only complaint of three of the patients was that their families collected their DGs and did not give it to them, but used the money for purchasing food, electricity and clothes for the extended family, many of whom had no form of employment and income and made no attempt to obtain employment. With regards to their smoking and drinking habits before and after they became symptomatic there appears to be a slight decrease in the amount of alcohol consumed, but an increase in smoking (Table 53 and Table 54) which is contrary to the findings in other studies (Reynolds 2004; Jensen et al 1998; Livneh and Antonak 1997; Falvo 1991).

	Not at all 2	A few 3	Moderately 4	Heavily 5
W14 How much did you smoke before you developed symptoms of HD?	6	1	11	23
W15 How much did you smoke after you developed symptoms of HD?	8	2	5	26

Table 53: Smoking habits before and after symptoms (n=41)

	Not at all 2	A few 3	Moderately 4	Heavily 5
W18 How much did you drink before you developed symptoms of HD?	12	7	7	15
W19 How much did you drink after you developed symptoms of HD?	24	6	4	7

Table 54: Drinking habits before and after symptoms (n=41)

The patients explained that they did not drink as much afterwards as they were taking medication for their movements or sleeping tablets and they thought it would be dangerous. Their families also limited their intake of alcohol by not making it available and they could not walk to the shops independently. Smoking became more and more of a safety problem as patients frequently burnt themselves or their clothing and were at-risk of setting fire to their informal wooden houses. Most of them were restricted by their families to smoking only in the rooms which had tiled or cement floors.

Disability

Many studies on people with disabilities have found that individuals appeared to accept their disabilities as a fact of life and simply as one of the many personal characteristics they possessed, which was not the most significant or important influence on their lives. The findings of other authors confirm that most disabled considered their disabilities natural and a mundane inconvenience only and relatives of the disabled who had had close contact with the disabled for many years had a less tragic, succumbing view of disability than outsiders (Futter 2004; Lonsdale, 1990; Gething, 1985; Weinberg and Williams, 1978). However, this was not the case with the patients with HD in the WC who believed that their disabilities were the worst thing that possibly could have occurred in their lives and 92.6% (38/41) found it to be a problem which affected and compromised every aspect of their daily living. Their quotes below describe the many different ways in which HD affects their lives:

"I used to be a proper person who could work, do my housework, and look after my children and my husband. Now I can't work anymore and we really need the money and every day almost there is something else I cannot do and my family has to do it for me. It is very depressing because I am just making more and more work for them exactly the same as when I looked after my mother when I was young and she had the sickness".

"When I walk in the streets everybody thinks I am drunk and they move away from me. Yesterday when my mother was taking me to the clinic and we had to cross the road the taxi driver thought I was drunk and wouldn't let me get in until my mother shouted at him that I was !! sick and not !! drunk".

"He doesn't enjoy going out to restaurants anymore because people stare at him when he drops his knife or fork or messes his food and bumps into the tables when he walks. The family also get embarrassed by his behaviour and so we have decided not to go out anymore to public places with him for his own sake". (Proxy speaking on behalf of patient with dysarthria).

"Yesterday I had to go to the supermarket and the security would not let me in because they said I was drunk. When I tried to tell him I had a sickness I could hear myself that I sounded drunk. It is very embarrassing as there were a lot of people around and it made me feel terrible to be called drunk when I really wasn't".

"Each day I wake up and wonder what the next deterioration is going to be. Last week I was unable to remember my multiplication tables and so I can't do our family budget anymore. I've always done that every month and was good at it. I just envisage going down the same miserable degrading pathway as my father did before he reached the end of his tether and committed suicide. I sometimes think that he made the right decision". This was also found by Evers-Kiebooms and Decruyenaere (1998).

"I don't like to have friends visit me or go to them because I can't take part in the conversation and they get tired of talking and talking and nothing from me. My husband forces me to socialise to keep in touch with people, but I find it very stressful as I used to be a very chatty person and now I just can't keep up with the conversation".

Table 55 indicates the severity of the disease triad and the capacity of the patients to function in society.

	No problem 1	Mild problem 2	Moderate problem 3	Severe problem 4	Complete problem 5	
Engagement in occupation	0	0	4	1	36	F33
Capacity to handle financial affairs	0	2	1	1	36	F34
Capacity to manage domestic responsibility	0	0	4	5	32	F35
Capacity to engage in social activities	4	2	15	4	15	F36

Table 55: Capacity of patients to function in society (n=41)

4.10.4 SOCIAL SUPPORT

Table 56 presents the kinds and extent of emotional support the patients received. It shows that the majority of patients felt that social support was available to them 'most' to 'all of the time'. Only a few patients had social support available to them between 'none' and 'some of the time'.

		None of the time	A little of the time	Some of the time	Most of the time	All of the time
M1	Someone to help you if you have to stay in bed	3	1	1	0	36
M2	Someone you can count on to listen to you when you need to talk	6	1	2	0	32
M3	Someone to give you good advice about a crisis	4	3	1	1	32
M4	Someone to take you to the doctor if you needed it	3	0	2	0	36
M5	Someone who shows you love and affection	5	1	1	0	34
M6	Someone to have a good time with	7	0	2	0	32
M7	Someone to give you information to help you understand a situation	5	1	1	0	34
M8	Someone to confide in or talk to about yourself or you personal problems	6	2	1	0	32
M9	Someone who hugs you	5	1	1	2	32
M10	Someone to get together with for relaxation	5	1	2	0	33
M11	Someone to prepare your meals if you were unable to do it yourself	4	0	1	0	36
M12	Someone whose advice you really want	5	2	1	1	32
M13	Someone to do things with to help you get you mind off things	7	1	1	0	32
M14	Someone to help with your daily chores if you were sick	3	1	2	0	35
M15	Someone to share your most private worries and fears	6	1	2	0	32
M16	Someone to turn to for suggestions about how to deal with a personal problem	6	2	1	0	32
M17	Someone to do something enjoyable with	7	1	1	0	32
M18	Someone who understands your problems	7	1	1	0	32
M19	Someone to love and make you feel wanted	5	1	2	0	33

Table 56: The Medical Outcomes Study Social Support Survey (MOS) (n=41)

The Cronbach Alpha coefficient based on 41 patients and 19 items measuring 'social support' was 0.973 showing a high level of internal consistency. This allowed the multiple-item scale to be transformed into a single additive scale.

For further analysis the rating categories were recoded into three categories: 'none' to 'some' social support (1), social support 'most of the time' (2) and social support 'all of the time' (3). The result is summarised in Table 57.

	Frequency	Percentage	Cumulative Percentage
Some of the time	6.8	16.5	16.5
Most of the time	0.2	0.5	17.0
All of the time	34	82.9	100
Total	**41**	**100.00**	

Table 57: The amount of time social support was available

These findings are confirmed by Lonsdale's (1990) research in the UK where she found that living in extended families was helpful for people with disabilities because there was no need for them continually to explain their needs and, as the extended families provided company, it meant that the people with disabilities were never lonely. This also happened in strong supportive communities which were always willing to help and provide the disabled people with company (Lonsdale, 1990).

The results of a 2-dimensional CAT-PCA, explained 97% of the total variance. Table 58 presents the component loadings of the 19 items of the scale.

		Dimension	
		1	2
M1	Someone to help you if you have to stay in bed	.768	.627
M2	Someone you can count on to listen to you when you need to talk	.987	-.148
M3	Someone to give you good advice about a crisis	.986	-.157
M4	Someone to take you to the doctor if you needed it	.802	.553
M5	Someone who shows you love and affection	.933	.101
M6	Someone to have a good time with	.980	-.181
M7	Someone to give you information to help you understand a situation	.933	.101
M8	Someone to confide in or talk to about yourself or you personal problems	.987	-.148
M9	Someone who hugs you	.989	-.142
M10	Someone to get together with for relaxation	.961	-.084
M11	Someone to prepare your meals if you were unable to do it yourself	.798	.589
M12	Someone whose advice you really want	.986	-.164
M13	Someone to do things with to help you get you mind off things	.972	-.204
M14	Someone to help with your daily chores if you were sick	.789	.603
M15	Someone to share your most private worries and fears	.970	-.211
M16	Someone to turn to for suggestions about how to deal with a personal problem	.970	-.211
M17	Someone to do something enjoyable with	.980	-.181
M18	Someone who understands your problems	.970	-.213
M19	Someone to love and make you feel wanted	.965	-.065

Table 58: Component loadings of the 19 items of the MOS

Figure 27 shows the component loadings graphically and reveals three clusters of items. The top one is constituted by items M1, M14, M11 and M4 which appear to relate to 'physical support' required to assist them with their basic needs of living when they are unable to be functionally independent. The middle one is constituted by items M7 and M5 relating to 'empathy' where a person who 'shows emotions of love and affection' is able to 'provide them with information which helps them understand the difficulties of a situation'. The next cluster M19 and M10 appears to relate to 'relaxing with somebody' who 'loves and makes them feel wanted'. The bottom one consists of the remaining variables M9, M8, M2, M12, M3, M6, M17, M13

M15, M16 and M18 which relate to a compassionate person who understands the patient's worries and fears and whom he/she can trust and confide in as well as have confidence in his/her advice. Unfortunately the labeling of the vectors is not clear as they are located too close together for clarity.

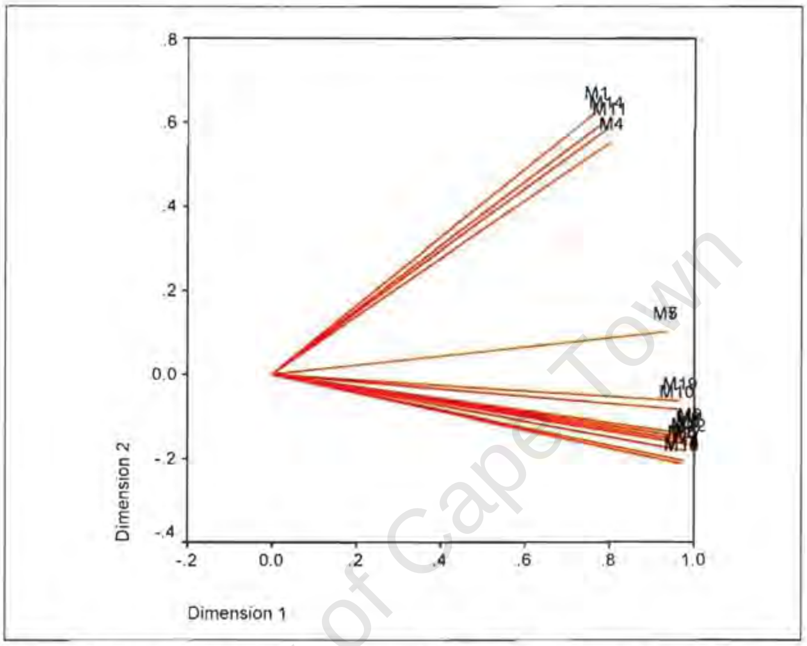

Figure 27: Component loadings of the 19 items of the MOS

In response to the question, "What has a negative impact on the amount of your socialisation?", patients' responses are recorded in Table 59.

	No problem 1	Mild problem 2	Moderate Problem 3	Severe problem 4	Complete problem 5
M38 Embarrassment of your disability	7	4	11	9	10
M39 Your disability	0	0	7	11	23
M41 Transport difficulties	17	5	5	4	10
M42 Fear of criminals	31	0	4	3	3
M43 No money	12	4	14	1	10

Table 59: Aspects having a negative impact on patient's socialisation (n=41)

A 2-dimensional CAT-PCA explaining 75% of the total variance, reveals how the items are related. Figure 28 shows that items M41 'transport difficulties', M42 'fear of criminals' and M42 'no money' have mobility, lack of money and security in common, determining the first dimension. The second dimension appears to be determined by aspects directly related to the symptoms of the disease.

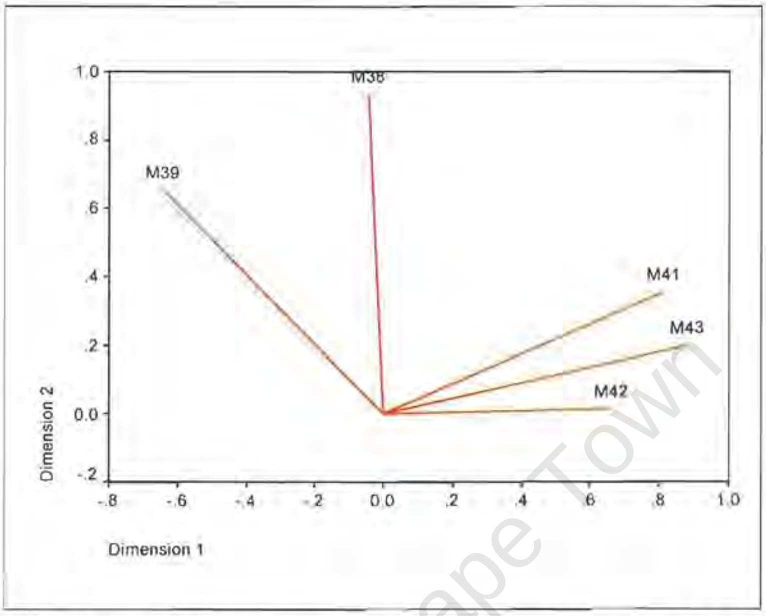

Figure 28: Problems affecting level of socialisation of patients

The main problems in the lives of the individuals and families with HD

When asked what the biggest problem was in their daily lives with regards to HD the responses were listed as in Tables 60 (patients) and 61 (families). Categories were not mutually exclusive.

Category	Symptoms	Frequency	Percentage
Motor	Involuntary movements	2	4.8
	Weakness	1	2.4
	Dysarthria	30	73.2
	Unable to balance	15	36.6
	Falling	7	17.1
	Gait	36	87.8
	Dysphagia	4	9.8
	Total	**95**	**25.6**
Cognitive	Concentration	7	17.1
	Forgetfulness	9	21.6
	Ability to think clearly	5	12.2
	Total	**21**	**5.6**
Affective	Irritability	3	7.3
	Anxiety	24	58.2
	Depression	12	29.3
	Total	**39**	**10.5**
Other	Loss of independence	39	95.1
	Deterioration	38	92.6
	Withdrawing from socialisation	15	36.6
	Worry about personal future	36	87.8
	Loneliness	5	12.2
	Boredom	3	7.3
	Unable to socialise	12	29.3
	Embarrassment of symptoms	24	58.2
	Worry about future of children	10	24.4
	People think I'm drunk	35	85.4
	Total	**217**	**58.3**

Table 60: Biggest problems in the daily life of the patients

It is interesting to note that the majority of patients found 'loss of independence', 'deterioration', 'worry about personal future', 'people think I'm drunk' and dysarthria their biggest problems and not their 'involuntary movements', 'falling', 'dysphagia' and 'boredom'. In contrast the family members found the patients 'lack of insight', 'forgetfulness', 'ability to think clearly' and 'aggression' to be their greatest concerns with coping with them (Table 61). These cognitive and affective disabilities were far more of a problem to the families than the motor components, apart from that of 'dysphagia' and confirm the findings of Quarrell and Cook (2004).

Category	Symptoms	Frequency	Percentage
Motor	Restlessness	15	36.6
	Chorea	5	12.2
	Falling	16	39.2
	Dysphagia	17	41.5
	Total	**53**	**11.4**
Cognitive	Concentration	19	46.3
	Forgetfulness	36	87.8
	Ability to think clearly	35	85.4
	Lack of insight	37	90.2
	Total	**127**	**27.2**
Affective	Apathy	12	29.3
	Irritability	19	46.3
	Disinhibition	20	48.8
	Depression	2	4.8
	Aggression	35	85.4
	Compulsive behaviour	9	21.6
	Paranoia	21	51.2
	Total	**118**	**25.2**
Other	Personality change	16	39.2
	Loneliness	3	7.3
	Boredom	5	12.2
	Worry about future of relative	34	83.0
	Not sleeping	28	68.4
	Unable to socialise	17	41.5
	Embarrassment	24	58.2
	Safety of children	4	9.8
	Alcohol abuse	12	29.3
	Stubbornness	26	63.4
	Total	**169**	**36.2**

Table 61: Biggest problems for families coping with the patients

4.11 EVALUATION OF NEUROGENETIC CLINIC

Knowledge of HD

	PT group		Patient group	
	Knows	Does not know	Knows	Does not know
B1 How do people get HD?	25	2	30	11
B2 What part of the body does it affect?	27	0	20	21
What signs can you see that a person has HD?				
B3 Unwanted movements/slow movements/unable to move	27	0	31	10
B4 Loss of memory/decreased intellectual capacity/dementia	27	0	17	24
B5 Anxiety/irritability/depression/personality changes	18	9	17	24
B6 How old are people when they first start to show the signs of HD?	25	2	31	10
B7 Is there a cure for HD?	27	0	33	8
B8 What is the treatment for HD?	27	0	33	8
What are the risks of your family having HD?				
B9 brothers and sisters	25	2	20	21
B10 parents	25	2	17	24
B11 children	26	1	19	22
B12 yourself	24	3	17	24
What kind of genetic disorder is HD?				
B13 autosomal	1	26	0	41
B14 dominant	3	24	0	41
B15 Are males and females equally affected with HD?	27	0	30	11
Mean	**81.3%**		**20.8%**	

Table 62: A comparison of the level of knowledge of HD between the PT group and the patient group

At the time of the interview all patients or patients and carers had attended the Neurogenetic Clinic on at least 4 occasions. Sixty-eight percent of them did not know the name of the disease with which they were affected was called Huntington disease. Table 62 presents the patients' and patients'/carers' level of knowledge of HD. As was found in the PT group, the patients and carers had difficulties with items B14 and B15 and none of them could answer the question correctly. When asked how people developed HD the majority knew that it was a family disorder, but had no idea about the genetic pattern of inheritance and when asked how they inherited the disorder from their parent the following responses are typical of their understanding of it.

"Well it's like my father had HD and so I also got it. Then he got TB so I also got it. Not all of my brothers got HD and only two of us got TB so those things are just in the family. It skips a generation because you know my grandmother had it and then none of her children had it and now two of my cousins have started with it".

"I got it from my mother. She got it from her mother because they looked just the same and they had the same temper. Then me and my two brothers and one sister got it because we look just like my mother and we are all stubborn people and get cross very quickly".

"You can say it's just like a weakness in the family. Like some people are very tall in one family and then every now and then suddenly there is one short one born that doesn't look like the family at all. In our family we all get it at the same age and it's only in the men. They start at about thirty and then die when they are nearly fifty".

"When I was young I had bad friends. We used to use a lot of tik (crystal methamphetamine) and once I was unconscious and had to go to hospital. Six months after I came out of hospital then these things started. I couldn't walk properly and it was difficult to speak. It's all from that tik-taking when I was in the gang".

These perceptions confirm the findings of Meiser et al (2001) that there are general beliefs amongst families with inherited diseases about family patterns of inheritance being based on appearance, mannerisms and behaviour. The concepts of inheriting a disorder are difficult for individuals to comprehend, particularly if they have a low standard of education. In reality it also means that if the individuals have transport difficulties it frequently happens that all the information has be given by the counsellor in one session. Under these circumstances all the information has to be given during this session and does not permit a period of reflection and consolidation by means of repetition before hearing further information about the disease. This frequently leads to misunderstandings and they may never have these misconceptions clarified.

A comparison of the knowledge of HD of the two groups shows a large difference in accuracy of information (Table 62). The mean score of the patient group was 20.8% which was significantly less than the PT group with 81.3%. Reasons for this discrepancy are the different levels of education (Eggers et al 1999), income (Bloch 1989) and possibly the manner in which they received genetic counselling. Usually they are provided with genetic information at their first attendance at the neurogenetic clinic. This is a stressful time as the information evokes anxiety and interferes with the processing of information and not all information receives the same attention or is remembered to the same extent (Kessels 2003; Pilnick and Dingwell 2001; Hammond 1998). The patients are undoubtedly focused on their medical problems and follow-up appointments so the genetic information is not processed in memory and is forgotten. When the patient returns for confirmation of the diagnosis the genetic counsellor revises the information, but after that the patient does not see the counsellor again as the review appointment is usually for 6 months later. Therefore, the patients do not have much contact with the genetic counsellor after the diagnosis is made.

Items B13 'autosomal' and B14 'dominant' appeared to be difficult to answer for both groups: only 5.8% (4/68) of the total number of respondents were able to answer the question correctly. This information is provided by the genetic counsellors and the majority of PT participants had access to the internet and commented on the amount they used it for keeping up with new information so it there is no reason for them finding difficulty with it. It is understandable in the case of the patients and their caregivers as their average number of years at school was so low and they probably had not yet reached the stage of attending biology or physiology courses and thus they had difficulty in retaining the information provided them during their counselling session.

The relationship between the items and the group variable was explored by a CAT-PCA on the 13 remaining items and group. A two-dimensional solution explained almost 65% of the total variance. Figure 29 reveals three clusters.

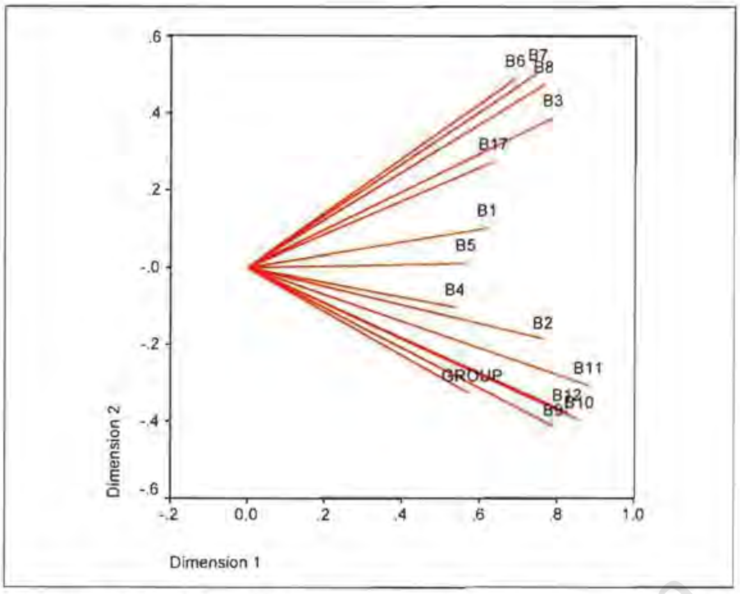

Figure 29: A plot of the component loadings and the 13 items

The cluster of interest, namely, the one relevant to the group variable, consists of items B4 'loss of memory', B2 'part of the body affected', B11 'risks of children', B12 'risk of yourself', B10 'risk of parents', and B9 'risk of siblings', suggesting that these items discriminate predominantly between the PT group and patient group (Figure 30). A plot of the category scores shows that the PT group (denoted as 'a' in the graph) is associated with 'knowing' and the patient group (denoted as 'b' in the graph) with 'does not know'.

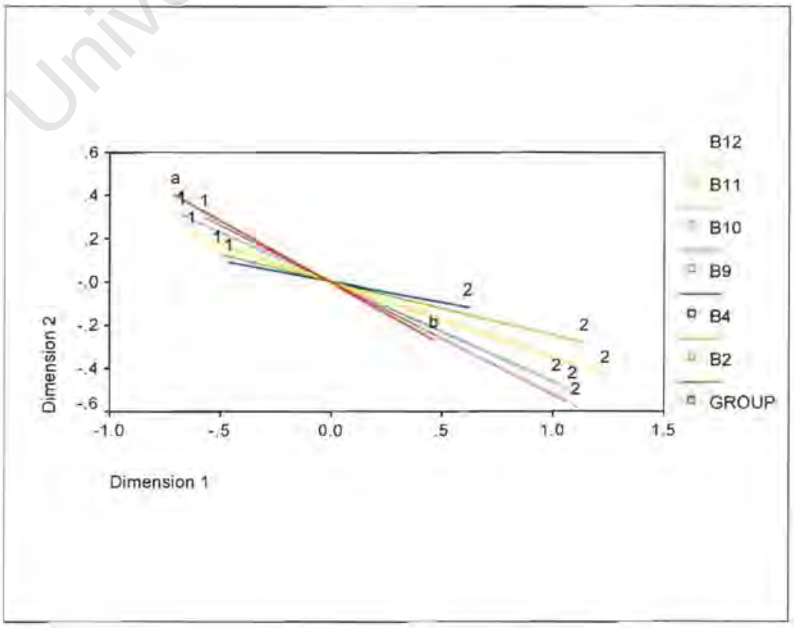

Figure 30: Plot of category scores of selected items

This is confirmed by a logistic regression analysis on these five items as independent variables: the model[1] predicted that 21 respondents correctly belong to the PT group and 32 to the patient group (Table 63).

It was clear that the patients understood that some of their children could develop the symptoms, but appeared to be unaware that their grandchildren were also at-risk.

Observed	Predicted		
	Group		Percentage Correct
	PT group	**Patient group**	
PT group	21	6	77.8
Patient group	9	32	78.0
Overall Percentage			**77.9**

Table 63: Classification based on logistic model

SATISFACTION WITH THE NEUROGENETIC CLINIC

Knowledge of PT

All of the patients knew about the PT programme and all of them who had children over 18 years of age had informed them of the testing that was available to them, but to date none of the children had availed themselves of the opportunity. Their parents' perceptions of their adult children's reactions to being told of the availability of the test is presented in Table 64. The categories are not mutually exclusive. Generally when the researcher personally discussed testing with the adult children caring for an affected parent their attitude was most accepting and fatalistic.

"It doesn't' really matter whether I get it or not. It's a family thing you know and in our family we have always just accepted responsibility for those that get the sickness. One day if I get it one of my sisters will look after me and if they get it I'll look after them so what it the use of knowing about it?"

---

[1] $\text{prob (event)} = \frac{1}{1+e^{-z}}$

"We have lots of family affected. Cousins, uncles, aunts, grandparents. When we see somebody is dancing to their own tune then we know that they have started with this thing. Its just one of those things so I don't really bother about it. I did want to get tested at one time, but now my wife and I just accept that if I get it its' too bad. There's nothing you can do about it anyway".

Response	Reaction	Frequency	Percentage
Denial	Never spoke about it again	3	7.3
	Avoided the topic when HD was discussed	4	9.8
	Distanced themselves from their parents	2	4.8
Fatalistic	Didn't mean much to them either way	15	36.6
Decided against testing	No treatment or cure available	17	41.5
Would consider testing when planning children	Procrastination – never presented for testing	20	48.8
Don't want to know status	No discussion in the family	5	12.2

Table 64: Parents' perceptions of adult children's responses to being told of availability of HD test

Satisfaction Scale of the Neurogenetic Clinic

Items		Very unsatisfactory	Unsatisfactory	Satisfactory	Very Satisfactory	Highly satisfactory
		1	2	3	4	5
N19	The manner in which your symptoms are explained to you	0	7	14	15	5
N20	The staff listen to what you have to say	0	0	1	12	28
N21	The staff understand what is worrying you	0	7	12	12	10
N22	The staff answer all your questions	0	0	0	21	19
N23	The staff show you a lot of caring	0	0	1	14	26
N24	Privacy when seeing the doctor	0	16	15	9	1
N25	The clinic helps you to cope better with your problems	0	0	4	14	23
N26	There is enough time given to you at each session	0	0	1	18	22
N27	The clinic environment	6	11	10	9	4
N28	The staff reassure you	1	0	2	14	24
N29	The manner in which the diagnosis was presented to you	0	0	3	16	22
N30	There is time to have your questions answered	0	0	0	20	21
N31	The time you wait to get an appointment if there is a need out of your regular clinic appointments	0	0	0	18	23
N32	The time you wait to see the doctor at each visit	7	9	16	5	4
N33	The staff are interested in you as a person	1	0	2	17	21
N34	Decisions are made for you with full explanations provided to you	0	0	3	16	22
N35	Clinics/visits provide you with a lot of support	7	8	13	11	2
N36	General administration and concern shown by genetic sisters	0	0	1	16	24
N37	Overall, how do you rate the neurogenetic clinic?	0	0	1	23	17

Table 65: The rating frequencies per item on the satisfaction scale (n=41)

Table 65 has been re-coded by merging 'very unsatisfactory' and 'unsatisfactory' ratings to decrease the variance. The result is presented in Table 66.

	Items	Very unsatisfactory plus Unsatisfactory 1	Satisfactory 2	Very Satisfactory 3	Highly satisfactory 4
		1	2	3	4
N19	The manner in which your symptoms are explained to you	7	14	15	5
N20	The staff listen to what you have to say	0	1	12	28
N21	The staff understand what is worrying you	7	12	12	10
N22	The staff answer all your questions	0	0	22	19
N23	The staff show you a lot of caring	0	1	14	26
N24	Privacy when seeing the doctor	16	15	9	1
N25	The clinic helps you to cope better with your problems	0	4	14	23
N26	There is enough time given to you at each session	0	1	18	22
N27	The clinic environment	18	10	9	4
N28	The staff reassure you	1	2	14	24
N29	The manner in which the diagnosis was presented to you	0	3	16	22
N30	There is time to have your questions answered	0	0	20	21
N31	The time you wait to get an appointment if there is a need out of your regular clinic appointments	0	0	18	23
N32	The time you wait to see the doctor at each visit	16	16	5	4
N33	The staff are interested in you as a person	1	2	17	21
N34	Decisions are made for you with full explanations provided to you	0	3	16	22
N35	Clinics/visits provide you with a lot of support	15	13	11	2
N36	General administration and concern shown by genetic sisters	0	1	16	24
N37	Overall, how do you rate the neurogenetic clinic?	0	1	23	17

Table 66: Recoding of Table 65 by merging 'very unsatisfactory' and 'unsatisfactory' ratings (n=41)

Table 67 presents the results of a three-dimensional CAT-PCA on the items measuring satisfaction.

Dimension	Variance Accounted For	
	Eigenvalues	% of Variance
1	3.020	16.778
2	2.657	14.763
3	2.152	11.954
Total	**7.829**	**43.496**

Table 67: Results of a three-dimensional CAT-CPA on the items measuring satisfaction

The accompanying component loadings are presented in Table 68.

Items		Dimension		
		1	2	3
N19	The manner in which your symptoms are explained to you	-.192	-.551	-.190
N20	The staff listen to what you have to say	.379	.631	-.150
N21	The staff understand what is worrying you	.663	-.353	-.041
N22	The staff answer all your questions	.079	-.246	-.076
N23	The staff show you a lot of caring	-.125	.702	-.048
N24	Privacy when seeing the doctor	.600	-.017	.543
N25	The clinic helps you to cope better with your problems	-.102	.823	-.019
N26	There is enough time given to you at each session	-.477	.330	.010
N27	The clinic environment	-.394	-.065	-.158
N28	The staff reassure you	.403	-.154	.050
N29	The manner in which the diagnosis was presented to you	.010	.185	-.381
N30	There is time to have your questions answered	.419	.058	-.087
N31	The time you wait to get an appointment if there is a need out of your regular clinic appointments	-.357	-.459	.251
N32	The time you wait to see the doctor at each visit	.436	.216	.521
N33	The staff are interested in you as a person	.627	.270	-.406
N34	Decisions are made for you with full explanations provided to you	-.226	.270	.645
N35	Clinics/visits provide you with a lot of support	.625	-.122	.353
N36	General administration and concern shown by genetic sisters	.364	-.088	-.753

Table 68: Component loadings per item of a three-dimensional CAT-PCA

Items N21, N24, N33 and N35 are strongly related and contribute relatively strongly to dimension 1. Item N21 'staff understand what is worrying you', N24 'privacy during consultations', N33 'staff are interested in you as a person' and N35 'clinic provides you with a lot of support' appear to refer to the level of empathy received during clinic visits (dimension 1). Item N25 'clinic helps you to cope better with your problems' has the highest contribution to dimension 2, followed by items N23 and N20. Thus dimension 2 appears to be the amount of caring received which helps the patients to cope better with their problems by the staff listening to what they have to say. Item N36 contributes relatively strongly to dimension 3 followed by N34, N32 and N24. This psychological construct appears to be the amount of concern displayed during the time they had to wait for the doctor and providing them with information in privacy.

Table 69 presents the Spearman Rank correlation between Item N37 'the overall rating of the neurogenetic clinic' and the objectscores of the three dimensions. The correlation between item N37 and the objectscores on dimension 2 are the highest indicating that the overall rating of the clinic is somewhat determined by dimension 2: the amount of caring received from the staff.

Correlation Coefficient	Object-scores dimension 1	Object-scores dimension 2	Object-scores dimension 3
Item N37	.252(**)	.418(**)	-.007(**)
Sig. (2-tailed)	.117	.007	.968
N	41	41	41

** Correlation is significant at the 0.01 level (2-tailed)

Table 69: Spearman Rank correlation between item N37 and objectscores of dimensions 1, 2 and 3

As patients and their families comprehend that there is no medical treatment for HD they attend the neurogenetic clinic for communication with the staff members to be reassured that everything possible is being done for the patient and that the caregivers are providing their affected relatives with the best possible care. Thus with this neutral approach to their management it appears that their level of satisfaction would start from a negative position (Shiloh et al 1990). However, from the actual evaluation scores the patients rated their satisfaction with the neurogenetic clinic positively. Figure 31 shows the median per item with the green line indicating the median of item D19 which refers to 'the overall satisfaction with the testing program'. The majority of patients considered the various components of the clinic to be 'very satisfactory' or 'highly satisfactory' and all the items were rated at least as 'satisfactory'.

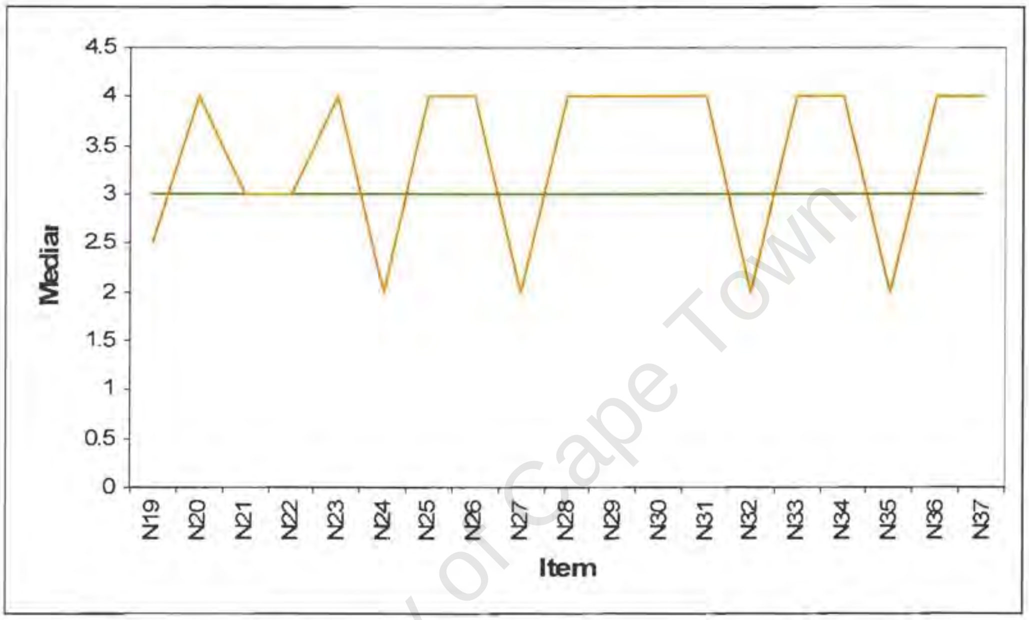

Figure 31: Median of the rating scores per item

The median and mode statistics and the 75th percentile of the nineteen items of the satisfaction scale are presented in Table 70.

	N19	N20	N21	N22	N23	N24	N25	N26	N27	N28
N	41	41	41	41	41	41	41	41	41	41
Median	2.50	4.00	3.00	3.00	4.00	2.00	4.00	4.00	2.00	4.00
Mode	3	4	2	3	4	1	4	4	1	4
75th Percentile	4.00	5.00	4.00	5.00	5.00	3.00	5.00	5.00	4.00	5.00

	N29	N30	N31	N32	N33	N34	N35	N36	N37
N	41	41	41	41	41	41	41	41	41
Median	4.00	4.00	4.00	2.00	4.00	4.00	2.00	4.00	3.00
Mode	4	4	4	2	4	4	1	4	3
75th Percentile	5.00	5.00	5.00	3.00	5.00	5.00	4.00	5.00	5.00

Table 70: Non parametric statistics per item

Item N24 'privacy when seeing doctor', item N27 'clinic environment', item N32 'time waiting to see doctor' and item N35 'clinic provides you with a lot of support' which were rated as 'satisfactory' only were followed up by in-depth questioning to investigate the reasons for not scoring the items more positively and to suggest how these aspects could be improved upon (Figure 32).

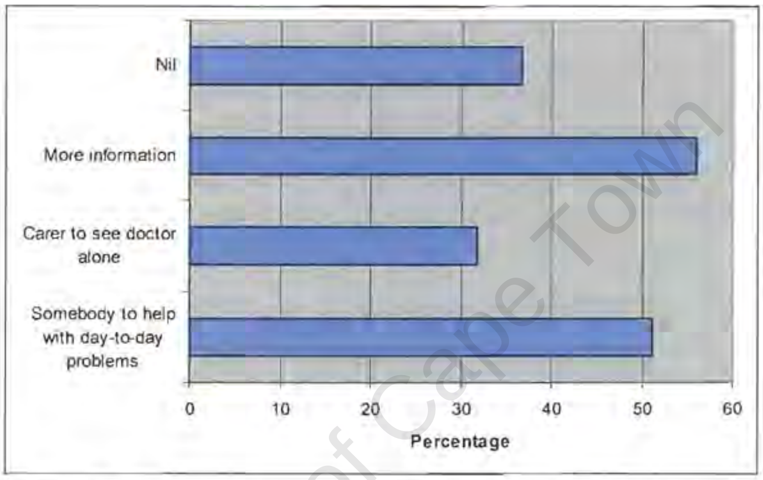

Figure 32: Patients' recommendation for improvements to genetic counselling service

It is interesting to note that, although one of their problems was 'waiting for the doctor' they did not have any suggestions to change this. They appeared to accept it as inevitable and something that simply had to be tolerated in order to receive the service. Fifty-six percent (23/41) requested more information about the disease and its management. Although they had been issued with leaflets at the time of diagnosis they were in English and the patients and their families claimed that they were too difficult to understand because of their low level of education and because their home language was Afrikaans. The carers of patients who had psychiatric symptoms with obsessive-compulsive tendencies and sexual disinhibition felt strongly that the carers should be seen separately by the doctor as they could not always report their problems in front of the patients and so they went away from the clinic with their most difficult problems unheard. Fifty-two percent (21/41) requested to talk with somebody who could help them with the management of their relatives at home and to reassure them that what they were doing was the best or to be able to obtain advice on their management. Some of the families discussed that this service was available when there was an active HD support group, but since its demise they had nobody to whom they could turn. They expressed the need for this to be made available for them at the clinic.

Difficulties experienced with attending Neurogenetic Clinic

The outcomes of the open-ended question, "What difficulties do you have in attending the Neurogenetic Clinic?" are presented in Table 71. The difficulties reported were mostly in connection with getting to the clinic and waiting for different components of the visit. The categories are not mutually exclusive and the quotations below are self explanatory about the problems experienced.

Problems experienced	Frequency	Percentage
Distance	18	43.9
Costs	33	80.5
Transport	18	43.9
Early start in the morning	15	36.6
Waiting for folder	18	43.9
Waiting for doctor	24	58.2
Waiting for medication	14	34.2
Accessibility of social worker	5	12.2
Nobody to help with day-to-day care problems	23	56.1
Nil	8	19.6

Table 71: Difficulties experienced with attending Neurogenetic Clinic

"Going to the clinic is a big problem for me. I have to ask a friend for a ride to the bus station. I have to get up at half past four in the morning to dress and feed my wife and pack food for her for the long morning to be in time for the ride. Then it is so difficult to get her into the bus. We get to the hospital and have to wait for the folder. Then we have to wait for the doctor. Then we have to wait for the tablets and all the time my wife is being difficult because she is impatient. And when she gets impatient she makes a big fuss in front of all the people which is very embarrassing for me. The only time she sits quietly and waits is when she is waiting for the doctor. She loves to go on the outing, but for me it's a complete nightmare".

"It is very difficult to always go to the clinic for our appointment. It costs us a lot of money for the taxi and train and the tablets don't really help. The staff is always very nice to us, but what can they do. There's nothing that helps this disease and the biggest problems are the difficulties at home and there is nobody to help us with those problems. Like what to do at home when he gets up at night and wanders around the house so I can't sleep for worrying

about what he's going to do. He wanders into the girls' bedroom and disturbs them and when I take him back to our bedroom he shouts and gets aggressive with me".

4.12 SUMMARY OF PATIENTS' PROFILE

The majority of the patients were from a low educational and socio-economic group. Their mean number of years of education was 9.4 years and 54% (22/41) had elementary occupations during the time they had been employed. The mean AOO of symptoms was 39.6 years and the first symptoms to appear were cognitive and affective disturbances. Twenty-nine percent (12/41) of the patients had been disabled for more than 10 years. Forty-five percent (18/41) of them had major problems with their activities of daily living and 34% (14/41) were severely dependent. Thus they were severely compromised with regards to independence and quality of life as they were unable to perform basic functional activities. The anxiety and depression scores of the HAD scale showed the presence of anxiety in 60.1% (25/41) and depression in 29.3% (12/41) of the patients. The amount of the DG was frequently insufficient to meet their special needs as a direct result of their disability. The major social problems of 78% (32/41) of the patients experienced were lack of money, access to transport and inadequate and substandard housing without basic amenities such as bathrooms, toilets, running water. A variety of inexpensive simple assistive devices in the homes would have allowed greater independence for the patients and decreased burden for the caregivers (Figure 23, page 207). Although 60.1% (25/41) experienced abuse during their lives it is not possible to say this was as a result of being in a dysfunctional family with HD or because of the high level of domestic violence and general violence in the suburbs in which they lived. Eighty-two percent (34/41) of patients reported receiving social and emotional support 'all the time'. Patients perceived their biggest problem in life to be their loss of independence caused by the disease; whereas their family found their cognitive and affective symptoms were the most difficult with which to deal.

The patients' knowledge of the genetics of HD was poor achieving a mean of only 20.8% on their knowledge of the disease and the genetic pattern of inheritance. Their overall satisfaction with the neurogenetic clinic counselling service was rated as 'very satisfactory' (3/4) by the majority of patients for 15/19 items. Recommendations for improvements to the counselling service were made for four items (N6 'privacy when seeing the doctor', N9 'clinic environment', N14 'time you had to wait for the doctor' and N17 'clinics provided you with a lot of support') rated as 'satisfactory'.

Difficulties in attending the clinic were provided and contribute towards explaining the reasons for them frequently not attending their clinic appointments.

4.13 GENERAL NEEDS OF FAMILIES WITH HD

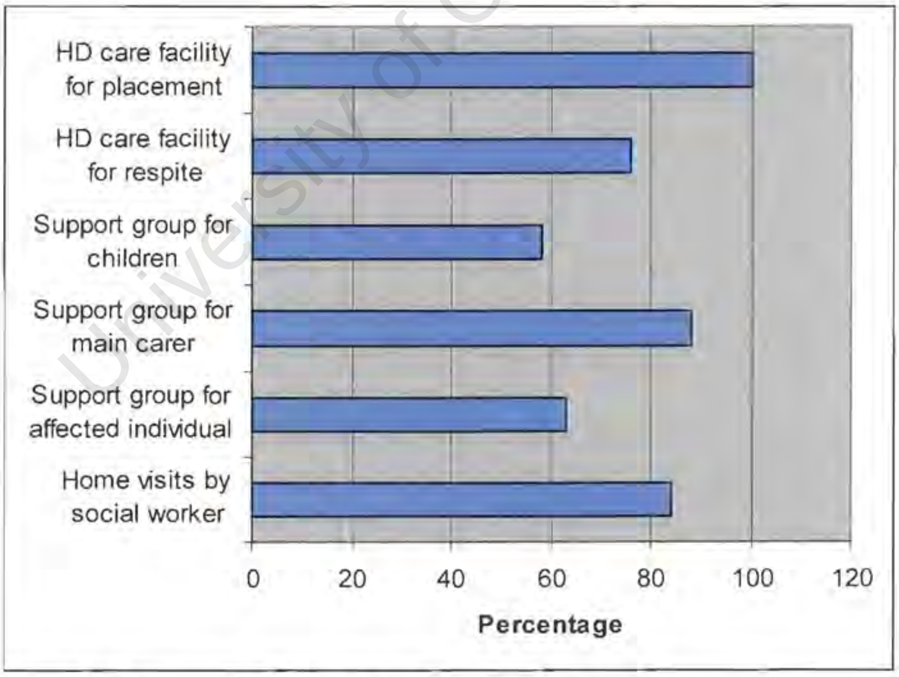

Figure 33: General needs of families with HD (n=68)

PT participants and patients were asked what they felt the general needs for families with HD in the WC were (Figure 33). Many of them and their families had experienced support from the HD Support Group when it was operational and considered that there was an urgent need for it to be revitalised. However this need can only be met if the patients and their families mobilise themselves and speak for themselves as a support group as their life experience and expertise is what is needed to achieve changes to the physical, social and attitudinal environment (DPSA 2001; Lang 2000; Leavitt 1995; Groce 1996; Marks 1996; Bickenbach et al 1999; Department of Health 1990).

With the changes in health care system most families were struggling to find suitable placement for their family members in the late stages of HD or else any placements which would accept patients with HD. This was difficult because the majority of families could not afford private fees from their grants and pensions and resulted in the carers having the burden of caring for bedridden patients who had urgent nursing needs. Although many expressed the wish for a support group where they could meet to discuss problems and share experiences, from a practical point of view, it is difficult to envisage how the majority of families would be able to attend meetings due to their financial and transport difficulties. Those who had previously attended support group meetings when it was functioning were from the higher socioeconomic group who lived in close proximity to the city and/or had their own transport.

The majority 84% (57/68) suggested that a social worker was urgently needed to visit the homes of the families with HD. Carers from the patients group maintained that when they took their affected relatives to the neurogenetic clinic they were well controlled and calm and the doctors did not realise what the families were experiencing at home. Some of the participants who had been raised in families where a parent was affected thought that they had suffered unnecessarily and that had a social worker visited and observed what their home circumstances were like, there would have been some intervention to ensure that they were safe and were receiving adequate care. Twelve percent (8/68) had been removed from their parents when they were young and were adopted or placed in foster care as their mothers were unable to care for them adequately.

4.14 CAREGIVERS

There were 38 carers of whom 13% (5/38) were males and 87% (33/38) females. Three percent (26/38), 2 males and 8 females, had to leave their jobs in order to stay at home to take care of their affected relative. Their age distribution is presented in Figure 34. The mean age was 48.7 years. All carers lived in the same house as the patient. At the time of the initial interview only 7.8% (3/38) caregivers received a grant-in-aid for caring for the patient.

Figure 34: Age distribution of the caregivers (n=38)

The relationship of the carer to the patient and the number of years of caring for the patient is presented in Table 72.

Number of years of caring	Partner	Child	Sibling	Parent	Friend	Total
1	0	0	0	0	1	1
2	2	1	0	1	0	4
3	4	1	0	1	0	6
4	1	2	1	0	0	4
5	2	3	1	1	0	7
6	2	0	1	1	0	4
7	2	1	0	0	0	3
10	1	0	0	0	1	2
11	1	0	0	0	0	1
13	1	0	0	0	0	1
14	1	3	0	0	0	4
15	1	0	0	0	0	1
Total	**18**	**11**	**3**	**4**	**2**	**38**

Table 72: Relationship of carer with patient and years of caring

The number of years of caring for the patient is presented in Figure 35 and the number of hours per day of caring in Table 73. At the time of the first interview 93.1% (36/38) of the carers were involved in caring activities for more than 12 hours per day. The mean number of hours that they spent in actual caring tasks, supervising or having to be available for the patient, was 23 hours.

Figure 35: Number of years of caring for patient

		Hours				Total
		2	**12**	**16**	**24**	
Caregiver	Male	1	0	1	4	6
	Female	0	1	0	31	32
Total		**1**	**1**	**1**	**35**	**38**

Table 73: Number of hours caregivers spent caring for affected Relative (n=38)

As shown in Table 74 the nature and frequency of the tasks with which they assisted the patients involved a great deal of physical effort and emotional strain. Although they did not always have to assist the patient they had to be available at all times to supervise the tasks by prompting the sequence of the activity or checking the standard of performance. In addition, this required a great deal of patience and frequently meant the carer had to assist or redo tasks causing outbursts of anger from the patient.

	Not at all 1	Supervise only 2	Seldom 3	Sometimes 4	Often 5	Always 6
U11 Eating	0	18	1	4	0	15
U12 Bathing/showering	0	21	1	2	0	14
U13 Personal hygiene	0	21	0	2	1	14
U14 Getting on and off toilet	7	18	0	3	0	10
U15 Dressing	2	18	0	6	3	9
U16 Getting in and out of bed	7	17	1	3	0	10
U18 Giving medication	5	3	0	0	1	29
U19 Doing errands	2	11	0	1	2	22
U20 Walking	8	9	2	1	2	16
U21 Shopping	0	9	0	1	3	25
U22 Housework	0	8	0	0	2	28
U23 Preparing meals and cooking	1	7	0	0	2	28
U24 Laundry	0	7	0	0	2	29
U26 Listening and talking	0	7	0	0	2	29

Table 74: Nature and frequency of the rating of the tasks with which the carer assisted the disabled person (n=38)

Table 75 gives the ratings of the carers on items referring to the level of burden they experienced.

Items	1 Not at all	2 Seldom	3 Sometimes	4 Often
General strain				
U31 Do you find yourself facing practical problems in the care of your relative that you think are difficult to solve?	5	1	8	24
U32 Do you think you have to take too much responsibility for your relative's welfare?	15	3	3	17
U33 Do you sometimes feel as if you would like to run away from the entire situation you find yourself in?	14	3	2	19
U34 Do you feel tired and worn out?	14	9	2	13
U35 Do you feel tied down by your relative's problems?	10	3	3	22
U36 Do you find it mentally tiring to take care of your relative?	10	0	4	24
U37 Do you think your own health has suffered because you have been taking care of your relative?	28	1	0	9
U38 Do you think you spend so much time with your relative that you do not have enough time for yourself?	12	6	6	14
Isolation				
U39 Do you avoid inviting friends and acquaintances home because of your relative's problem?	16	3	6	13
U40 Has your social life, e.g. with family and friends, been reduced?	9	2	2	25
U41 Has your relative's problem prevented you from doing what you had planned to do in this phase of your life?	18	2	1	17
Disappointment				
U42 Do you feel that life has treated you unfairly?	22	0	1	15
U43 Did you expect that your life would be different from what it is at your age?	17	1	2	18
U44 Do you feel lonely and isolated because of your relative's problem?	17	5	2	14
U45 Do you find it physically tiring to take care of your relative?	22	5	1	10
U46Have you had to make financial sacrifices because you have been taking care of your relative?	12	2	5	19
Emotional involvement				
U47 Are you sometimes ashamed or embarrassed by your relative's behaviour?	14	5	4	15
U48 Do you ever feel hurt and angry with your relative?	11	5	0	22
U49 Do you feel embarrassed by your relative's behaviour?	17	3	5	13
Environment				
U50 Does the physical environment make it difficult for you to take care of your relative?	24	0	1	13
U51 Do you worry about not taking care of your relative in the proper way?	15	0	2	21
U52 Is there anything in the neighbourhood which makes it difficult for you to take care of your relative?	28	0	0	10

Table 75: Caregivers' rating of level of burden (CBS) (n=38)

These CBS ratings justify a re-scaling of the four rating categories into two: 'none to seldom' and 'sometimes to often'. The cross-tabulation of hours per day of caring and CS are presented in Table 76.

		Hours per day				
		2	12	16	24	Total
Burden	**Scale**					
General Strain	none/seldom	0	0	0	14	14
	sometimes/often	1	2	1	20	24
Isolation	none/seldom	0	0	1	17	18
	sometimes/often	1	2	0	17	20
Disappointment	none/seldom	1	2	1	17	21
	sometimes/often	0	0	0	17	17
Emotional Involvement	none/seldom	0	0	1	17	18
	sometimes/often	1	2	0	17	20
Environment	none/seldom	1	1	1	24	27
	sometimes/often	0	1	0	10	11

Table 76: Hours per day of caring cross-tabulated with CBS (n=38)

It is very evident that those carers whose caring tasks involved less than 16 hours per day were not particularly affected by any burden. However, those whose caring involved 24 hours experienced considerable strain, isolation, disappointment and emotional involvement. These findings are corroborated by other authors (Taylor et al 1995; Askham 1992; Hayden et al 1980c). It is interesting to note that, in spite of the difficult housing and accessibility circumstances under which the majority of families lived, only 28.9% of the carers found their environment to be a burden.

When asked what their main problems were as carers they expressed their needs as presented in Figure 36.

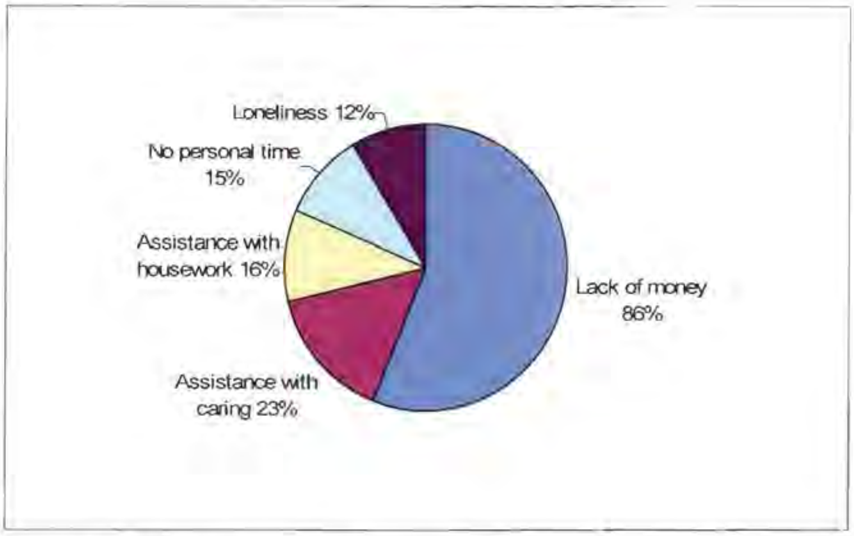

Figure 36: Needs expressed by carers

The 7.3% (3/38) carers who were <20 years (Figure 36) were all females who had been caring for their mothers from an early age. They were resentful about being expected to be the sole full-time carers while there were none of these expectations of their male and younger sister siblings who were able to leave home at will and lead normal teenagers' lives. This is confirmed by other studies (Forrest Keenan et al 2007; Siskowski 2006; Downing 2005; National Alliance for Caregiving 2005). On the other hand spouses, on the whole, seemed affected very little by their association with HD and its concomitant responsibilities of caring which is similar to the findings of Helder et al (2002).

"As I say she and I have been together for thirty years so I must look after her because what must she do if I don't do it for her. It is very difficult sometimes when she gets stubborn or when I have to do her personal washing and things like that. But that is what I've got to do and I must just get on with it and do it".

"When we first heard that he had the disease we were both terribly upset and we sat down and spoke to it with the Lord and asked him for guidance. Once we had worked out that He wanted us to be together well I just do everything possible to make things easier for my husband and look after him as best I can. It is not always easy, but then I think of what the Lord's expectations of me is and I am thankful for the fact that we have ourselves and our children".

"She has had the sickness for many years now and it's just a fact of life that we have to get on with it the best we can".

There were several spouses who expressed resentment about their husband's families who seldom came to visit the affected individual and never provided any form of support for the caring spouse in spite of having lived with affected relatives and personally having experienced the difficulties of caring for deceased relatives affected with HD. They were particularly resentful about the spouse's parent with regards to this aspect. Others complained that their adult children appeared to stay away and avoid the fact that they could or should assist the carer in any practical or emotional way. This avoidance of contact with affected relatives was also found in research on caregivers of HD by Lowit and Van Teijlingen (2005).

4.15 SUMMARY OF CAREGIVERS' PROFILE

Caregiving tasks have a major impact on the lives of the carers of patients with HD. Many of them had to give up jobs in order to stay at home to care for their relatives. This increased their level of poverty as the grant-in-aid which some of them received was less than their wages earned saving the state a great deal of money by not having to pay for the patient's care in an institution. Others had no form of income once they left their jobs and were reliant on the patient's DG.

The nature and responsibilities of the tasks were onerous as the average age of the carers was 48.7 years and the number of hours that they spent in caring for their relatives was 23 hours per day.

Caregivers whose hours of caring were <16 did not report a significant amount of burden, but those who were responsible for and who had to care for relatives for 24 hours per day experienced a considerable level of burden.

The major problem experienced by the caregivers was lack of money.

4.16 ADVICE AND ASSISTANCE PROVIDED TO PATIENTS AND THEIR FAMILIES DURING HOME VISITS

During the follow-up visits the discussions mostly revolved around the problems the caregivers were encountering with regards to the care of their affected relatives. These have to be seen within the context of families with HD living in extreme poverty, who have no medical, nursing, therapy or social support services provided in the form of home visits or respite centres. As the researcher had had 40 years of experience of working in multidisciplinary rehabilitation therapy teams as a physiotherapist, she was able to provide advice and assistance with many of their day-to-day non-pharmacological management problems. Consecutive follow-up visits allowed her to confirm that the advice was assisting them and enabling the patients to be optimally independent, thereby decreasing caregiver burden. Table 77 shows the types and frequency of advice and tasks with which the patients and their families required assistance. These itmes were not mutually exclusive.

Tasks and advice	Frequency	Percentage
Safe smoking habits	18	43.2
Reassurance of carer	17	41.5
Nutrition	15	36.6
Eating and swallowing	15	36.6
Application for social grants	11	26.8
Avoidance of aggressive outbursts	11	26.8
Exercise	8	19.6
Referral to neurologist, psychiatrist	7	17.1
Suitable clothing for independence	5	12.2
Letters of advice on patient's diagnosis	5	12.2
School fees rebates	3	7.3
Removal of walking frames and crutches	3	7.3
Guns to be handed in at police station	2	4.8
Corporal punishment of children	2	4.8

Table 77: Types of advice and assistance requested during home visits

The researcher referred patients and their families to the social workers at their local Community Health Centres to ensure that their applications for the social grants to which they were entitled were processed and that they did actually receive them. On two occasions this entailed helping the carers to apply for their identity documents before they could apply for their grants-in-aid.

Three families had children who had just had birthdays and were no longer entitled to the children's grant which meant that they had to meet the school fees themselves. This was approximately R350 per year in addition to the normal school requirements of books, writing materials and other daily expenses. The researcher was able to write to the principals of the schools explaining the nature of HD and that the parent was not abusing alcohol and, therefore, should be entitled to a fees rebate.

During two visits the researcher noticed that there appeared to be more children than what had originally been accounted for in the demographic data section. On further questioning it transpired that these children had been born to neighbours who were young mothers with severe alcohol abuse problems. As they were not taking care of the babies adequately the carers just took them on as though they were their own children and raised them as part of the family. They were referred to the social services to apply for a foster carer grant and child care grant to which they were entitled.

If the patient appeared to have progressed a great deal and required being seen by the neurologist for medication for poor sleeping or depression or severe chorea an appointment was made through the genetic sisters for this to be done as soon as possible.

Two of the patients reported that there was a great deal of domestic violence that was occurring in the families, mostly between themselves and their young adult children. After a lengthy discussion the researcher queried whether they had any firearms in the house as she could foresee real harm being done if that were the case. They both acknowledged that they had unlicensed firearms to protect themselves and their families from gangsters in the area, but when the researcher explained that, with their severity of chorea, they would not be able to use the firearm and that their children would be able to overpower them and use it on them during a bout of family violence, they agreed to hand over these weapons to the police station. It was fortunately during a period of firearm amnesty so there were no questions asked and they were provided with receipts for the transactions.

Two fathers admitted to giving their young teenage girls corporal punishment when "they were doing something that wasn't good" and the researcher observed their wives being very distressed during this discussion. The researcher arranged for them to be seen by the psychiatrist immediately for assessment and appropriate management. The final outcome for one of these families was that the spouse was given medication which worked very effectively to control his aggressive outbursts. In the other case the wife was advised to leave her husband for the children's safety as her husband's family provided her with no support in spite of living one block away and her own family lived in another province. The wife was severely depressed and had been living in constant fear of her husband's next attack on her or her daughters who were petrified of him and were living very dysfunctional lives by trying to avoid being at home with him when their mother was at work.

4.17 PARTICIPANTS' KNOWLEDGE OF HIV/AIDS

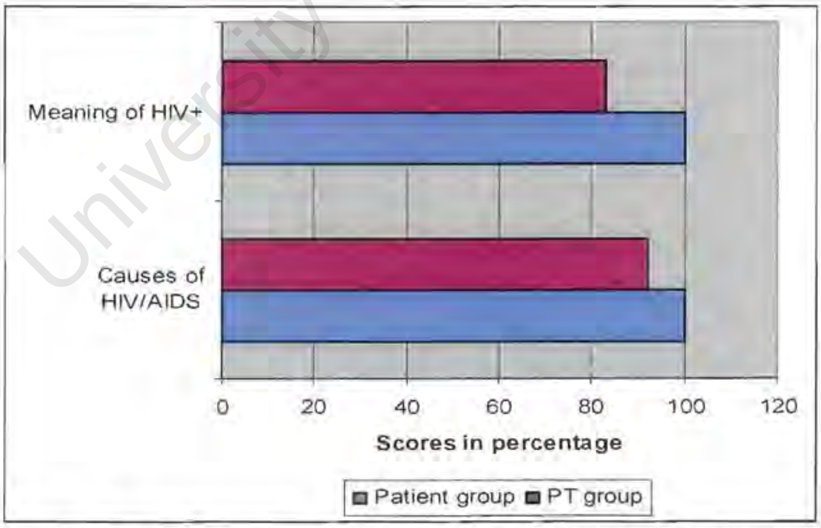

Figure 37: A comparison of the level of knowledge of HIV/AIDS between the PT group (n=27) and the patient group (n=41)

Figure 37 shows that the PT participants were fully informed about HIV/AIDS. The majority could name three or four causes and understood what HIV-positive signified. Eight percent (3/41) of the patient group, on the other hand, did not have any idea of the cause and only a few could cite more than one cause. Seventeen percent (7/41) thought that being HIV-positive was a pleasing result as it meant that the person did not have the HIV virus. It appears that these people associated the word positive with the pictures of a healthy diet together with a HIV+ sign on the posters in the clinic and associated this with a positive result. Twenty-one percent (9/41) were confused between the HD confirmatory test they had undergone at the neurogenetic clinic and believed it was a bloodtest for their HIV status.

The sources from which the participants obtained their information is presented in Figure 38. The categories are not mutually exclusive.

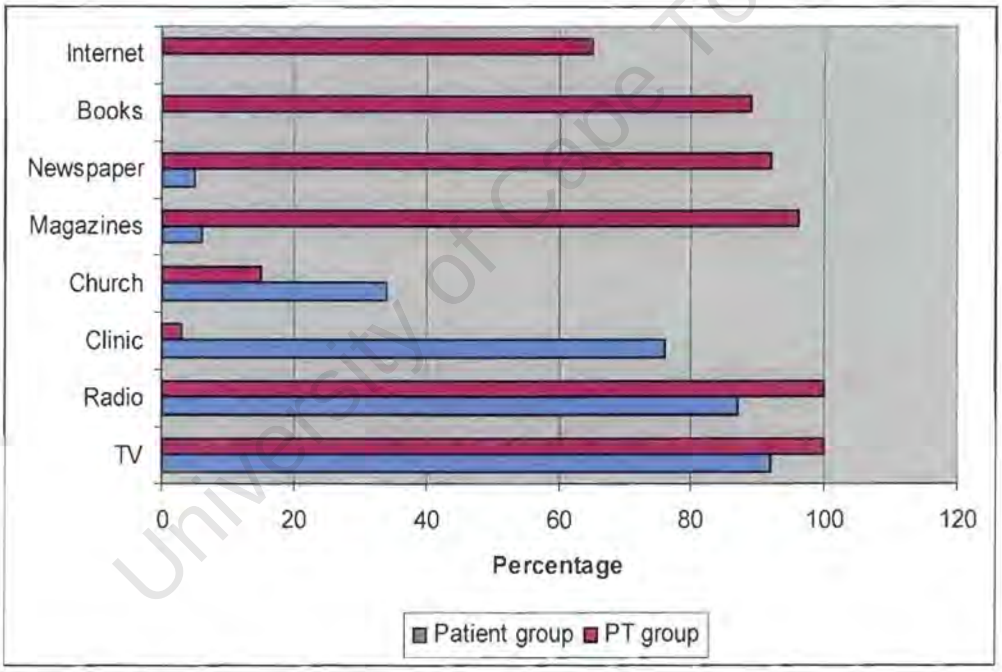

Figure 38: Information sources for HIV/AIDS (PT group n=27, patient group n=41)

It is very clear to see that the PT group obtained their information from sources requiring a higher educational level, whereas the majority of the patient group relied on sources which did not require any form of reading.

The major findings and implications of the study will be summarised in the conclusions in Chapter Five.

CHAPTER FIVE

CONCLUSIONS

Although the prevalence of HD is low and it might not be considered to be one of the priorities in the health system it has substantial implications for the immediate families and future generations of the affected individuals. Because of these broad repercussions in families with HD there has been a vast amount of research published on HD in the UK, Europe, USA, Australia and Canada. The literature contains extensive data on the prevalence and age at onset; genetics; molecular biology, mitotic stability and meiotic variability of the $(CAG)_n$; neuropathology; psychopathological, cognitive and motor changes associated with the symptoms; PT, psychological consequences of PT and adverse affects of PT testing; and pharmacological and non-pharmacological management of the symptoms of HD. These have largely been studied independently of the psychosocial and cultural factors that may be affecting the individuals and their families and which tend to cause discrimination, stigmatisation and marginalisation.

There have been a few studies of the prevalence in South Africa (Greenberg 1990; Hayden et al 1980a and 1980b), but no research has been published on the social profiles of either the patients affected with HD or at-risk individuals who have undergone PT. This research is the first to study HD from a social model framework, namely, the aspects of society which impact negatively on the individuals with HD and their families. Previous research has been based on the medical model which centres on pathology and is not adequate in providing assistance to individuals with this condition. The complexity of HD requires a social model approach whereby the social barriers which impact on the affected individuals' and their families' quality of life and the social support mechanisms necessary to ameliorate these obstacles are considered and researched. The focus has to be placed on reducing the negative impact of the disease through changes in social policy and institutional practices and can only be brought about by identifying the day-to-day needs of the patients and their families and evaluating the extent to which they impact negatively on their lives in their home environment. This was particularly so in this study as the majority of the cohort are from mixed ancestry and live in historically disadvantaged communities.

It was indispensable for this research to be conducted in the homes of the PT participants, HD patients and caregivers. It provided an opportunity to observe the conditions under which they were living so that the researcher could elicit questions which gave her a deeper insight into the problems which confronted them. This could not have been achieved in a hospital or clinic as the housing and social environment could not be evaluated accurately by participant reports of their social conditions. Thus it was essential for the researcher to first become knowledgeable about the low-income suburbs with regard to their history, demographic features, socio-economic situation of the general population, physical environment, housing, nature of crime, community facilities and services offered. This provided important information about the patients with regard to the historical events which had influenced their development and the environmental conditions which affected their lives and that of their families at the time of the research.

The aims of this research were firstly to evaluate the effectiveness of the genetic counselling processes of the PT programme and neurogenetic clinic from the user's perspective; secondly to identify the components and determinants which ensure that the genetic counselling services are evaluated positively; thirdly to implement changes and additions to these services as recommended by the users; fourthly to explore the nature and extent of the social barriers experienced by the users of the PT programme and neurogenetic clinic; and fifthly to improve the quality of life of the families of symptomatic patients by assisting them with the management of their affected relatives and providing them with psychological support.

This study reports on the results for PT at UCT over 11 years of direct mutation testing from April 1995 to April 2006. The data were generally similar to those of international studies apart from the lower uptake rate and substantially increased frequency of major adverse reactions immediately after the test disclosure and one year after PT. The participants' level of knowledge of HD was excellent with regard to recall and understanding which is part of the aims of genetic counselling. The majority of participants evaluated the PT programme as being at least 'very satisfactory' and was satisfied with the majority of the aspects of the programme. The determinants of satisfaction were found to be 'the quality of information' provided and the 'amount of empathy' displayed by the genetic counselling team members. Only two items, D17 'follow-up visits provided a lot of support' and D7 'the counselling helped you to cope better with your problems', were rated as 'very unsatisfactory' and 'unsatisfactory' respectively. The participants recommended modifications to address these items (Chapter Six, page 266), amongst which was post-test counselling. When implemented these ratings could be increased to 5/5 'highly satisfactory' which is what a centre of excellence such as the Division of Human Genetics aspires to be and is expected from it. This will also ensure that the PT programme conforms to the practice and recommendations of other international centres (Blain and Brooks 2007; Tassicker et al 2006; Davey et al 2005; Van Riper 2005; Myers 2004; Sarangi et al 2004; Shannon 2004; Taylor 2004; Hayden et al 2001). Bearing in mind that there are an average of only 3 counsellees per year these recommendations should not be a significant challenge to or be a drain on the present resources and may assist in decreasing the frequency of depression in participants after PT. Generally there were few social problems experienced by the PT group apart from 3/27 individuals who had developed symptoms and were dependent on the state DG.

The clinical manifestations of the patients were similar to that described in the literature. The type and level of support provided by the carers and family members was described as being available to them 'all' or 'most of the time' in spite of them finding their relatives' cognitive and affective impairment to be the most burdensome aspects with which to cope. The patients' and caregivers' knowledge of the genetics of HD was poor in spite of having attended the neurogenetic clinic on at least four occasions. Their overall satisfaction with the neurogenetic clinic counselling service was rated as 'very satisfactory' (3/4) by the majority of participants. The determinants of satisfaction with the neurogenetic clinic sounselling were found to be the amount of caring, empathy and concern displayed by the staff. Recommendations for improvements to the service made by the patients and caregivers are provided in the next chapter.

This is the first time that an inventory of all the factors contributing to the social barriers of patients and families with HD have been documented and data have been collected on the extent to which each of the factors affected the research cohort of families with HD. Without clear documentation of what is effective and ineffective, family and caregivers will continue to face their problems without guidance and health care providers will not be able to provide anticipatory guidance to facilitate each transition during this progressive disease (Moskowitz and Marder, 2001). The majority of the patient group was from historically disadvantaged communities and was still bearing the consequences of the 'apartheid' system with its cornerstone of an inferior education system for Black and Coloured ethnic groups (Kallway 2002). The factors identified are common in low-income suburbs in South Africa, namely, low level of education; poverty; transport; housing; safety; accessibility of health and social services; physical and emotional abuse; and their disability. The housing conditions contributed significantly to the hardships endured by the patients and their families. The problems experienced with their housing demonstrates that the housing of many of the disabled do not meet the basic criteria of what a "house" requires in order to comply with the Reconciliation and Development Programme definition (ANC 1994) and that the social security net is not alleviating the poverty of the most vulnerable of the population. The majority of participants found the amount of the DG as being a problem to them and that it affected their ability to meet basic food, clothing, fuel and rental costs. It also had a major impact on their ability to pay for transport which, in turn, impacted on their access to health services. Although they had different special needs relating to their disabilities and different responsibilities with regard to the number of dependents they had to support they all received the same amount of DG. There is urgency for social security to be addressed in order to alleviate the poverty of the disabled. It should ensure that all beneficiaries of the DG are reached and that, in the cases where disabled have dependent children their child support grants should continue for them until they have completed their education and become self-supporting. The amount of the grant-in-aid (180 per month) also impacted negatively on the family when the caregiver had to leave his/her employment to care for the disabled relative as it was only a portion of what they received when they were working in the open market. The Integrated National Disability Strategy (1997) claims that the disabled have a minimum livelihood provided by means of social benefits. However, this is not possible on R820 per month with their living expenses and social responsibilities toward their families and they become further involved in the cycle of poverty. The cycle of poverty with the patients and families with HD appears to be unavoidable and inevitable for future generations unless the BIG is introduced when individuals will be able to receive a basic income grant and will be able to remain at school to complete their matriculation certification and have access to employment in higher income categories than at present. This can only happen if, in addition, the state provides facilities for the care of patients with HD once they are unable to perform activities of daily living or require constant supervision during the daytime to free the caregivers to return to employment. With the funds that it will require to address these issues no one expects that the facilities and resources the patients with HD need can be met overnight. However, planned change efforts are essential if their basic rights are to be met and their quality of life is to improve. To date, none of the legislation, policies and white papers regarding people with disabilities (Disability Rights Charter 2000; White Paper on an INDS 1997; Bill of rights Section 9 and 28 1996) have been implemented in the Cape Flats and this is nine years down the line from the time they were legislated by the relevant government departments. There is a total absence of formal support services accessible to them in the community clinics which they could attend periodically for assistance with mobility, feeding, swallowing, communication and activities of daily living.

As the mean number of children of patients was twice that of the PT participants in this study, hopefully with improved education levels in the future many of the families with HD will limit their number of children to affordable numbers and, with increased knowledge of HD, might present for PT before having children which will then reflect a higher uptake of PT than at present. An increased level of education will also allow greater opportunities for employment and in this way decrease the number of social barriers with which they are currently confronted.

Caregivers found the responsibility and caring for their relatives to be an onerous task which was made more burdensome by the lack of money. This was particularly so if they had to leave their employment to take on the caregiving role which placed their families at an even greater disadvantage with regard to their general living costs. They also had the concern that they would have to care for their relatives without any respite and with the anxiety that they would not be able to afford placing them in appropriate institutional care once they required proper nursing care.

The home visits proved most beneficial as a means of ensuring that the families were receiving all the social grants to which they were entitled. Many of the carers also needed reassurance that their relatives' deterioration was not due to neglect and that they were doing their best to care for them. The majority needed some form of assistance with the management of their relatives with regard to seating, nutrition, eating and swallowing and general daily exercise. They appeared to benefit by discussing their relatives' aggression and the need for daily routine and communication. It was important to refer some of the caregivers to the appropriate specialists for the management of their relatives' psychiatric conditions which they had not discussed previously when attending the neurogenetic clinic.

At present the management of HD is provided within the medical model framework where HD is perceived to have its locus in the individual and, in the majority of cases, medical intervention is palliative, seeking to control symptoms or make them less severe, but not being able to offer a cure.

Consequently, maximising the welfare of these individuals means maintaining or improving their quality of daily life, rather than attempting to eradicate the disease process itself (Ross and Deverell 2004). The neurologists at the clinic prescribe medication to attempt to alleviate the symptoms of HD, but once patients return home their caregivers are expected to cope and function in their homes and communities on their own without any form of health and social service support. However, according to the social model that is precisely where and when they need the most assistance with HD as their needs at this stage are not health-related. They should be the responsibility of the departments of social services, housing, transport, roads and sewerage, labour and education as it is in these domains that they and their families experience great difficulties rather than only with their disabilities.

The general needs of the individuals and the families with HD were care facilities provided by the state where affected patients could be placed when the families could not longer cope with their nursing requirements. Home visits by a social worker were considered to be essential by more than 80% of the cohort. This was not only for assistance with social grant applications, but also for assessing and advising on the management of the children when an affected parent was unable to care for them adequately and/or they were being abused by the parent and/or members of the family or community. They also have an urgent need for an active social support group to which the carers, in particular, could turn to for support and advice. Thus, once more, the problems experienced by families with HD would be relieved to a great extent if their disease was managed by a social model of health rather than the medical model which can provide them with little in the management of the onset of the disease or their symptoms once the disease manifests itself, but social support services could provide the essential support needed in order to ensure an improved quality of life for the patients as well as their families.

CHAPTER SIX

RECOMMENDATIONS

After the data had been analysed and the findings completed the recommendations made by the PT participants; patients and caregivers; and the researcher were discussed with the genetic counselling team members and neurogenetic clinic staff. As described below the majority of the suggestions to improve the respective programmes were approved and were or were being implemented before the write-up of this thesis had been completed.

Recommendations for improving the PT programme made by the participants were as follows:

1. More psychological counselling sessions during the PT programme. In retrospect some of the participants realised that they had needed counselling to overcome their issues of being raised in a family with an affected parent before undergoing the testing. As this recommendation would have a significant impact on the resources of the genetic counselling team and the majority of the individuals are covered by medical insurance, the researcher recommends that at the first counselling session the genetic counsellor advise them to consult a private psychologist for counselling to assist with their longstanding psychological problems. This has been implemented.

2. A legal consultant to be included in the genetic team. Participants expressed their anxiety experienced during the PT programme when they realised that their and their children's medical and life insurance, and access to housing bonds may be affected, compromised or refused. They considered it essential to have a consultant conversant with the implications of genetic disorders and insurance with whom they could discuss their financial and legal concerns. This request has

already been put into practice. A legal consultant is in the process of drawing up an information manual which addresses these issues in context of the present legislation of the country. She is also able to refer individuals with specific concerns to appropriate legal consultants.

3. Change of venue. Due to the psychological impact of past memories of attending the hospital clinic with affected relatives or the impact of observing patients in advanced stages of HD for the first time, participants requested that the clinic venue be changed so that they do not have to have contact with patients and are not part of the hospital environment. The venue has subsequently been changed from the hospital clinic to an office in the Faculty of Health Sciences away from the hospital.

4. Change of format of results session. Many participants described that they were given their results and then simply abandoned by the genetic team. Others expressed their feelings of numbness and that they were unable to think of questions they wished to ask immediately after hearing the result. These experiences were expressed by participants receiving both gene-positive and –negative outcomes. They requested that they be given a short recess alone with their support partner and then see a member of the genetic counselling team to discuss any queries or concerns. This has been put into practice.

5. Post-test follow-up counselling. Participants conveyed the need for essential post-test counselling sessions as part of the PT programme. Many of them felt that had they had these they might not have developed depression as they would have received professional support by people who understood their personal situation and the implications it might have on their families. They also expressed the need for these sessions to include their spouses/partners and children if necessary. These post-test counselling sessions are now part of the

PT programme. They are continued for one year after the test disclosure.

The following suggestions were made by the patients and their caregivers for improving the neurogenetic clinic genetic counselling service:

6. Written information about HD in Afrikaans and at al level they would understand with their low levels of formal education. A booklet in Afrikaans has been written for this purpose (Appendix viii).

7. Caregivers to be given the opportunity of consulting the neurologist separately to discuss the management of difficult psychiatric symptoms and obsessive-compulsive tendencies of sexual disinhibition which would not be appropriate to discuss in the presence of the patient. This has been implemented by having a genetic counsellor complete the HDQoL-C questionnaire with the carer. This gives them the opportunity of discussing these issues on their own rather than in the presence of the patient and without rousing the patient's suspicions about the nature of a private conversation between the care and the genetic counsellor. Based on this discussion the genetic nurses arrange for the caregiver to see the neurologist in a private consultation immediately before the patient joins them. During this time the genetic nurse arranges further referrals or appointments with the patient.

8. A health worker who they can spend time with and talk to at the clinic regarding their day-to-day problems at home to ensure that they are doing everything they can for their relative's management. This is related to guidance with feeding, choking, dressing, bathing and other activities of daily living as well as the difficult behaviour of their affected relatives. This aspect has been implemented.

The researcher's recommendations are:

9. A review of the HD protocol distributed to individuals requesting PT. The protocol was developed several years ago and incremental changes have taken place and are understood by the staff, but have not been formalised on the information leaflet. This has been revised (Appendix ix).

10. Individuals requesting PT are provided with a post-test counselling programme for one year after the result has been disclosed in order to comply with international practice and recommended by the UK Huntington's Prediction Consortium (Craufurd and Tyler 1992) in an attempt to minimise the levels of depression reported in the participants in this study. Berkenstadt et al (1999) and Shiloh et al (1997) found that using perceived personal control as an outcome of genetic counselling provided the participants with the ability to cope with stressful events in a manner that reduces its perceived threat; the ability to alter the physical characteristics of the threatening event so that its actual threat is reduced; and the opportunity to choose amongst options. This should be done at monthly intervals post-test to measure how effectively the individual is coping with their particular situation. Post-test counselling sessions have been introduced as part of the predictive testing programme for HD and all new participants have since been offered these additional sessions as part of the predictive testing programme.

11. The individuals should complete an HAD form at each post-test counseling session and, if they are found to be depressed, should be referred for further psychological counselling.

12. Standard letters on hospital headed paper be available at clinic visits and be issued to patients if they are experiencing difficulties with community people reacting to them as though they are intoxicated. This has been put into practice (Appendix x).

13. For information on the inheritance pattern, availability of PT, contact numbers for the service, advice on non-pharmacological management of HD to be published in Afrikaans at a literacy level which the majority of families would be able to read and understand. This has been completed (Appendix viii) and is being reviewed with the families attending the neurogenetic clinic and by the researcher when she does follow-up visits to inform the study participants of the outcomes of the research to which they contributed.

14. The patient and caregiver receive additional genetic counselling at each follow-up session of clinic attendance to clarify any misunderstandings, repeat content of counselling sessions and consolidate their knowledge of the inheritance pattern of HD. According to Krupst et al (1975) patient restatement with feedback from the counsellor is the most effective means of increasing long-term recall. It is possible that by implementing greater emphasis on regular counselling sessions at each clinic visit more presymptomatic school leavers will present for predictive testing before they enter a cycle of early unplanned pregnancies with little knowledge of the inheritance pattern of the disease. This has been implemented.

15. In order to level the playing fields and provide the socio-economically disadvantaged families with the same opportunities of accessibility to PT, these individuals should have their transport costs paid by a bequest fund for HD available in the Division. When these individuals request PT they should be informed that their transport costs for the six sessions of pre-test counselling and the post-test counselling sessions for a year will be paid for by the fund. This has already been put into practice.

16. A staff member spends time with each patient and carer when they attend neurogenetic clinics to explore their day-to-day difficulties and assist them with non-pharmacological approaches in dealing with

them in order to reduce the burden of care and to ensure the patient is functioning at an optimal level. This is in operation.

17. More time devoted to the caregivers at the neurogenetic clinics. The caregivers' needs are as important as those of the patient, yet they often do not receive the attention they deserve as there is so much time spent on the patients' needs. This is being addressed by having the genetic counsellor complete the HDQoL-C with the caregiver and providing them with the opportunity of someone listening and showing them caring. If necessary, they are seen by the doctor separately from the patients, based on the counsellor's findings during her/his interview.

18. Future research is conducted on the PT programme after 10 further individuals have completed the pre- and post-test genetic counselling sessions to evaluate whether the protocol changes brought about by the recommendations of the present research have been positively appraised.

19. Re-evaluation of the clinical genetic counseling service be done after a year of the recommendations being implemented.

20. Family pedigrees of the larger families in the Western Cape to be updated. Special attention should be given to including "voorkinders", children born before the marriage, as the participants' responses frequently only included the children born within a marriage and omitted others born before marriage or out of wedlock, yet they are also at-risk for HD, but are unknown to the neurogenetic clinic.

21. The HDQoL-C scale be translated into Afrikaans and validated in collaboration with the authors (Aubeeluck and Buchanan 2007) who developed it and have kindly made it available for use at the GSH neurogenetic clinic.

22. A cost-benefit analysis of social care economics be performed whereby home visits by a primary health care nurse acquainted with HD and the management of its symptoms, are compared to the costs of transport and distress undergone by caregivers attending routine visits when their affected relatives are not experiencing any specific medical problems.

23. There is an urgent need for a social worker to provide home visits to assess the circumstance of the family members, especially the children, and to ensure they are receiving all social benefits to which they are entitled. This will need additional resources and funding for such a post will need to be found. This is currently being sought from international funders.

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UNIVERSITY OF CAPE TOWN PREDICTIVE TESTING PROTOCOL FOR HUNTINGTON DIEASE

The predictive test for Huntington disease is a new procedure which allows an individual at risk for having inherited the gene for Huntington disease a chance of knowing whether he or she has the affected gene, before the clinical signs present themselves.

To take the test is a very serious decision. Therefore, it is important that you are well informed and understand the programme and procedures you will have to go through to finally get the result. A programme or protocol has been recommended by doctors and geneticists based on their experiences in dealing with individuals and families at risk for the disease.

If, after careful consideration, you decide to take the test, you will be requested to come to the Groote Schuur Hospital Neuro Genetic Clinic or the Department of Human Genetics on at least four occasions to see the doctors involved in running the programme.

FIRST MEETING (NEUROLOGIST, GENETICIST AND GENETIC NURSE IN ATTENDANCE)

The neurologist will perform a clinical and neurological examination. The doctor and geneticist will explain in detail the nature and requirements of the test which are as follows:-

1. As you know, the affected gene for Huntington disease has been found and we are now able to offer a blood test that will predict whether or not you will get the disease.

   ***IMPORTANT:** It is very important at the beginning that you understand that there is a small possibility that the test may not show a clear result with absolute certainty. If this occurs in your case, then unfortunately your risk of carrying the gene remains exactly the same, i.e. an even chance (a 50% chance of carrying the gene or not), as before you entered the programme.* Therefore, the final result of your test could be one of three:

   * *Positive* - you have the affected gene and will develop the symptoms at some stage in the future. We have no way of knowing when and how it will present.

- *Negative* - based on our current knowledge of this gene, you will not develop Huntington disease.

- *Uninterpretable* - no result. You are still at risk and have a chance of carrying the affected gene.

2. There is currently no additional charge for the blood test to GSH patients at the Groote Schuur Hospital Clinic. Costs will be discussed for private patients and those with medical aid.

3. We strongly recommend that you inform your family doctor of your decision to undertake the test. If you do not have a family doctor we recommend you find one. As part of the policy of this programme, we believe that the on-going medical care and support your doctor is able to give you is very important to you and your family. A letter will be sent to your doctor to inform him/her about the programme after the third visit.

4. Support is essential and we therefore strongly advise that you choose a family member or a trusted friend to accompany you to all the meetings.

5. The final result will be given to you approximately two months after your blood samples have been taken and will be strictly confidential. No result will be given to you by telephone. Written confirmation of your result will be sent to the genetic centre that referred you. With your permission, your family doctor will be contacted and given the result.

6. If at any stage in the programme you decide you do not wish to continue, the decision is entirely yours. Your decision will in no way prejudice our relationship with you or your family. We will be happy to continue to offer you all the support and help you need.

SECOND MEETING (PSYCHOLOGIST IN ATTENDANCE)

1. You must be accompanied by your support partner or trusted friend to this meeting.

2. A psychologist will make an assessment to exclude any serious psychological problems.

3. During this meeting the psychologist will discuss your reasons for requesting predictive testing as well as the possible effects that a positive or negative result may have on you, your partner and your family. Counselling may vary widely. For example, people who have the gene may suffer a sense of shock and grief however well they may have been prepared beforehand. People whose risk is negative may feel relief but at the same time suffer guilt and anxiety. For those with no result their feelings could be anger and frustration.

4. You will be asked to make an appointment to see the geneticist approximately one month after this meeting .

5. It may be necessary to include another interview in the pre-test period if either you or we may need further information or discussion.

THIRD MEETING (GENETICIST AND GENETIC NURSE IN ATTENDANCE)

1. You must be accompanied by your chosen support partner (family member or friend).

2. At this meeting the doctors will need your written consent to take blood samples from you. Again there will be careful explanation of the test and the manner in which the final result will be given:- about two months after the blood samples have been taken you will be contacted to make an appointment to see the neurologist and psychologist when you will be given the final result. It may also be necessary to obtain blood from other close family members, with their consent.

3. Your GP will be contacted once your blood has been taken and a letter will be sent to him/her about the test. Once the result is available and has been given and discussed with you, your GP will be informed telephonically again, with your consent. No result will be given to you over the telephone, whatever the result may be.

4. Due to the complexity of the test it may take longer than two months. We cannot guarantee this. Waiting is a very difficult and anxious time - so everyone involved will try to make it as short as possible.

FOURTH MEETING (NEUROLOGIST, PSYCHOLOGIST AND GENETIC NURSE IN ATTENDANCE)

1. You will be contacted and an appointment made to finally receive your result. No result will be given over the telephone, whatever it might be. ***Please note that the genetic nurse does <u>not</u> know the result when she calls you.***

2. The doctors in attendance, and again only with your consent, will give you the final result. You must be accompanied by your chosen support partner at this meeting.

3. Once the result has been discussed with you and again, with your consent, your GP will be informed telephonically.

FOSTER CHILD GRANT

The foster child must pass the means test and there must be a court order indicating foster care status. The amount is R590 per month.

CHILD SUPPORT GRANT

Applicants must be the primary care giver of the child concerned and meet the requirements of the means test. This grant is available to children aged under seven years of age and the value is R190 per month (Government Gazette Act Nos R417 of 1998; 106 of 1997 and 59 of 1992).

WESTERN CAPE HUNTINGTON DISEASE RESEARCH PROJECT

INTERVIEW SCHEDULE

Study Code Number: ....

SECTION A DEMOGRAPHIC DATA

1. Gender:		Male ^{1} Female ^{2}	A1
2. Date of birth:		Age	A2
3. Ethnic group:		White ^{1} Mixed ^{2}	A3
4. Occupation: at present:	Previously:	Salary	A4
5. Level of education: number of years at		Junior school ^{1}	A5
		High school ^{2}	A6
		College/University ^{3}	A7
6. Why did you not continue at school or college?:			A8
7. Home language:		English ^{1} Afrikaans ^{2} Other ^{3}	A9
8. Marital status:	Single ^{1} Widow ^{2}	Married/Partner ^{3} Divorced ^{4}	A10
9. Number of children before this marriage:			A11
10. Number of children from this marriage:			A12
11. Names of children:	Date of birth:		
12. Number of grandchildren:			A13
13. Names of grandchildren:	Date of birth:		
14. Affected parent: Male ^{1}	Female ^{2} Unknown ^{3}	Unknown paternity ^{4}	A14
15. Relationship of proxy to person interviewed:	Self ^{1} Partner ^{2} Child ^{3} Relative ^{4} Parent ^{5}		A15
	Sibling ^{6} Other ^{7}: Specify		
16. Age at first visit for predictive testing:			A16
17. Number of years since completion of PT:			A17
18. Significant life event triggering presentation for predictive testing:			A18
19. Subtle symptoms observed at first visit		Yes ^{1} No ^{2}	A19
20. Symptoms developed since PT:		Yes ^{1} No ^{2}	A20
21. Year in which PT was undertaken	1995 – 2006		A21
22. Test result a) CAG repeat length	Positive ^{1} Negative ^{2}		A22
23. Describe your emotional and psychological state when you first came for predictive testing			A23

SECTION B KNOWLEDGE OF HD

1. How do people get HD? inherited/genetic disorder	Knows ^{1} Does not know ^{2}	B1
2. What part of the body does it affect?		
brain/head/nervous system	Knows ^{1} Does not know ^{2}	B2
3. What signs can you see that a person has HD?		
a) unwanted movements/slow movements/unable to move	Knows ^{1} Does not know ^{2}	B3
b) loss of memory/decreased intellectual capacity/dementia	Knows ^{1} Does not know ^{2}	B4
c) anxiety/irritability/depression/personality changes	Knows ^{1} Does not know ^{2}	B5
4. How old are people when they first start to show the signs of HD? 20-30 ^{1} 30-50 ^{2} 50-70 ^{3} Any age ^{4}	Knows ^{1} Does not know ^{2}	B6

5. Is there a cure for HD?	Knows ^{1}	Does not know ^{2}	B7
6. What is the treatment for HD?	Knows ^{1}	Does not know ^{2}	B8
7. What are the risks of your family having HD?			
a) brothers and sisters	Knows ^{1}	Does not know ^{2}	B9
b) parents	Knows ^{1}	Does not know ^{2}	B10
c) children	Knows ^{1}	Does not know ^{2}	B11
d) yourself	Knows ^{1}	Does not know ^{2}	B12
9. What kind of genetic disorder is HD?			
a) autosomal	Knows ^{1}	Does not know ^{2}	B14
b) dominant	Knows ^{1}	Does not know ^{2}	B15
11. Are males and females equally affected with HD?	Knows ^{1}	Does not know ^{2}	B17

14. Where did you learn about HD?

PREVIOUS CONTACT WITH HD

1. Who in your family has/had HD?:	At what age were they diagnosed?	

2. Have you lived with any family members with HD? C1
Who? C2
For how many years? C3

3. What effect has this/did this have on you with regard to?:						
	No problem 1	**Mild problem 2**	**Moderate problem 3**	**Severe problem 4**	**Complete problem 5**	
a) Emotional strain						C4
b) Financial strain						C5
c) Marital discord						C6
d) Family discord						C7
e) Social isolation						C8
f) Feelings of resentment						C9

4. Have you cared for a family member with HD? C11
Who? C12
For how many years? C13

5. What effect has this had on you with regard to?:						
	No problem 1	**Mild problem 2**	**Moderate problem 3**	**Severe problem 4**	**Complete problem 5**	
a) Emotional strain						C14
b) Physical strain						C15
c) Financial strain						C16
d) Coping with normal family duties						C17
e) Marital discord						C18
f) Family discord						C19
g) Physical caring of affected person						C20
h) Lack of respite						C21
i) Lack of personal time						C22
j) Deterioration of personal health						C23
k) Isolation						C24
l) Feelings of resentment						C25
m) Other: specify						C26

6a. By who and were you first told about the fact that you/your family members were at risk of HD? C27a

6b. How old were you at the time C27b

7. What was your initial response to knowing you/your family members were at risk? C28

8. What level of support did you receive from others? C29

SECTION C THE PREDICTIVE TESTING PROGRAMME

9. How did you/your family find out about the predictive testing programme? C30

10. Why did you/your family member decide to go for predictive testing? C31

11. How did you respond when you were told about the predictive testing protocol that was available? C32

13. What do you think the purpose of the testing protocol and programme are? C49

15. How many sessions did you/your family attend altogether for the testing? C51

24. Before the test did you think that your test result would be positive or negative?
+ve ^{1} -ve ^{2} ambivalent ^{3} C59

25. Who did you tell about the test result after you heard it? C60
26. Why did you tell him/her/them? C61

27. Why did you not tell everybody? : C62

28. How many months did the whole testing process take? C63

30. How did you respond when you received your/family member's test result? C65

31. Describe the period immediately after receiving the results C66

32. How did your family cope with the results? C67

33. Describe how you are coping today C68

34. What has changed in your life since you/your family member received the test result? C69

35. What aspect has been the most difficult for you with regard to the PT result? C70

36. What aspect has been the most difficult for your family? C71

37. How have the test results impacted on your plans for the future? C72

38. How would you rate your current life situation? C73

39. Do you regret having had the test done? Yes^1 No^2 C74

41. Would you advise other individuals with HD to have the test? Yes^1 No^2 C76

42. If 'yes', why? C77

44. Do you think that children should be tested if they request it themselves without coercion? Yes^1 No^2 C79

45. If 'yes', why? C80

46. If 'no', why not? C81

47. Have you discussed your test result with your children? Yes^1 No^2 N/A^3 C82

49. If 'no', why not? C84

50. Has your result influenced your choice to have children/more children? C85

51. What do you feel about termination of pregnancy if the foetus were tested and found to have the HD gene? C86

52. Have you/your family member been for follow-up sessions at GSH since the predictive result? Yes^1 No^2 C87

54. If 'no', why not? C89

55. Have you/your family member ever been contacted again by any member of the professional team? Yes^1 No^2 C90

56. Describe the reasons for the interaction/s C91

SECTION D SATISFACTION WITH THE PREDICTIVE TESTING PROGRAMME

	Very unsatisfactory 1	Unsatisfactory 2	Satisfactory 3	Very Satisfactory 4	Highly satisfactory 5	
1. The manner in which HD was explained to you						D1
2. The doctor/counsellor/sister listened to what you had to say						D2
3. The doctor/counsellor/sister understood what was worrying you						D3
4. The doctor/counsellor/genetic sister answered all your questions						D4
5. The doctor/counsellor/genetic sister showed you a lot of caring						D5
6. The counselling helped you in making your decision about being tested						D6
7. The counselling helped you to cope better with your problems						D7
8. There was enough time given to you at each session						D8
9. The clinic environment						D9
10. The doctor/counsellor/genetic sister reassured you						D10
11. The manner in which the test results were presented to you						D11
12. There was time in the visits to have your questions answered						D12
13. The time you waited to get an appointment for your first visit						D13
14. The time you waited to see the counselling staff at each visit						D14
15. How satisfied were you with the information you received during the counselling sessions?						D15
16. The extent to which the team met your expectations of them						D16
17. Follow-up clinics/visits provided you with a lot of support						D17
18. General administration and concern shown by genetic sisters						D18
19. Overall, how do you rate the HD Predictive testing programme						D19

20. What could be done to improve the HD predictive testing programme? D20

21. Personal files indicate genetic sisters: Made follow-up call [1] Did not make follow-up call [2] Called for research consent only [3] D21

22. At the moment what is the biggest problem in your life (not necessarily related to HD)? D22

SECTION E HEALTH PROFILE OF PARTICIPANT

1. Since you last visited the clinic/hospital have you had to be hospitalised for some illness related to HD Yes [1] No [2] E1

If 'Yes' give reasons

2. Since you last visited the clinic/hospital have been put onto medication for some health problem related to HD? Yes [1] No [2] E2

3. Since you last visited the clinic/hospital have received any psychological support/counselling?
Yes ¹ No ² E3

If 'Yes' give reasons

4. Since you last visited the clinic/hospital have you noticed any changes in your personality or mood or intellectual ability or movement?
Yes ¹ No ² E4

5. How long after PT did they start? E5

SECTION F DISABILITY PROFILE OF PARTICIPANT/AFFECTED INDIVIDUAL

1. At what age did you first notice your symptoms of HD? F1

2. What were the first symptoms you noticed? F2

HEALTH ASSESSMENT QUESTIONNAIRE (HAQ)

	Without <u>any</u> difficulty	**With <u>some</u> difficulty**	**With <u>much</u> difficulty**	**<u>Unable</u> to do**	
DRESSING & GROOMING					
Are you able to:					
Dress yourself including tying your shoelaces and doing your buttons?	0	1	2	3	F3
Shampoo your hair?	0	1	2	3	F4
GETTING UP					
Are you able to:					
Stand up from an armless chair?	0	1	2	3	F5
Get in and out of bed?	0	1	2	3	F6
EATING					
Are you able to:					
Cut your own meat?	0	1	2	3	F7
Lift a full cup or glass to your mouth?	0	1	2		F8
Open a new milk carton?	0	1	2	3	F9
WALKING					
Are you able to:					
Walk outdoors on flat ground?	0	1	2	3	F10
Climb up and down five stairs?	0	1	2	3	F11

What <u>Aids or Devices</u> do you usually use for any of the above activities?

Specify ____

For what activities do you usually need help from another person?

_________1 Dressing & Grooming
_________2 Getting up
_________3 Eating
_________4 Walking

	Without any difficulty	With some difficulty	With much difficulty	Unable to do	
HYGIENE					
Are you able to:					
Wash and dry your entire body?	0	1	2	3	F12
Bath in a proper bath/shower?	0	1	2	3	F13
Get on and off the toilet?	0	1	2	3	F14
REACH					
Are you able to:					
Reach and get down a 2.5 kg object such as a bag of sugar from just above your head?	0	1	2	3	F15
Bend down to pick up clothing from the floor?	0	1	2	3	F16
ACTIVITIES					
Are you able to:					
Run errands and shop?	0	1	2	3	F17
Get in and out of a car?	0	1	2	3	F18
Do chores such as housework or gardening?	0	1	2	3	F19

What Aids or Devices do you usually use for any of the above activities?

Specify ____

What activities do you usually need help from another person?

_________1 Hygiene
_________2 Reach
_________3 Gripping and Opening Things
_________4 Errands and Chores

How much pain have you had because of your disability over the past week?

NO PAIN **VERY SEVERE PAIN**

Place a mark on the line to indicate the severity of the pain

0 1 2 3 4 5 6 7 8 9 10

F20

Comments of disabled person ……………………………………………………………………………

………

Physiotherapist's assessment of what assistive devices and/or mobility aids would improve the quality of life of the disabled person by making daily activities possible or easier

Wheelchair	Yes 1 No 2	F21
Bath board	Yes 1 No 2	F22
Grab bar	Yes 1 No 2	F23
Bed pan	Yes 1 No 2	F24
Male urinal	Yes 1 No 2	F25
Commode	Yes 1 No 2	F26
Chair for shower	Yes 1 No 2	F27
Bannister railing	Yes 1 No 2	F28
Walking frame	Yes 1 No 2	F29
Crutches	Yes 1 No 2	F30
Walking sticks	Yes 1 No 2	F31
Other		F32

LEVEL OF ABILITY TO FUNCTION IN SOCIETY

	No problem 1	**Mild problem 2**	**Moderate problem 3**	**Severe problem 4**	**Complete problem 5**	
Engagement in occupation						F33
Capacity to handle financial affairs						F34
Capacity to manage domestic responsibility						F35
Capacity to engage in social activities						F36

What do you find to be the biggest problem in your daily life? F37

What does the family find is the biggest problem in coping with the disabled person in their daily lives? F38

Have you heard about predictive testing? Yes 1 No 2 F39

Do your children know about the predictive testing programme? Yes 1 No 2 N/A 3 F40

Have any of them decided to be tested? Yes 1 No 2 Too young 3 N/A 4 F41

HOSPITAL ANXIETY AND DEPRESSION SCALE

I feel tense or 'wound up'	A
Most of the time	3
A lot of the time	2
From time to time occasionally	1
Not at all	0
I still enjoy the things I used to enjoy	D
Definitely as much	0
not quite as much	1
Only a little	2
Hardly at all	3
I get a sort of frightened feeling as if something awful is about to happen	A
Very definitely and quite badly	3
Yes, but not too badly	2
A little, but it doesn't worry me	1
Not at all	0
I can laugh and see the funny side of things	D
As much as I always could	0
Not quite as much now	1
Definitely not so much now	2
Not at all	3
Worrying thoughts go through my head	A
A great deal of the time	3
A lot of the time	2
From time to time, but not too often	1
Only occasionally	0
I feel cheerful	D
Not at all	3
Not often	2
Sometimes	1
Most of the time	0
I can sit at ease and I feel relaxed	A
Definitely	0
Usually	1
Not often	2
Not at all	3

I feel as if I am slowed down	D
Nearly all the time	3
Very often	2
Sometimes	1
Not at all	0
A get a sort of frightened feeling 'butterflies in the stomach'	A
Not at all	0
Occasionally	1
Quite often	2
Very often	3
I have lost interest in my appearance	D
Definitely	3
I don't take so much care as I should	2
I may not take quite as much care	1
I take just as much care as ever	0
I feel restless as if I have to be on the move	A
Very much indeed	3
Quite a lot	2
Not very much	1
Not at all	0
I look forward with enjoyment to things	D
As much as I ever did	0
Rather less than I used to	1
Definitely less than I used to	2
Hardly at all	3
I get sudden feelings of panic	A
Very often indeed	3
Quite often	2
Not very often	1
Not at all	0
I can enjoy a good book or radio or TV programme	D
Often	0
Sometimes	1
Not often	2
Very seldom	3

Total anxiety score ____________	F42
Total depression score	F43

SECTION G SOCIAL PROBLEMS EXPERIENCED

The following problems affect many families who have a member who suffers from a disability. How much does each of them affect you?

	No problem 1	Mild problem 2	Moderate problem 3	Severe problem 4	Complete problem 5	
1. Lack of money						G1
2. Transport						G2
3. Housing						G3
4. Safety						G4
5. Unemployment						G5
6. Isolation and loneliness						G6
7. Privacy						G7
8. Health Services						G8
9. Social Services						G9
10. Domestic assistance						G10
11. Attitude of the community						G11
13. Abuse						G13
14. Disability						G14
15. Health						G15
16. Other ?						G16

SECTION H SOCIAL GRANTS

1. Do you receive a disability grant or an Old Age pension? Yes ^{1} No ^{2}	H1
2. How do you supplement your disability grant or Old Age pension? Family ^{1} No other income ^{2} Other ^{3}: specify	H2
3. For how many years have your received a Disability Grant and/or Old Age pension?	H3
4. How many people does this grant support?	H4
5. How are they related to you? Spouse/partner ^{1} Children ^{2} Parents ^{3} Family ^{4} Other ^{5}	H5
7. Do you or you family receive all other social grants to which you are entitled? Yes ^{1} No ^{2}	H7
If "Yes" what grants do you receive? Disability Grant ^{1} School fees rebates ^{2} Grant-in-aid ^{3} Care dependency grants ^{4} Child support ^{5}	H8
If 'No' what grants are you entitled to? Disability Grant ^{1} School fees rebates ^{2} Grant-in-aid ^{3} Care dependency grants ^{4} Child support ^{5}	H9
8. Do you believe that your disability has been a financial burden on your family? Yes ^{1} No ^{2}	H10

SECTION J TRANSPORT

Which form of transport do you have access to?

	No access 1	For general use 2	For medical purposes 3	For disability pay-point 4	
Own vehicle					J3
Vehicle in the household					J4
Vehicle of family or friend					J5

Train					J6
Bus					J7
Taxi					J8
Dial-a-ride					J9
Other: specify					J10

What difficulties do you have with the public transport? J11

SECTION K HOUSING

1. Is the dwelling	self-constructed formal ^{1} built by the council ^{2} homeless ^{3} self-constructed informal ^{4}	K1
2. Type of dwelling:	house ^{1} flat ^{2} backyard shack ^{3}	K2
3. Do you or a family member:	own your home ^{1} rent the home ^{2}	K3
4. Do you live with:	as a boarder ^{1} legal or common law spouse ^{2} parents ^{3} siblings ^{4} children ^{5} relatives ^{6} friends ^{7} alone ^{8}	K4
5. How many rooms does the dwelling have?		K5
6. How many people permanently live in the household?:		K6
7. In total, how many people live in the:		
main house		K7
backyard shack		K8

What problems do you have with your housing? K9

SECTION M SOCIAL SUPPORT

THE MEDICAL OUTCOMES STUDY SOCIAL SUPPORT SURVEY (MOS)

People sometimes look to others for companionship, assistance, or other types of support. How often is each of the following kinds of support available to you if you need it?

(Circle one number on each line)

	None of the time	**A little of the time**	**Some of the time**	**Most of the time**	**All of the time**	
Someone to help you if you have to stay in bed	1	2	3	4	5	M1
Someone you can count on to listen to you when you need to talk	1	2	3	4	5	M2
Someone to give you good advice about a crisis	1	2	3	4	5	M3
Someone to take you to the doctor if you needed it	1	2	3	4	5	M4
Someone who shows you love and affection	1	2	3	4	5	M5
Someone to have a good time with	1	2	3	4	5	M6
Someone to give you information to help you understand a situation	1	2	3	4	5	M7
Someone to confide in or talk to about yourself or you personal problems	1	2	3	4	5	M8
Someone who hugs you	1	2	3	4	5	M9

Someone to get together with for relaxation	1	2	3	4	5	M10
Someone to prepare your meals if you were unable to do it yourself	1	2	3	4	5	M11
Someone whose advice you really want	1	2	3	4	5	M12
Someone to do things with to help you get you mind off things	1	2	3	4	5	M13
Someone to help with your daily chores if you were sick	1	2	3	4	5	M14
Someone to share your most private worries and fears	1	2	3	4	5	M15
Someone to turn to for suggestions about how to deal with a personal problem	1	2	3	4	5	M16
Someone to do something enjoyable with	1	2	3	4	5	M17
Someone who understands your problems	1	2	3	4	5	M18
Someone to love and make you feel wanted	1	2	3	4	5	M19

Additional comments ……………………………………………………………………………………………………

………

34. What has a negative impact on the amount of your socialisation? M20

SECTION N HEALTH SERVICES

PRESENT UTILISATION OF HEALTH SERVICES

Which health services do you use regularly?

3. Doctor at the clinic Yes ^{1} No ^{2} N5

4. Private doctor Yes ^{1} No ^{2} N7

6. Groote Schuur Hospital Yes ^{1} No ^{2} N13

7. Do you receive medication regularly? Yes ^{1} No ^{2} N17

8. From which institution(s)? Day hospitals ^{1} Privately ^{2} N18

SATISFACTION WITH GROOTE SCHUUR HOSPITAL NEUROGENETIC CLINIC

	Very unsatisfactory 1	Unsatisfactory 2	Satisfactory 3	Very Satisfactory 4	Highly satisfactory 5	
1. The manner in which your symptoms are explained to you						N19
2. The staff listen to what you have to say						N20

	Very unsatisfactory 1	Unsatisfactory 2	Satisfactory 3	Very Satisfactory 4	Highly satisfactory 5	
3. The staff understand what is worrying you						N21
4. The staff answer all your questions						N22
5. The staff show you a lot of caring						N23
6. Privacy when seeing the doctor						N24
7. The clinic helps you to cope better with your problems						N25
8. There is enough time given to you at each session						N26
9. The clinic environment						N27
10. The staff reassure you						N28
11. The manner in which the diagnosis was presented to you						N29
12. There is time to have your questions answered						N30
13. The time you wait to get an appointment if there is a need out of your regular clinic appointment						N31
14. The time you wait to see the doctor at each visit						N32
15. The staff are interested in you as a person						N33
16. Decisions are made for you with full explanations provided to you						N34
17. The clinics/visits provide you with a lot of support						N35
18. General administration and concern shown by genetic sisters						N36
19. Overall, how do you rate the Neurogenetic Clinic						N37

20. What difficulties do your experience with the Neurogenetic Clinic? N38

21. What recommendations can you make to improve the service? N39

SECTION R ABUSE

People associated with families with HD are often abused sometime during their lives. This abuse can take the form of physical, emotional or psychological abuse.

If you have been abused, describe your experiences of abuse as a child.

If you have been abused, describe your experiences of abuse as an adult.

SECTION U CARER

Study Code Number ……………………………………………………..

1. Gender:	Male ^{1} Female ^{2}	U1
2. Age:		U2
3. Are you the major carer?	Yes ^{1} No ^{2}	U3
4. What is your relationship to the person with the disability?		

Partner [1] Child/child-in-law [2] Sibling/sibling-in-law [3] Friend [5] Parent [6] Other [7] Relative [8]	U4
5. Do you stay in the same household as the disabled person? Yes [1] No [2]	U5
6. Are you employed? Yes [1] No [2]	U6
7. Do you receive a "grant-in-aid" from the Social Services Department? Yes [1] No [2]	U7
8. For how many years have you cared for the disabled person?	U8
9. Did you have to leave your job specially to stay at home and care for the	
disabled person? Yes [1] No [2]	U9

10. With which of the following tasks do you assist the disabled person?

	Not at all 1	**Supervise only 2**	**Seldom 3**	**Some-Times 4**	**Often 5**	**Always 6**	
Eating							U11
Bathing/showering							U12
Personal hygiene							U13
Getting on and off toilet							U14
Dressing							U15
Getting in and out of bed							U16
Giving medication							U18
Doing errands							U19
Walking							U20
Shopping							U21
Housework							U22
Preparing meals and cooking							U23
Laundry							U24
Listening and talking							U26

12. How many hours per day do you spend looking after the person with a disability? U29

CAREGIVER BURDEN SCALE (CBS)

- **Please check the one response which best describes your feelings over the past week:**

(Tick one block on each line)

	Not at all 1	Seldom 2	Some-Times 3	Often 4	
GENERAL STRAIN					
Do you find yourself facing practical problems in the care of your relative that you think are difficult to solve?	1	2	3	4	U31
Do you think you have to take too much responsibility for your relative's welfare?	1	2	3	4	U32
Do you sometimes feel as if you would like to run away from the entire situation you find yourself in?	1	2	3	4	U33
Do you feel tired and worn out?	1	2	3	4	U34
Do you feel tied down by your relative's problems?	1	2	3	4	U35
Do you find it mentally tiring to take care of your relative?	1	2	3	4	U36
Do you think your own health has suffered because you have been taking care of your relative?	1	2	3	4	U37
Do you think you spend so much time with your relative that you do not have enough time for yourself?	1	2	3	4	U38
ISOLATION					
Do you avoid inviting friends and acquaintances home because of your relative's problem?	1	2	3	4	U39
Has your social life, e.g. with family and friends, been reduced?	1	2	3	4	U40
Has your relative's problem prevented you from doing what you had planned to do in this phase of your life?	1	2	3	4	U41
DISAPPOINTMENT					
Do you feel that life has treated you unfairly?	1	2	3	4	U42
Did you expect that your life would be different from what it is at your age?	1	2	3	4	U43
Do you feel lonely and isolated because of your relative's problem?	1	2	3	4	U44
Do you find it physically tiring to take care of your relative?	1	2	3	4	U45
Have you had to make financial sacrifices because you have been taking care of your relative?	1	2	3	4	U46
EMOTIONAL INVOLVEMENT					
Are you sometimes ashamed or embarrassed by your relative's behaviour?	1	2	3	4	U47
Do you ever feel hurt and angry with your relative?	1	2	3	4	U48
Do you feel embarrassed by your relative's behaviour?	1	2	3	4	U49

	Not at all 1	Seldom 2	Some-Times 3	Often 4	
ENVIRONMENT					
Does the physical environment make it difficult for you to take care of your relative?	1	2	3	4	U50
Do you worry about not taking care of your relative in the proper way?	1	2	3	4	U51
Is there anything in the neighbourhood which makes it difficult for you to take care of your relative?	1	2	3	4	U52

NEEDS OF CARER

What are your biggest problems with being a caregiver? U53

SECTION W PARTICIPANTS IN GENERAL

Do you agree that families with HD have the following needs?:

Home visits by social worker	Yes ^{1}	No ^{2}	W4
Support group for affected individual	Yes ^{1}	No ^{2}	W6
Support group for main carer	Yes ^{1}	No ^{2}	W7
Children's support group	Yes ^{1}	No ^{2}	W8
Interest group for information	Yes ^{1}	No ^{2}	W9
HD Care Facility for nursing care	Yes ^{1}	No ^{2}	W10
HD Care Facility for respite care	Yes ^{1}	No ^{2}	W11

LIFESTYLE CHANGES WITH REGARD TO PREDICTIVE TESTING AND DIAGNOSIS OF HD

With regards to your smoking habits

	N/A 1	Not at all 2	A few 3	Moderately 4	Heavily 5	
How much did you smoke before your PT?						W12
How much did you smoke after your PT?						W13
How much did you smoke before you developed symptoms of HD?						W14
How much did you smoke after you developed symptoms of HD?						W15

UNIVERSITY OF CAPE TOWN

DIVISION OF HUMAN GENETICS

HD PREDICTIVE TESTING FOLLOW-UP STUDY

PROTOCOL FOR INITIAL CONTACT WITH INDIVIDUALS

1. Telephonic contact will be made by a genetic sister/counsellor to all individuals who have undergone predictive testing to explain the purpose of the research to be conducted, namely:

- To investigate how effective the participants found the counselling and support processes of the predictive testing programme offered by the Division of Human Genetics, UCT;
- To investigate the impact that predictive testing has had on the individual and family members; and
- To determine the nature and extent of the social, physical, attitudinal and environmental barriers confronting the participants.

2. The participant will be informed that a home visit will be undertaken by a researcher who has not previously been part of the HD programme or genetic counselling team in order to interview the individual. The participants will be reassured that:

- the researcher does not know the test result
- all information provided to the researcher during the interview will be kept confidential apart from publication in a scientific journal where names will not be used;
- the information will not be discussed with extended family members;
- the information will not be discussed with the predictive testing genetic counseling team;
- enrolment is completely voluntary and that they may choose not to participate or can withdraw from the study at any time without jeopardising their or their families' access to the medical and genetic services to which they are entitled; and
- the researcher is a physiotherapist who has had many years of experience in dealing with people with physical disabilities and that, if any of their family members are affected, she may be able to offer them advice and assistance in their management to enable them to live as independently as possible and to reduce the burden on the caregiver.

3. The individuals will be invited to participate in the study. If they agree, they will be informed that Dr Merle Futter will be given their contact details and will contact them personally to arrange a convenient time and place for the interview.

4. If the individual does not wish to participate in the study the genetic sister/counsellor is requested to ask the participant for the reason for the study purpose. This will be conveyed to the researcher verbally to maintain confidentiality.

M J Futter
June 2005

If you are HIV positive it is very important to prevent passing the infection on to others. Find out more about safer sex, and use a condom every time you have sex.

As jy HIV-positief is, is dit bale belangrik om te voorkom dat jy hierdie infeksie aan ander mense oordra. Vind meer uit oor veiliger seks, en gebruik 'n nuwe kondoom elke keer as jy seks het.

Ukuba une-HIV kubaluleke kakhulu ukuba uthintele ukugqithisela olu losulelo kwabanye abantu. Fumana inkcazelo ebanzi malunga nokulalana okukhuselekileyo, kwaye sebenzisa ikhondom qho xa ulalana (usabelana ngesondo).

Fa o nale HIV go botlhokwa gore o seke wa fetisa tshwaetso e, go bangwe. Batlisisa ka mekgwa ya go dira thobalano e e sireletsegileng mme o dirise khondomo nako nngwe le nngwe fa o dira thobalano.

ign 1907/98 © 1998. Tel (012) 312-0121 Fax: (012) 326-2891 Private Bag X828, Pretoria, 0001

If you have any questions about HIV/AIDS you can phone the free 24-hour AIDS Helpline at 0800-012-322.

As u enige vrae oor HIV/VIGS het, kan u die gratis 24-uur VIGS hulplyn skakel op **0800-012-322.**

Ukuba unayo nayiphina imibuzo nge HIV/AIDS, ungafowunela inombolo yasimahla yoNcedo ye-AIDS efumaneka iiyure ezingama 24 ethi **0800-012-322.**

Fa o nale dipotso ka HIV/AIDS o ka leletsa mogala mo go *Helpline* ya AIDS ya diura di le 24 e e sa duelelweng kwa go **0800-012-322.**

There are a number of other leaflets in this series that give more information about AIDS.

Daar is 'n aantal ander pamflette in hierdie reeks wat meer inligting verskaf oor VIGS verwante kwessies.

Kukho nezinye iincwadana ezininzi kolu luhlu ezinika inkcazelo ebanzi ngemibandela ephathelelene ne-AIDS.

Go nale dipapetsana tse di mmalwa mo serising eno tse di nang le tshedimosetso ka ga AIDS.

G.P.-S.

FORMESET PRINTERS

Living with HIV/AIDS

AFRIKAANS	Hoe om met HIV/VIGS saam te leef	A
ISIXHOSA	Ukuphila une-HIV/AIDS	X
SETSWANA	Go tshela ka HIV/AIDS	T

AIDS HELPLINE
☎ **0800-012-322**

AIDS is a disease that affects millions of South Africans. It is caused by a virus called HIV that slowly weakens a person's ability to fight off other diseases.

VIGS is 'n siekte wat miljoene Suid-Afrikaners raak. Dit word veroorsaak deur 'n virus genaamd HIV, wat geleidelik 'n persoon se weerstand teen ander siektes aftakel.

I-AIDS sisifo esichaphazela iimiliyoni zabantu baseMzantsi Afrika. Ibangelwa yintsholongwane (vayirasi) ebizwa ngokuba yi HIV, yona eqhubeka kancinane (ethi chu) ukuqoba amandla omntu okulwa nezinye izifo.

AIDS ke bolwetse jo bo amang dimilione tsa batho fa Aferika Borwa. E tlhodiwa ke virase e e bidiwang HIV e e dirang ka go koafatsa mmele wa motho go o palledisa go lwantsha malwetse a mangwe.

If you are infected with HIV you can still lead a normal healthy life for many years by:

☐ taking care of your health
☐ being positive about life
☐ getting support from those around you
☐ practicing safer sex.

As jy met HIV besmet is, kan jy steeds baie jare lank 'n normale, gesonde lewe lei deur:

☐ om te sien na jou gesondheid
☐ positief te wees oor jou lewe
☐ ondersteuning te kry van mense na aan jou
☐ veiliger seks te beoefen.

Ukuba wosuleleke yi-HIV usengahlala ubomi obunempilo iminyaka emininzi ngokwenza ezi zinto zilandelayo:

☐ ngokukhathalela impilo yakho
☐ ngokuba nethemba nokuqiniseka ngobomi
☐ ngokufumana inkxaso kwabo bakungqongileyo
☐ ngokulalana (ukwabelana ngesondo) okukhuselekileyo.

Fa o tshwaeditswe ke HIV o ka tshela botshelo jo bo tletseng jo bo nonofileng mengwaga e le mentsi ka:

☐ go tlhokomela boitekanelo jwa gago
☐ go nna o ne le dikakanyo tse di ntle ka botshelo
☐ go bona tshegetso go tswa go batho ba o tshelang le bona
☐ go dira thobalano e e sireletsegileng.

Taking care of your health includes:

☐ eating properly
☐ getting exercise
☐ getting enough rest.

Jy kan na jou gesondheid omsien deur onder andere:

☐ reg te eet
☐ liggaamlike oefening te kry
☐ genoeg te rus.

Ukukhathalela impilo yakho kubandakanya:

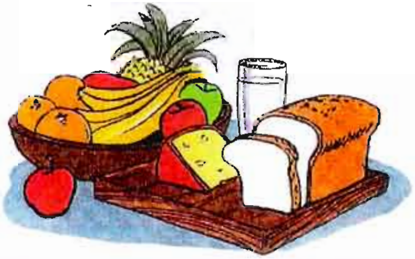
☐ ukutya ngokufanelekileyo
☐ ukuqhelisa umzimba (uku-eksesayiza)
☐ ukuphumla ngokwaneleyo.

Go tlhokomela boitekanelo go akaretsa:

☐ go ja sentle
☐ go ikatisa
☐ go netefatsa gore o bona khutso e o e tlhokang.

Eating healthily can help your body fight diseases. This includes:

☐ eating a lot of fresh fruits and vegetables
☐ eating whole grains such as samp and brown bread
☐ avoiding junk foods
☐ drinking lots of water
☐ avoiding alcohol, smoking and drugs
☐ keeping your cooking and eating area clean



As jy reg eet, help jy jou liggaam om siektes te bestry. Doen die volgende:

☐ eet baie vars groete en vrugte
☐ eet volgraanvoedsel soos samp en bruinbrood
☐ vermy kitskos
☐ drink baie water
☐ vermy alkohol, sigarette en dwelms
☐ hou jou kook- en eetgebied skoon.

Ukutya ngokunempilo kunganceda umzimba wakho ulwe izifo. Oku kubandakanya:

☐ ukutya kakhulu iziqhamo ezifreshi kunye nemifuno
☐ ukutya iinkozo ezipheleleyo ezifana nomngqusho kunye nesonka esibhrawuni
☐ uzame ungatyi ukutya okuphekwe okanye okufrayishwe ngokukhawuleza (fast foods)
☐ usele amanzi amaninzi
☐ uzame ungaseli utywala, ungatshayi kwaye ungasebenzisi iziyobisi
☐ ugcine indawo yakho yokuphekela nokutyela icocekile.

Appendix vi

UNIVERSITY OF CAPE TOWN

Research Ethics Committee
E52 Room 24 Old Main Building Groote
Schuur Hospital, Observatory, 7925
Queries : Lamees Emjedi
Tel : (021) 406-6338 Fax: 406-6411
E-mail : lemjedi@curie.uct.ac.za

17 June 2005

REC REF: 362/2003

Dr. MJ Futter
Human Genetics

Dear Dr. Futter

PREDICTIVE TESTING FOR HUNTINGTON DISEASE IN THE WESTERN CAPE: THE IMPACT OF THE PROTOCOL, THE PROGRAMME AND THE PROFESSIONALS ON PARTICIPATING INDIVIDUALS AND THEIR FAMILIES

Thank you for your letter to the Research Ethics Committee dated 01/06/2005.

It is a pleasure to inform you that the Ethics Committee has formally approved the above-mentioned study including the amendments referred to in the above-mentioned letter.

Please quote the REC. REF in all your correspondence.

Yours sincerely

PROF T ZABOW
CHAIRPERSON

WESTERN CAPE HUNTINGTON DISEASE RESEARCH PROJECT

INFORMATION AND CONSENT FORM

STATEMENT BY PARTICIPANT

I, ………………………………………….., living at (address) …………………..……………

confirm that:

1. I have been invited to participate in the above research project which has been initiated through the Division of Human Genetics, University of Cape Town/Neurogenetic Clinic, Groote Schuur Hospital because I have been through the programme predictive testing programme are presently attending the Neurogenetic Clinic.

2.1 I understand that the objective of this study is to investigate:

- whether the predictive testing and support programme meets my and my family's needs;
- how I and my family are coping with the test result;
- what I and my family feel about the way the protocol that had to be followed before my blood was taken for testing;
- how I feel about the different staff involved and how helpful they were to me during the testing process;
- what effects the test result had on me and my family;
- what effects the test result had on me and my family's future plans;
- what changes are necessary to make the testing programme more helpful for me and my family;
- the problems I experience in my daily life in my home and community;
- the different aspects that contribute to these problems;
- whether my or my family's quality of life can be improved by learning basic management skills to assist with my affected family member/s so that they can remain as independent as possible.

2.2 I understand that the interview will take part in my home or another venue of my choice and that it may take one or two visits of two hours each.

2.3 I am aware that this is a once-off procedure that will be implemented in 2005/6 at a time convenient to me and my family.

2.4 I understand that some of the questions may make me angry or sad, but the risks to me from the study are minimal. The researcher is an experienced physiotherapist and she will refer me to my HD genetic counsellor, if necessary. She will show me respect, acceptance and empathy during the interview.

3.1 I have been assured that all information will be handled confidentially. Information may be used for a thesis, publications in scientific journals and presentations at professional congresses, but names and addresses will not be included and will not be made available to anybody else other than the researcher.

3.2 I understand that the interview with be tape recorded so that the researcher does not have to write too much during the interview. The tape will be stored in a safe until the research has been written up and will then be destroyed immediately. The data stored on the computer will have a numerical code only and my name will not appear anywhere.

4. I have been assured that the recorded and transcribed information discussed at the meeting will only be made available to members of the HD programme with my study code

number and that they do not know that it refers to my name or address. The photographs may be used for presentation or teaching purposes locally or overseas.

5. I have not been persuaded to consent to taking part in the study and I have been informed that I may refuse to participate in this project and that I may stop participating at any stage, and that such refusal or stoppage will not in any way negatively affect my future access to medical and genetic services to which I am entitled.

6. Dr Merle Futter has explained the information of the study to me in English/Afrikaans. I am proficient in that language and my questions have been answered satisfactorily.

7. I understand that there are no medical benefits to me from this study, but there will be a chance for me to talk to a professional about how I am coping with my/relative's HD predictive test result and how it has affected my/our lives and that the results of this study may benefit other individuals who need the test in the future.

8. I have been assured that participation in this project will not lead to additional costs for me or my family and I will not benefit from it financially.

I HEREBY DECLARE THAT I WILL VOLUNTARILY PARTICIPATE IN THE ABOVE RESEARCH STUDY

Signed at (address) .... on 2005/6

....
Participant's/proxy signature

I HEREBY DECLARE THAT I AGREE TO HAVE MY INTERVIEW AUDIOTAPE RECORDED

Signed at (address) .... on 2005/6

....
Participant's/proxy signature

I HEREBY DECLARE THAT I AGREE TO HAVE MY PHOTOGRAPH TAKEN, TO BE SHOWN FOR TEACHING PURPOSES LOCALLY OR OVERSEAS OR PUBLISHED IN A THESIS OR SCIENTIFIC JOURNAL WITHOUT MY FACE BEING OBSCURED

Signed at (address) .... on 2005/6

....
Participant's/proxy signature

IMPORTANT INFORMATION

Dear Participant

Thank you for participating in this study. Should you have any questions during the duration of this study regarding:

1. problems as a result of the research, or
2. questions regarding information about the project

please contact me at the following telephone number: Dr Merle Futter: (021) 406-6425
Cell 083-697-9118
Prof Jacquie Greenberg: (021) 406-6299

If you have any questions about your rights as a research participant please contact Prof T Zabow, Chair of the Research Ethics Committee, Faculty of Health Sciences, University of Cape Town Ethics Review Committee on (021) 406-6492.

AFRIKAANS BOOKLETS

The author wrote the booklets in English and they were then translated into Afrikaans. As the average number of years of schooling, the participants requesting information in Afrikaans, was seven years a school teacher who taught Grade 7 learners at school translated the text to ensure that it would easily be understood by people with that level of education.

For the benefit of the examiners the booklets included are the English version.

UNIVERSITY OF CAPE TOWN
DIVISION OF HUMAN GENETICS

Level 3, Wernher & Beit North Building, Faculty of Health Sciences, Observatory 7925, Cape Town, South Africa
Fax: 021-406-6826, Telephone: 021-406-6297

UNIVERSITY OF CAPE TOWN PREDICTIVE TESTING PROTOCOL FOR HUNTINGTON DISEASE

The predictive test for Huntington disease is a process which allows an individual at risk for having inherited the gene for Huntington disease a chance of knowing whether he or she has the affected gene, before showing signs of the disease.

To take the test is a very serious decision as the knowledge of your test result will remain with you for life. Therefore, it is important that you are well informed and understand the programme and procedures you will have to go through before being given your result. A programme or protocol has been recommended by international genetic counselling teams based on their experiences in dealing with individuals and families at risk for the disease.

Predictive testing is not generally available to

- individuals under 18 years of age;
- pregnant couples; or
- individuals who have only recently become aware of Huntington disease being in their family as it is advisable to wait for a year before undergoing the predictive testing programme.

If, after careful consideration, you decide to undergo the test, you will be requested to come to the Groote Schuur Hospital Neurogenetic Clinic or the Division of Human Genetics on at least five occasions to see the different members of the genetic counselling team.

FIRST MEETING (GENETIC COUNSELLOR AND GENETIC NURSE IN ATTENDANCE)

The genetic counsellor will ask you for your family history and explain the protocol. He/she will ask you for your reason for requesting a predictive test and will explain the inheritance pattern of the disease and the methods and limitations of the predictive test.

1. As you know, the affected gene for Huntington disease has been found and we are now able to offer a blood test that will predict whether or not you will get the disease.

***IMPORTANT:** It is very important at the beginning that you understand that there is approximately 1% possibility that the test may not show a clear result with absolute certainty. If this occurs in your case, then unfortunately your risk of carrying the gene remains exactly the same as before you entered the programme (i.e. 50%).* When the final result is given, it could be one of three:

* *Positive* - you have the affected gene and will develop the symptoms at some stage in the future. We have no way of knowing when and how it will present.

* *Negative* - based on our current knowledge of this gene, you will not develop Huntington disease.

* *Uninterpretable* - no result. You are still at risk and have a 50% chance of carrying the affected gene.

2. We strongly recommend that you inform your family doctor of your decision to undertake the test. If you do not have a family doctor we recommend you find one. As part of the policy of this programme, we believe that the on-going medical care and support your doctor is able to give you is very important to you and your family. A letter will be sent to your doctor to inform him/her about the programme after the third visit.

3. Support is essential and we therefore strongly advise that you choose a family member or a trusted friend to accompany you to all the meetings.

4. The final result will be given to you approximately six weeks after your blood samples have been taken and will be strictly confidential. No result will be given to you by telephone. Written confirmation of your result will be sent to the genetic centre that referred you. With your permission, your family doctor will be contacted and given the result.

5. If at any stage in the programme you decide you do not wish to continue, the decision is entirely yours. Your decision will in no way prejudice our relationship with you or your family. We will be happy to continue to offer you all the support and help you need.

6. In order to ensure confidentiality, family members who request testing together will attend individual counselling sessions with the genetic team members.

SECOND MEETING (NEUROLOGIST AND ANOTHER MEMBER OF THE GENETIC TEAM) (APPROXIMATELY FOUR WEEKS AFTER FIRST MEETING)

The neurologist will perform a neurological examination to exclude early symptoms.

THIRD MEETING (PSYCHOLOGIST IN ATTENDANCE) (APPROXIMATELY FOUR WEEKS AFTER SECOND MEETING)

1. You must be accompanied by your support partner to this meeting.

2. A psychologist will make an assessment to exclude any serious psychological problems.

3. During this meeting the psychologist will discuss your reasons for requesting predictive testing as well as the possible effects that a positive or negative result may have on you, your partner and your family. Counselling may vary widely. For example, people who have the gene may suffer a sense of shock and grief however well they may have been prepared beforehand. People whose risk is negative may feel relief but at the same time suffer guilt and anxiety. For those with no result their feelings could be anger and frustration.
4. You will be asked to make an appointment to see the genetic counsellor approximately one month after this meeting.

5. It may be necessary to include another interview in the pre-test period if either you or we may need further information or discussion.

FOURTH MEETING (GENETIC COUNSELLOR AND GENETIC NURSE IN ATTENDANCE) (APPROXIMATELY FOUR WEEKS AFTER THIRD MEETING)

1. You must be accompanied by your support partner.

2. At this meeting the genetic team will need your written consent to take blood samples from you. Again there will be careful explanation of the test and the manner in which the final result will be given: about six weeks after the blood sample has been taken you will be contacted to make an appointment to see the genetic counsellor and psychologist when you will be given the final result.

3. Your GP will be contacted once your blood has been taken and a letter will be sent to him/her about the test. No result will be given to you over the telephone, whatever the result may be.

4. Due to the complexity of the test it may take longer than six weeks.

FIFTH MEETING (PSYCHOLOGIST, GENETIC COUNSELLOR AND/OR GENETIC NURSE IN ATTENDANCE) (APPROXIMATELY FOUR WEEKS AFTER FOURTH MEETING)

1. You will be contacted and an appointment made to finally receive your result. No result will be given over the telephone, whatever it might be. ***Please note that the genetic nurse does not know the result when she calls you.***

2. The genetic team in attendance, and again only with your consent, will give you the final result. You must be accompanied by your support partner at this meeting. You will be given a short recess alone with your partner after the test result has been given and this will be followed by a discussion to answer any questions you may have.

3. Once the result has been discussed with you and again, with your consent, your GP will be informed telephonically.

4. The genetic nurse will make an appointment for one week later and then for one month, three months, six months and one year later for follow-up visits.

Contact persons: **Sister Diane Sklar** **Tel: (021) 404 6235 or 406 6304**
or
Sister Trish Legg **E-mail: Diane.Sklar@uct.ac.za**
Patricia.Legg@uct.ac.za

03 July 2007

PROVINCIAL ADMINISTRATION : WESTERN CAPE
Department of Health
PROVINSIALE ADMINISTRASIE : WES-KAAP
Departement van Gesondheid
ULAWULO LWEPHONDO : INTSHONA KOLONI
Isebe Lezempilo

TO WHOM IT MAY CONCERN

.... suffers from Huntington Disease which is a medical condition that affects the brain. This causes him to have involuntary movements of his arms, legs and face and to talk and walk as though he is drunk. He has a neurological disease and regularly attends the Groote Schuur Hospital Neurogenetic Clinic.

WIE DIT MAG AANGAAN

.... lei aan 'n mediese toestand wat Huntington siekte genoem word. Hierdie toestand affekteer die brein. Dit veroorsaak dat hy nie beheer het oor die bewegings van sy arms, bene en gesig nie. Dit veroorsaak ook dat hy/sy loop en praat asof hy/sy dronk is. Hy het 'n neurologiese siekte en besoek gereeld Groote Schuur Hospitaal Neuro-genetiese Kliniek in die verband.

Signature .... Date ....

Name ....

Contact persons: Sr Diane Sklar or Sr Trish Legg
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