

**An investigation into the level of genetic knowledge of parents of sons
with Duchenne Muscular Dystrophy and their satisfaction with the
genetic counselling service at Red Cross War Memorial Children's
Hospital.**

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ABSTRACT

Duchenne muscular dystrophy (DMD) is the most severe and common form of muscular dystrophy with a prevalence in the region of 1 in 3500 live male births. Red Cross War Memorial Children's Hospital (RCWMCH) has a weekly neuromuscular clinic where, amongst others, boys with DMD and their parents are seen for clinical management by a paediatric neurologist. South African parents who have a son with DMD are confronted with unique circumstances and barriers. The aims of this study were to determine the level of understanding of genetics of parents who have sons with DMD; to determine their level of satisfaction with the genetic counselling service at RCWMCH; and to investigate the impact that DMD has had on the family.

A qualitative approach was selected as it aims to understand and provide descriptions that portray the richness and complexity of real-life events from the perspective of the participants. Ten semi-structured qualitative interviews were conducted with parents who had sons with DMD between the age of 8 years and 15 years of age. Interviews were conducted in the language of the participants' choice and signed informed consent was obtained prior to the interview.

The majority of participants demonstrated a high level of satisfaction with the service received at RCWMCH neuromuscular clinic; although there were some areas of the service which they indicated could be enhanced. The accessibility of the clinic varied among the higher and lower socioeconomic groups. A common complaint from the participants was that they were inadequately educated about DMD. There was generally an inadequate level of understanding of the genetics of DMD. The level of understanding was identified as being related to socioeconomic status as well as level of education. Most of the problems experienced by the participants were of a practical nature. Financial problems, difficulties with public transport and lifting the affected boy were among the most frequently mentioned by the participants.

This was the first research of its kind in South Africa. The findings of this study will help healthcare professionals involved in the RCWMCH neuromuscular clinic to better understand the barriers that South African parents of sons with DMD face. The service that is delivered at the RCWMCH may be improved by having a genetic counsellor as a member of the interdisciplinary team involved with all families in which a diagnosis of DMD is made, to facilitate information giving and assist in providing the necessary psychosocial support.

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LIST OF TERMS AND ABBREVIATIONS

CF	Cystic Fibrosis
Dial - a - Ride	Transport service for people with special needs which is funded by the City of Cape Town, the Provincial Government of the Western Cape and the National Department of Transport.
DMD	Duchenne Muscular Dystrophy
DNA	Deoxyribonucleic acid
MDF	Muscular Dystrophy Foundation
RCWMCH	Red Cross War Memorial Children's Hospital
SA	South Africa
TB	Tuberculosis
Temp	Temporary Worker
TOP	Termination of Pregnancy
UK	United Kingdom
USA	United States of America

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Chapter 1

INTRODUCTION

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1.1 INTRODUCTION

Duchenne muscular dystrophy (DMD) is the most severe and most common form of muscular dystrophy with a prevalence in the region of 1 in 3500 live male births (Mueller and Young 2001; Emery 1991). The estimated world-wide prevalence of DMD is 63 people per million. Approximately 8000 males in the United States of America (USA) are affected with DMD (Emery 1991). To date there is no incidence or prevalence statistics known for South Africa (SA).

The Red Cross War Memorial Children's Hospital (RCWMCH) has a weekly Neuromuscular Clinic where, amongst others, boys with DMD and their parents are seen for clinical management by a paediatric neurologist. The clinic has a multidisciplinary team approach involving neurologists and physiotherapists, and a counsellor from the Cape Branch of the Muscular Dystrophy Foundation of SA. This is the only clinic of its kind in SA and most affected boys from the Western Cape area are seen at this clinic. Once the diagnosis of DMD has been confirmed clinically and if possible molecularly, the family of the affected boy is given information about the genetics of DMD. This includes the implications for other family members and the prognosis of the disease. This information is then repeated at following visits especially if the parents are considering having more children. When a diagnosis of DMD is made it has a profound impact on the family both emotionally and practically. It was at this clinic that the researcher became aware of the unique circumstances and barriers with which South African parents that have a son with DMD are confronted. This includes poverty and minimal access to resources. Many families do not have running water or electricity in their homes and some have to share outside toilet facilities with other families. As there is a paucity of research into the psychosocial impact of DMD on the parents and the family there was a great need to determine what the impact of DMD has been on these families and to improve the support the families are receiving at the RCWMCH by improving the genetic counselling service.

1.2 AIMS

The aims of this study were to:

- Investigate the genetic knowledge of parents with a son with DMD;
- Identify the problems experienced by parents with a son with DMD; and
- Investigate the level of satisfaction the parents experience with the genetic counselling service at RCWMCH.

1.3 OBJECTIVES

The objectives of the research were to:

- Investigate the level of knowledge of the inheritance of DMD and knowledge of the carrier status of female family members;
- Investigate the sociodemographics of the group;
- Investigate the impact that DMD has had on the family; and

1.4 ORGANIZATION OF STUDY

In Chapter Two a literature review will be presented on various aspects of DMD. The methodology is described in Chapter Three. The process of identifying and contacting the participants is explained and the measurement instruments are described in terms of appropriateness and validity/trustworthiness. A brief description of the data gathering and analysis is provided. The procedure of the entire research study is also described. The findings of the study are presented and discussed in Chapter Four. The conclusion in Chapter Five summarizes the main findings of the research.

Chapter 2

LITERATURE REVIEW

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2.1 INTRODUCTION

This chapter includes literature reviews of the clinical aspects of DMD, including diagnosis and management thereof, the level of understanding of genetics following genetic counselling; satisfaction with genetic counselling service, problems experienced by parents of sons with DMD; and the impact DMD has on a family. Literature searches were performed using PubMed, Ebscohost, ScienceDirect and Google Scholar research databases. There was no literature available on the South African experience and all literature refers to experiences of participants in developed countries, such as the UK and USA.

2.2 CLINICAL ASPECTS OF DMD

DMD is a genetic condition which is caused by a mutation in the DMD gene which codes for the protein dystrophin. The DMD gene is one of the largest human genes to be identified to date and was isolated in 1987 (Mueller and Young 2001). Since DMD is inherited in an X-linked recessive manner, it predominantly affects boys (Emery 1998).

Clinical signs of the disease can be noted as early as 2 to 3 years of age. Males with DMD usually present with delayed motor development, including delays in sitting and standing independently, with approximately half of these boys failing to walk until the age of 18 months (Bridgeman and Prior 2005). Muscle weakness and wasting is usually progressive and symmetrical, affecting the lower limbs first, followed by the upper limbs. The proximal muscles are affected before the distal muscles. Proximal weakness results in other early characteristics which include an unusual waddling gait, inability to run quickly, difficulty in climbing stairs and lumbar lordosis (Bridgeman and Prior 2005; Mueller and Young 2001). Most boys with DMD have enlarged calves which are firm to palpitation. Usually by age five, rising from the floor can only be achieved by the child "climbing up" his legs and pushing down on his thighs with his hands in order to extend his hips and trunk (Gowers' sign) (Mueller and Young 2001). Another important clinical manifestation of DMD is joint contractures. As the disease progresses

the contractures increasingly develop and may lead to asymmetrical spinal deformities (scoliosis). DMD progresses rapidly and affected males typically require a wheelchair by age 12 (Bridgeman and Prior 2005). Cardiomyopathy is a consistent finding in DMD, and increases steadily in the teenage years. More or less one third of boys with DMD have some form of cardiac involvement by age 14 years, one half by age 18 years and all individuals with DMD have cardiac involvement after age 18 years (Nigro et al 1990). Some degree of nonprogressive cognitive impairment is present in approximately 20% of boys with DMD and affects verbal ability more than non verbal performance. The cognitive impairment cannot be attributed only to the physical limitations of the child and although the neuropathological link for mental retardation in DMD has not been established, a specific isoform of the DMD protein, dystrophin, has been shown to be expressed in the brain (Bridgeman and Prior 2005) and this may be part of the explanation.

2.2.1 Diagnosis of DMD.

A positive family history which is compatible with X-linked recessive inheritance and the clinical features mentioned above can be used to make a clinical diagnosis of DMD. There are various tests that can be done to assist in making the diagnosis. These include serum creatine phosphokinase concentration testing; muscle biopsy with dystrophin studies; and identifying the DMD gene mutation to confirm the diagnosis. A brief description of the studies follows:

a) *Serum Creatine Phosphokinase testing:* In patients with DMD there seems to be an increased permeability of the muscle membrane as a result of the dystrophic process and this leads to an escape of muscle enzymes into the blood. Serum creatine kinase levels are dramatically elevated in patients with DMD. The levels can be increased more than tenfold normal levels and this can be used to confirm the diagnosis (Mueller and Young 2001).

b) *Skeletal muscle biopsy:* Early in DMD non-specific dystrophic changes in the muscle histology include variation in fibre size, foci of necrosis and regeneration, and hyalinization. Later in the disease the histology shows deposition of fat and connective tissue. The dystrophin levels in muscle biopsies of patients with DMD are very low or absent (<3% of normal levels) (Hoffman et al 1988).

c) *DMD gene mutation identification:* The DMD gene encodes the protein dystrophin which is expressed predominantly in smooth, skeletal and cardiac muscle (Mueller and Young 2001). Approximately 60 - 65% of the mutations that cause DMD are large deletions which are generally clustered in specific regions of the gene. Therefore, when diagnostic testing is done for DMD these areas are generally tested first. There seems to be no relationship between the size of the deletion or its location and the severity and progression of the disorder. In the remainder of the patients with DMD who do not have large deletions there may be point mutations and small deletions and duplications that cause the disease (Bridgeman and Prior 2005). Molecular analysis of DMD gene mutations has greatly improved diagnosis, carrier detection and genetic counselling for DMD. In SA molecular testing is done across the known 'hotspot' regions of the DMD gene and can provide a molecular diagnosis for approximately 55% of patients (Goliath 2006).

d) *Carrier testing:* When a molecular diagnosis is confirmed in a patient with DMD accurate carrier detection can be achieved for most female relatives of the affected male (Mueller and Young 2001). Carrier status is determined by gene dosage and these determinations can be made from Southern blot or quantitative polymerase chain reaction (Bridgeman and Prior 2005). In SA these services are not currently available and carrier detection is based on linkage analysis. Linkage analysis is performed with markers that are highly polymorphic and informative, but due to the large size of the DMD gene there is a 12% risk of recombination across the gene (Mueller and Young 2001).

2.2.2 Management of DMD.

Currently there is no cure for DMD, although there are hopes of gene therapy being developed (Mueller and Young 2001). Appropriate management of the symptoms of DMD can prolong survival and improve the quality of life of patients with DMD. Issues that need special attention are weight control to prevent obesity, physiotherapy to prolong ambulation and prevent contractures, routine screening by a cardiologist to monitor evidence of cardiac involvement and monitoring for orthopaedic complications like scoliosis (Sussman 2002). Corticosteroids, including prednisone and a related compound, deflazacort, have recently been shown to delay the loss of muscle strength and prolong ambulation in boys with DMD (Sussman 2002), though in many boys these medications may have undesired effects such as weight gain, immunosuppression and glucose intolerance (Nereo et al 2003).

2.3 GENERAL PSYCHOSOCIAL ASPECTS OF CHRONIC ILLNESS AND DISABILITY.

Chronic illnesses and disability affect the physical, psychological, social, vocational and economic functioning of the affected individual and that of their families (Livneh and Antonak 1997). There is a profound effect on the lives of these persons due to the combination of the prolonged course of management, the often uncertain prognosis, the constant and intense psychosocial stress and the gradually decreasing level of independence with the performance of daily activities and life roles (Livneh and Antonak 1997). The diagnosis of a disabling condition tends to evoke strong emotional reactions in affected individuals as well as significant others in their environment such as parents, spouses or sibs (Ross and Deverell 2004).

When parents receive the news that their child has been diagnosed with a disabling condition it, in most people, produces emotions of loss that are associated with grief. The parents face the dual, conflicting tasks of mourning the lost, hoped for perfect child and addressing the complex needs and emotional impact of the actual child (Weil 2000).

Loss in any form precipitates grief, which can include a range of behaviours, physical sensations and emotions which, though varied are normal. The intensity and duration of the grief varies according to the individual and the nature of the loss (Ross and Deverell 2004). The main stages of the grieving process were described by Elizabeth Kübler-Ross in 1969; these stages include denial, bargaining, guilt, depression, anger and acceptance. These stages have no linear order, but tend to occur in a repeated cyclical fashion, until a new equilibrium is reached (Falek 1984). The various elements of mourning are interrelated. There are shifts back and forth from one set of emotions and thoughts to another and the process is cyclical at the same time as it evolves (Weil 2000). Individuals may oscillate between two stages, and specific life crises may plunge an individual into an emotional state which has successfully been negotiated in the past (Ross and Deverell 2004).

Shock, numbness and disbelief

Among persons with sudden onset disabilities or gradually progressive diseases, shock, disbelief and numbness are common reactions to the initial diagnosis of the condition, especially if the diagnosis is not anticipated (Livneh and Antonak 1997). This is an involuntary psychophysiological process that buffers the emotional impact and prevents potentially more serious psychological disorganization. At this stage information may be heard but not accepted because it does not appear to make sense (Weil 2000).

Denial

Denial is a coping mechanism that individuals use to avoid the reality of a situation (Falvo 1991). Denial may manifest in various ways, it may be the rejection of the diagnosis, its permanence or its impact. Individuals who reject the diagnosis may argue with the professional who made the diagnosis or refuse to accept their views. Other individuals may quietly accept the diagnosis, but do not follow through on recommendations or do not cooperate with healthcare professionals involved in the management (Ross and Deverell 1997). During the early stages of adjustment denial

may be beneficial, as it allows the individuals to adjust to the reality of the situation at their own pace, thereby preventing excessive anxiety. However, if denial continues it can prevent individuals from following healthcare advice or recommendations that may be helpful (Falvo 1991).

Bargaining

The bargaining stage is characterized by fantasy thinking, individuals may try to bargain with themselves, others or even with God to make the loss go away (Weil 2000; Ross and Deverell 2004). They lack confidence in dealing with the loss and look elsewhere for the answers. Bargaining is used as an attempt to delay the acceptance of the illness, to revert from the present to the past, to negotiate for a less severe form of the illness, and to obtain a more positive therapy prognosis (Ross and Deverell 2004).

Guilt

Guilt may occur in varying dimensions and may be expressed or not expressed (Falvo 1991). Guilt is often linked to concerns about the cause of the condition, mothers who have sons with DMD often express feelings of guilt about having transmitted the disease to their sons (Gagliardi 1991). Guilt is a normal necessary part of the grieving process and usually ceases once individuals have been able to re-evaluate their existing beliefs and values (Ross and Deverell 2004).

Depression

Depression is considered to be a typical reaction upon the realization of the reality, seriousness and implications of a chronic illness or disability (Livneh and Antonak 1997). Individuals may experience feelings of depression, helplessness and hopelessness, apathy and/or feelings of dejection and discouragement. Signs of depression may include sleep disturbances, changes in appetite, difficulty concentrating and withdrawal from activity. The extent of depression may vary between individuals and not all individuals experience prolonged depression with chronic illness and

disability. However, when depression is unresolved it can result in self destructive behaviours (Falvo 1991).

Anger

Anger is an integral part of mourning, during this stage individuals may ask questions such as “why?”, “why me?”, and “what did I do to deserve this?” Anger may also result from frustration with the situation. Parents of children who are diagnosed with a disability may feel anger towards the child, but since anger towards an innocent child is socially unacceptable, this anger may be displaced onto spouses, siblings or professionals (Ross and Deverell 2004). Anger may also be the expression of the realization that the situation is serious and the associated feelings of helplessness. Sometimes anger may be hidden in the struggle to stay in control through quarrelling, arguing, complaining or being excessively demanding (Falvo 1991).

Anxiety

Parents who are confronted with the reality of a child with a disability often demonstrate feelings of anxiety and panic. This anxiety is usually related to determining the right balance between responsibility for the welfare of the child and the right to have an independent life (Ross and Deverell 2004). For some parents the fear of the unknown or unpredictability of the illness may provoke anxiety. Anxiety and fear may render individuals unable to act; helping these persons regain a sense of control over the situation may be an important step in reducing anxiety (Falvo 1991).

Acceptance and coping

Individuals move through the grieving process at different rates, some may never reach the stage of acceptance, while others will move through the process rapidly. Some of the following characteristics may reveal that parents have achieved acceptance:

- The child's limitations are able to be discussed without feeling vulnerable or threatened;
- A balance is demonstrated between encouraging independence and showing love;
- They are able to work with professionals to formulate short- and long-term plans;
- Personal interests unrelated to the child are pursued;
- Appropriate discipline is exercised without experiencing undue guilt; and
- They can refrain from being either overprotective or unduly harsh towards their child (Ross and Deverell 2004).

The amount of time that an individual needs to reach acceptance depends on the personality of the person, the reaction of family members, life circumstances, available resources and the types of challenges that are faced (Falvo 1991). Because of individual variability, spouses or partners often have different primary defences and coping mechanisms, and they may proceed through the phases and cycles at different rates (Weil 2000). Although people who have come to accept the illness or disability still experience pain and grief, they are no longer overwhelmed by the disorder, but have integrated it into their lives (Ross and Deverell 2004).

Previous studies have made certain recommendations for individuals working with parents of children with disabilities which may be of value in this setting. These suggestions are: (a) feelings of loss should be recognised as normal and even encouraged; (b) it should be recognised that denial or anger may be the best coping mechanism available to parents who are not able to progress to the acceptance stage; (c) parents should be allowed to proceed through the stages at their own pace; (d) the parents' feelings should be accepted and may be constructively channelled into activities; (e) parents may need help in seeing that there is some value in being, even when no overt progress is being made; and (f) parents' back and forth movement between stages should be accepted (Ziolko 1991).

2.4 PROBLEMS EXPERIENCED BY PARENTS OF SONS WITH DMD.

Firth et al (1983) interviewed 66 British families who had a son with DMD. The questionnaire covered various topics, including problems experienced by the parents, the diagnosis, neonatal screening and the affects of DMD on the family. The authors identified three main categories of problems mentioned by the participants; service related problems, practical problems and emotional problems. Sixty-two percent of the problems experienced by parents were practical problems of a physical nature such as lifting, bathing, toileting, dressing and feeding. Twenty-three percent of the problems were service related, namely, difficulties in obtaining social and medical services, assistive aids and allowances or dissatisfaction with them. Fifteen percent of the problems identified were emotional problems such as depression of the affected boy, parent's emotional problems in watching their son deteriorate, parental isolation and awareness of society's attitude to handicapped people. Many of the parents also commented on the lack of communication about the disease within the family and that this issue was of great concern (Firth et al 1983).

Fitzpatrick and Barry (1986) found that most parents reported a great difficulty in communication within the family about DMD. This included informing female relatives of their potential carrier status. A possible reason for the lack of communication within these families was their cultural background, as direct communication about feelings or emotions are reported to be particularly difficult for the Irish. In a further study done by Fitzpatrick and Barry (1990) marked differences were found between Irish and American DMD families in the manner in which they communicated about the disease within their families.

Twenty-five American families who had a son with DMD were interviewed by Buchanan et al (1979) to determine how they had adjusted to having a child with the disorder. In contrast to the findings of Firth et al (1983), Buchanan et al (1979) found

that when parents were asked to identify a major problem they were facing, 76% of families identified a psychological problem and only 16% of families identified a problem of a physical nature such as lifting, carrying or turning. One of the major psychological issues that were raised was the participant's anticipation of a future stressor, for example, the unpredictability of the disease. No parent mentioned death as a major concern, although it was implied. Other psychological issues mentioned included anxiety of having to explain DMD to their sons and the reaction of friends and relatives. Only one participant mentioned loneliness and depression of the affected boy as a major concern (Buchanan et al 1979). Twenty-seven percent of the participants experienced a strain on their marital relationship significant enough to threaten the union. The primary difficulty in these relationships seemed related to DMD, childcare, discipline, constant fatigue from caring for the child and interference from extended family. In contrast to this four of the families believed that their marriage was stronger as a result of having a son with DMD (Buchanan et al 1979). Chronic emotional stress was reported by these parents as the most significant problem they faced in coping with a child with DMD due to the unrelenting, constant demands of medical, physical and emotional care required by the disease.

The findings by Bregman (1980) illustrated the variety of problems and difficulties faced by parents of children with disabilities. She noted four coping strategies that are generally used by these parents: families focussed on the present by taking things 'one day at a time'; they attempted to live as normal a life as possible, and reduced the risk of crises by having a proactive attitude towards the care and services provided for the child; and families developed coping resources based on personal strengths. The most common coping strategy identified by Buchanan et al (1979) was denial of some form usually in the early stage of adjustment. They also encountered magical thinking where mothers believed that their son was different from other boys with DMD and didn't show the same symptoms. Most of the participants exhibited over protection of the child which also manifested in a conscious lack of discipline, with the underlying implication that the child had suffered enough (Buchanan et al 1979).

DMD has a significant psychosocial impact on parents. Mothers of boys with DMD experience significantly greater stress than mothers of healthy children. This could be attributed to the possibility of increased problem behaviours in social interactions of these boys rather than physical manifestations of DMD (Nereo et al 2003). In a study by Witte (1985) parents of boys with DMD showed marked preoccupation with their sons, great stress and diminished expression of enjoyment. The boys showed a great need to communicate about the disease with their parents, but the parents had great difficulty in discussing the progressive and terminal nature of DMD with their sons (Witte 1985).

Holroyd and Guthrie (1986) found that parents and families of children with DMD seemed to exhibit more stress than parents of children with cystic fibrosis, renal disease and the control group of their sample. They suggest that this stress may be due to the almost total dependence of boys with DMD, the fact that many of them also have learning difficulties, and the repeated cycle of loss, adaptation and repeated loss as their physical condition worsens (Holroyd and Guthrie 1986).

Abi Daoud et al (2004) found that parents of boys with DMD had a greater likelihood of experiencing a major depressive episode and had a significantly lower self-esteem score than the control group. Thompson et al (1992) found that 57% of the parents of sons with DMD in their study had self-reported poor psychological adjustment. It has also been reported that parents of sons with DMD have significant feelings of guilt and difficulty discussing death issues with their sons (Reid and Renwick 2001).

In contrast to the reactions found by Buchanan et al (1979) who found that parents used magical thinking, overprotection and lack of child discipline as coping mechanisms, and Holroyd and Guthrie (1986) who found that parents of children with neuromuscular disease are pessimistic about outcomes regarding the disease and have negative attitudes towards the children, Webb (2005) found that parents coped realistically and positively. Parents began by taking one day at a time, gathering as much information about the

disorder as possible, interacting with other parents, adjusting their priorities and slowly becoming involved in new projects (Webb 2005). Parents have become more proactive over time as indicated by their knowledge of the disorder and disease progress, how to get suitable support and where to get the most up to date results of research studies. Many families of children with disabilities report life changing benefits and most are able to cope effectively (Webb 2005).

There is a lack of current literature available exploring the problems faced by parents who have sons with DMD in SA. South African parents who have a son with DMD may have unique circumstances and to date, no research has been done to explore the problems they face.

2.5 GENETIC COUNSELLING

The most often cited definition of genetic counselling was written by a subcommittee of the American Society of Human Genetics:

“Genetic counselling is a communication process that deals with the human problems associated with the occurrence or risk of occurrence of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or family to: (1) comprehend the medical facts including the diagnosis, probable course of the disorder and the available management, (2) appreciate the way heredity contributes to the disorder and the risk of recurrence in specified relatives, (3) understand the alternatives for dealing with the risk of recurrence, (4) choose a course of action which seems to them appropriate in view of their risk, their family goals, and their ethical and religious standards and act in accordance with that decision, and (5) to make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder” (Fraser 1974:637).

Recently a contemporary version of this definition has been developed. According to Biesecker and Peters (2001:194) genetic counselling can be defined as a “dynamic

psychoeducational process centred on genetic information. Within a therapeutic relationship established between providers and clients, clients are helped to personalize technical and probabilistic genetic information, to promote self-determination and to enhance their ability to adapt over time. The goal is to facilitate clients' ability to use genetic information in a personally meaningful way that minimizes psychological distress and increases personal control.”

The main elements to be covered during genetic counselling sessions are:

- Diagnostic and clinical aspects;
- Documentation of family and pedigree information;
- Recognition of inheritance patterns and risk estimation;
- Communication and empathy with those seen;
- Information on available options and future measures; and
- Support in decision-making and decisions made (Harper 2004)

According to Gagliardi (1991) individual, family and genetic counselling are essential early interventions for boys affected by DMD and their families. Health professionals need to be sensitized to the array of losses experienced by the boys and their families. It is important to be aware of the needs of the families as the boys evolve through physical, social and educational changes.

2.6 LEVEL OF UNDERSTANDING OF GENETICS

The aim of the counselling component of a genetic service is to assist families who are affected in understanding and dealing with a genetic disorder. This includes understanding the diagnosis, prognosis, progression of symptoms and available management (Davey et al 2005).

This raises the question of the level of understanding of genetics gained during a genetic counselling session. Several studies have been undertaken to evaluate this with varied outcomes (De Pina-Neto and Petean 1999; Eggers et al 1999; Mitchie et al 1997b; Somer et al 1988). The most frequent interaction between genetic counsellor and client is the delivery of factual information; therefore it is very important to evaluate client knowledge gain (Mitchie et al 1997a). However, client information recall is often selective and interpreted within a personal frame of reference (Mitchie et al 1997b).

De Pina-Neto and Petean (1999) undertook a retrospective study in which, among others, one of their specific objectives was to evaluate the level of understanding or recall of information following genetic counselling. Their results showed that their sample of parents had a predominantly inadequate level of understanding of 48.7% with much lower frequencies of satisfactory (28.8%) and adequate (22.5%) understanding levels. Compared to similar studies (Clarke et al 1990; Somer et al 1988; Abramovsky et al 1980) the level of understanding of genetics is among the lowest in the literature.

In this study (De Pina-Neto and Petean 1999) there was a distinct relationship between the level of understanding and time elapsed between the genetic counselling interview and the follow-up interview. Knowledge of the genetic disease in the family decreased with time. The level of understanding was also found to be significantly related to social class; and in this study the authors found that low socioeconomic-cultural level was a major factor that reduced the level of understanding.

The comprehension of issues discussed during genetic counselling is significantly better among patients with a higher socio-educational level. Eggers et al (1999) reported that as well as socio-educational level, comprehension was higher among those individuals who had a family member affected by or deceased from DMD.

In a large study conducted in Finland (Somer et al 1988), with the aims of improving counselling services and determining the post counselling knowledge of families, the following results were obtained: 80% of the respondents had an adequate knowledge of the mode of inheritance and 74% knew the recurrence risk of the genetic disorder; the knowledge of recurrence risk for X-linked inheritance was the poorest (61%) and this mode of inheritance often needed extra clarification. Possible reasons for the good recollection of the respondents were that 85% of them reported high motivation for counselling, and the level of education of the counselees was higher than that of the general population. After a counselling session families received a written summary letter which may be a further reason for the good recollection of the respondents in this study.

Mitchie et al (1997b) assessed the level of knowledge 2-4 weeks after a genetic counselling session. They investigated how much and what type of information given is recalled and whether recall is affected by receiving a summary letter of what was discussed in the session. They found that the average recall for genetic and medical knowledge was 68-78% and recall for risk information was 74-78%. Recall did not appear to be associated with the patients' satisfaction with the information given, their anxiety, level of concern or the extent to which their expectations were met. It also did not appear to be influenced by the specific hopes that the patients had for the consultation or whether or not a summary letter was received. This is in contrast with the findings of Somer et al (1988), but Mitchie et al (1997b) noted that the fact that the summary letter did not appear to influence patient recall could be a methodological artefact. Those that had not yet received a summary letter were those patients who had been most recently counselled. This may have been compensated for by not receiving the letter (Mitchie et al 1997b).

2.7 SATISFACTION WITH GENETIC COUNSELLING SERVICE

Satisfaction with genetic counselling has been studied by various groups (Mitchie et al 1997b; Clarke et al 1996; Shiloh et al 1990) and it has been found that genetic counselling clients

generally report a high level of satisfaction. This has also been reported by consumers of most medical services (Williams et al 1998). According to Williams et al (1998) high satisfaction ratings don't necessarily mean that patients have had good experiences with the service, but may often reflect attitudes such as "they are doing the best they can".

Clarke et al (1996) noted that attempts to assess global levels of satisfaction are not helpful in identifying either the strengths or weaknesses of a service, but rather that specific aspects of performance should be focussed on. Shiloh et al (1990) developed a comprehensive measure of satisfaction with genetic counselling. Three components are measured namely instrumental, affective and procedural. The instrumental component is the extent to which the client is satisfied that the professional has the required skills and gives the necessary treatment. The affective component is the extent to which the client is satisfied with the professional's affective behaviour towards the client as a person, showing interest and care. The procedural component concerns the client's satisfaction with the administrative aspects of the service (Shiloh et al 1990). They found that the affective aspects (dedication, interest in personal problems and the way in which information is communicated) were most important in determining the general level of satisfaction of the clients in their sample (Shiloh et al 1990).

Davey et al (2005) conducted a study to evaluate the effectiveness of genetic counselling, using among other measures, client satisfaction with the service received. They used measures which were developed by Shiloh et al (1990), to evaluate the degree to which clients were satisfied with the counselling service they received. They found that the majority of clients were very satisfied with their overall experience with a mean score of (93%). For the instrumental, affective and procedural components the mean scores were, 69%; 71% and; 85% respectively. For most of the participants in the study, affective and instrumental components of their interaction were essential for the development of overall satisfaction with the service (Davey et al 2005).

Research has supported an association between increased client satisfaction and meeting client expectations (Bernhardt et al 2000, Mitchie et al 1997a). It is widely recognized that measuring client satisfaction is difficult because clients are less likely to have clear ideas of what to expect from a genetic counselling session (Davey et al 2005).

When a diagnosis of DMD is made one might expect a high level of dissatisfaction among parents because it is such an unwelcome message. Green and Murton (1995) found that nearly half of their sample of 157 families indicated that they were very satisfied with the way in which the diagnosis was given to them showing that it is possible to deliver such negative news in a satisfactory way (Green and Murton 1995).

They further indicated that the following variables were significantly related to higher levels of satisfaction with how the diagnosis was given:

- Understanding and remembering what was said;
- Getting adequate information;
- Mother's level of education;
- Presence of father; and
- Father's level of education (Green and Murton 1995).

Variables such as location and the presence of lots of people were important to individuals but were not found to be statistically associated with satisfaction. Other factors which one may expect to influence satisfaction, for example length of time between suspecting a problem and receiving the diagnosis and whether there was any previous suggestion of the diagnosis were not found to be related to satisfaction. What is required is a healthcare professional who displays empathy and sensitivity to parents' emotional and informational needs (Green and Murton 1995).

This literature review highlights many aspects which may have an impact on parents who have a son with DMD. This includes the clinical and psychosocial aspects, parents' understanding of the genetics of DMD, their satisfaction with the genetic counselling service and the variety of problems they face. The literature included in this chapter describes the experiences of parents from the UK, USA and other developed countries. No literature was available to describe the experiences of SA parents. The results of this study which are presented in Chapter 4 will attempt to describe the experiences of the participants of this study and compare it that discussed in this chapter.

Chapter 3

METHODOLOGY

University of Cape Town

3.1 INTRODUCTION

In this chapter the methodological process is described and aspects of 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.

3.2 RESEARCH DESIGN

This qualitative research project was designed as an interview-based cross-sectional descriptive study conducted in partial fulfilment of the MSc Genetic Counselling course.

3.2.1 Qualitative Approach

A qualitative approach was selected as it aims to understand and provide descriptions that portray the richness and complexity of real-life events that occur in natural settings from the perspective of the participants (McMillan and Schumacher 2001). The researcher attempted to examine the experiences, feelings and perceptions of the participants (McMillan and Schumacher 2001; Holloway and Wheeler 1996) and to describe social reality from the points of view of the participants (Grinnell 1988).

In this cross-sectional study data were collected at one point in time by means of interviews conducted with the participants, not at several points in time as is done in a longitudinal study (Grinnell 1988). This method is less costly and simpler than longitudinal research, but it does not capture social processes or change over a period of time (Neuman 1999).

Descriptive research is concerned with the current or past status of some phenomenon and provides valuable data, especially when first investigating an area (McMillan and Schumacher 2001).

3.3 SAMPLE

3.3.1 Population

There are more than 40 boys with DMD who attend the neuromuscular clinic at the RCWMCH. They range in age from 2 to 21 years of age.

For the purpose of this study the parents of 12 boys with DMD, who met the inclusion criteria and from a variety of different socioeconomic groups, were identified by the neurologist involved in the neuromuscular clinic at the RCWMCH.

3.3.2 Sampling method

Both convenience and purposeful sampling methods were used for the study. Convenience sampling is when participants are selected on the basis of being accessible (McMillan and Schumacher 2001). Only participants who regularly attended the neuromuscular clinic at the RCWMCH and lived in the Cape Town area were interviewed as they were accessible to the researcher with respect to time and cost restraints. Purposeful sampling allows participants who provide the best information to address the aim of the research to be selected (McMillan and Schumacher 2001). In purposive sampling collecting information-rich data and understanding the ideas of the sample population is more important than generalisability (Holloway 1997). The neurologist involved in the neuromuscular clinic identified the first 12 families who attended the neuromuscular clinic who she considered to be most representative of the boys and parents. More than 12 families fulfilled the inclusion criteria, but due to time restraints, only 12 were included in the study.

3.3.3 Inclusion and Exclusion Criteria

Participants were included in the study if they:

- had a son with DMD between the age of 8 and 15 years [This age range was selected because parents of boys under the age of 8 years may still be coming to

terms with a recent diagnosis of DMD and these boys are usually still ambulant and not severely affected by DMD. While boys over the age of 15 are entering end-stage DMD and the researcher may not have had sufficient experience to deal with the issues that may have arisen at this stage of the disease];

- regularly attended the neuromuscular clinic at RCWMCH;
- lived in the Cape Town metropolitan area; and
- agreed to participate and have their interviews audio-taped.

Participants were excluded if:

- their son with DMD was under the age of 8 years or above the age of 15 years;
- they were not willing to participate if the interview was audio-taped; or
- they did not respond after three telephone calls from the researcher in order to arrange suitable interview meetings.

3.4 STUDY METHODS AND MEASURING INSTRUMENTS

3.4.1 Interviews

A semi-structured interview schedule, designed by the researcher, was used to gather the data (Appendix II). Individual interviews tend to be more useful to explore personal experiences and perspectives especially on sensitive topics (Giacomini and Cook 2000). The interview combined both closed-ended and open-ended questions. Closed-ended questions were used to obtain sociodemographic information from the participants. Open-ended questions were used to encourage free responses without the limitation of preset categories, allowing the parents to express themselves more precisely and in greater depth. As there was a limited time to conduct this research, prompt questions were used to guide the discussion so that the maximum amount of information was gathered in the allotted time. Obsequiousness bias was minimized by asking all

questions in a neutral manner and responses were probed where they did not correspond with other responses (Sacket 1979).

Certain sections of the interview were adapted from interviews used in previous research. In the section regarding the participants satisfaction with the counselling service at RCWMCH, selected questions were adapted from the comprehensive measure of satisfaction with genetic counselling which was developed by Shiloh et al (1990). In this study (Shiloh et al 1990) it was recommended that a shorter version of the interview be used. These items were incorporated among others in this section. In the section of the schedule regarding the level of understanding of the genetics of DMD, questions were adapted from an interview used by Somer et al (1988). Content validity was established by asking two experts in the field to critically review the content of the schedule (McDowell and Newell 1996).

All schedules, information sheets and consent forms were available in English and Afrikaans. As the researcher is fluent in English and Afrikaans the interview was conducted in the language of the participants' choice. The language used was simple to ensure that there was no ambiguity and that it was easily understood by the participants.

3.4.2 Research setting

Interviews were conducted in a private venue of the participants' choice. Ten interviews took place at the homes of the participants, and two who could not take time off work, were interviewed in a private room at the RCWMCH neuromuscular clinic at a time that coincided with a routine clinic visit. The home environment was the preferred site as individuals are most likely to be more comfortable in their home environment when responding to some of the questions which they might find sensitive. A period of observation in the home environment may also give more information than is obtained in a formal clinic setting (Smith et al 1995) as it allows the researcher the opportunity of interacting with the participants and their families by gaining a deeper insight into their

daily lives. It allowed the participants an opportunity to discuss and ask questions of concern regarding the course of the disease of their sons and problems they were experiencing in their daily lives with the management of their sons. It allowed the researcher the opportunity to observe and take detailed notes of the environment; relationships and interactions between the participants; and critical events that were important and influenced the lives of the participants.

3.5 PROCEDURE

The neurologist at RCWMCH informed the parents of the purpose and method of the study at a routine clinic visit. The consenting individuals were then contacted by the researcher to arrange interview times and venues.

3.5.1 Pilot interviews

A pilot study was conducted with two participants to test the constructed interview schedule to determine if the items were easily understandable. Pilot interviews were used to check for bias in procedures, the interviewer, or the questions. The procedures in the pilot interviews were identical to those implemented during the study. The interviewer took note of any cues such as body language, facial expression or inappropriate responses which suggested that the participants were uncomfortable or did not understand the questions (McMillan and Schumacher 2001). Following the two pilot interviews the schedule was adapted to ensure the logical order of the items and certain questions were simplified to aid the understanding of the participants. The pilot study ensured that no ambiguous questions were asked and established how much time was needed to complete the interviews. In this study the interviews were found to take approximately 90 minutes. The data obtained in the pilot study was not included in the results.

3.5.2 Recruitment

Participants were recruited by the neurologist involved in the neuromuscular clinic at RCWMCH. She informed the individuals of the research and asked if they would be willing to participate. The neurologist then, with the permission of the individual, gave their contact details to the researcher. The first 12 individuals who met the criteria and were willing to participate were recruited for the study; two of these became part of the pilot study. If both parents were available they were interviewed together or if only one parent was available or in the case of single parents only one parent was interviewed. When the researcher contacted the participants she informed them that there should be no extended family members present at the time of the interview, unless the family member was directly involved in the care of the boy with DMD. If it was not possible to conduct the interview in the participant's home without extended family members being present, the interview was scheduled at a venue of the participant's choice. Interview schedules were sent to the participants prior to the interview to allow them time to think about the questions.

3.5.3 Data Collection

Written informed consent was obtained by the researcher before the interviews were conducted (Appendix I). This also included permission for each interview to be taped by means of audio recording. Signed consent was obtained from both parents if they were both present at the interview. Participants were reassured that:

- All information provided during the interview (which would be tape-recorded) would be kept confidential apart from a possible publication in a scientific journal where names would not be used;
- The information would not be discussed with extended family members; and
- Participation was completely voluntary and that they could choose not to participate or could withdraw from the study at any time without jeopardizing their access to the medical services to which they were entitled.

The interviews were conducted by the researcher personally in a private venue of the participants' choice. The interviews were tape recorded as this allows a more complete record than hand written notes taken during the interview and enables the researcher to concentrate on how the interview is proceeding (Smith et al 1995). Interviews were transcribed for analysis by the researcher.

Due to the sensitive nature of the questions a follow-up visit was made if the participants felt there was a need. The researcher contacted the participants the day after their initial interview to clarify any uncertainties and to arrange a follow-up visit. The second interview provided the opportunity for counselling the parents regarding emotional issues evoked during the interview.

3.6 DATA ANALYSIS

This qualitative study used content analysis to capture the richness of the themes emerging from the semi-structured conversation with the participants, rather than reducing their responses to quantitative categories, as is obtained in structured interviews and questionnaires (Smith et al 1995).

Qualitative data analysis is mainly an inductive process of arranging data into categories and identifying patterns or relationships among the categories. Most categories and patterns emerge from the data, rather than being imposed on the data prior to collection (McMillan and Schumacher 2001). The data are read and reread to identify and index themes and categories. These categories may centre on particular phrases, incidents or types of behaviour (Pope et al 2000). The data obtained from the interviews was transcribed and organized into themes and categories through content analysis of the actual words used by the participants, and not the researcher's preconceived hypothesis (Smith et al 1995).

Qualitative research does not aim to identify a statistically representative set of respondents. Simple counts are used which provide a useful summary of some aspects of the analysis (Pope et al 2000). The responses of certain sections of the interview schedule were categorized by frequency and descriptive statistics such as percentages, means and averages were calculated to describe the occurrence of the data.

3. 7 TRUSTWORTHINESS/VALIDITY

In qualitative research trustworthiness is used to measure the validity of it. The research is trustworthy when it reflects the thoughts and reality of the participants. Trustworthiness exists when the findings of a qualitative study represent reality. Qualitative research recognizes that objective reality and subjective experiences possibly occur together in the research data (Holloway 1997).

According to Lincoln and Guba (1985) trustworthiness involves the following elements: credibility, transferability, dependability and confirmability. These four alternatives provide the foundation for demonstrating trustworthiness in qualitative research.

- 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. The researcher met regularly with her supervisor to ensure that the data obtained was interpreted in the same way.
- Transferability means that the findings in one context can be transferred to participants or situations that are similar (Holloway 1997). By describing the data accurately and in detail peers and readers are able to decide if the findings described may be transferred to other settings or participants.
- For a study to be dependable it must be consistent and accurate. This was demonstrated by providing detailed descriptions of the methodology so that readers can follow the path of the research (Holloway 1997).

- The last element of trustworthiness is confirmability. Confirmability means that the findings of the research are due to the results and not the 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 telephone calls and interviews to confirm that the participants agreed with the researcher's interpretation of certain aspects of the conversation that might have been meant differently. The findings of the study were also discussed with the researcher's supervisor.

3.8 ETHICAL CONSIDERATIONS

3.8.1 Ethical approval

This study was granted approval without reservations by the Medical Research Ethics Committee of the University of Cape Town (Reference number 360/2005) (Appendix III).

3.8.2 Consent

The participants were approached by the neurologist involved in the neuromuscular clinic at the RCWMCH who explained the purpose of the research to be conducted. 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 future medical management at the neuromuscular clinic.

3.8.3 Confidentiality

As confidentiality was of central concern, audio-taped recordings were transcribed as soon as possible after the interview. The audio-tapes and transcriptions 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, the tapes, the transcripts or spreadsheets.

3.8.4 Risk Benefit

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. The researcher was sensitive to the emotional state of the participants throughout the interviews. Participants had the opportunity of a second session where any emotional issues evoked during the initial interview were dealt with or any questions that the participant had were answered.

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 that attend the neuromuscular clinic at RCWMCH.

3.9 ASSUMPTIONS

The researcher assumed that the responses of the participants were honest and a true reflection of their lives.

3.10 LIMITATIONS AND STRENGTHS OF THE STUDY

3.10.1 Limitations of the study

- A limitation of the study was the small sample size. The qualitative interview schedule was time consuming and as a result of the time constraint only a small sample could be used. The results may not be able to be generalized to a larger population, but will only be valid in a population with similar characteristics;
- Data were gathered only at one point in time. This is a limitation of descriptive studies as changes in the sample could not be measured. As the boys in this study

were maturing and their symptoms were progressing it is likely that even six months later the problems experienced by them and their parents could have been much worse and of a different nature;

- A further limitation was evaluating the genetic counselling service by asking people who regularly attend the neuromuscular clinic for their opinion. It may be more valuable to gather data from the parents who do not regularly attend the clinic;
- The researcher had limited interviewing and counselling experience and, therefore might not have had the skills to gather the maximum information from the interviews;
- Not all participants received the interview schedule in the mail prior to the interview. This gave them limited time in which to think of their responses; and
- The paucity of information, literature and research concerning DMD in SA, means that there is no point of reference or comparison.

3.10.2 Strengths of the study

- The researcher was an outsider to the clinic team and therefore, had no vested interest or private agenda for personal gains. The participants were also able to discuss negative aspects of the programme more freely than with people they were accustomed to seeing at the RXWMCH neuromuscular clinic;
- The researcher conducted all the interviews personally;
- The interview schedule contained open-ended questions which allowed participants to express themselves freely without the limitation of preset categories;
- Certain sections of the interview schedule contained questions used in similar international research. This increased the trustworthiness of the study;
- All interviews were audio-taped to allow a more complete record than hand written notes by the researcher;

- Interviews were conducted in the participants homes where they were more likely to feel at ease about responding to emotional issues than in a clinical setting where their emotions are usually controlled; and
- Researcher bias was minimised as subtle signs and intimations during the interviews were followed up by in-depth questioning by the researcher which might have, to a certain extent, overcome this bias.

University of Cape Town

Chapter 4

FINDINGS AND DISCUSSION

University of Cape Town

4.1 INTRODUCTION

The findings of the research are presented in this chapter. Data are presented in tables followed by a discussion and when possible, reference is made to the literature in order to demonstrate similarities and differences to other studies on DMD. In total 10 interviews were conducted with the parents of boys with DMD.

4.2 INTERVIEW PROCEDURE

Six of the interviews were conducted with only the mother of the affected boy present. Fathers and mothers were present at two interviews. One interview was conducted with an aunt of a boy whose mother died two years prior to the study, and was now his primary caregiver. One interview was conducted with the mother and aunt of the affected boy as the aunt was directly involved with his care.

All of the interviews took less than one and a half hours to complete. None of the participants expressed the need to have a second interview. Nine of the interviews were conducted at the participants' homes. One interview took place in a private room at the RCWMCH at a time that coincided with a routine clinic visit, as the participant could not take time off from work.

For ease of reading and to ensure confidentiality the participants will be referred to by number throughout this chapter. The participant number refers to the adult/s who were interviewed. The researcher has included direct quotes of the participants' responses in order to provide the reader with greater insight into their thinking.

4.3 SOCIODEMOGRAPHIC INFORMATION OF PARTICIPANTS

A summary of the sociodemographic information of the participants is presented in Table 4.1 (p 37).

Table 4.1: Summary of the sociodemographic information of the participants

P. No	Age of affected son	Marital status	Level of education		Current occupation		Household income per month
			Mother	Father	Mother	Father	
1	12	Single	Gr 9	N/A	Unemployed	N/A	R780
2	15	Married	Gr 10	Gr 10	Temp	Sales representative	R1600
3	12	Married	Gr 8	Gr 10	Housewife	Builder	R1000
4	13	Remarried	Gr 9	N/A	Kitchen Staff	N/A	R2000
5	14	Married	Gr 7	Gr 7	Unemployed	Security guard	R2780
6	13	Married	Gr 12	Tertiary	Housewife	Industrial relations manager	R25 000
7	14	Married	Gr 12	Gr 12	Housewife	Restaurant owner	R13 000
8	11	Engaged	Gr 9	Gr 8	Data capturer	Handy man	R 2780
9	10	Single	Gr 12	N/A	Unemployed	N/A	R780
10	15	*	*	*	*	*	R3000

*Participant 10 was the aunt of the affected boy who was his foster parent. His mother died 2 years prior to the study and it is not known who his father is.

As Table 4.1 illustrates most of the participants had not completed high school. Only one participant had completed tertiary education while two participants had only completed primary school.

The monthly household income of the participants was very variable. Many participants were casual workers and could only estimate their monthly income. Eight of the participants had a monthly household income of R3000 or less, only two had an income above this. P6 and P7 were of a higher socioeconomic group than the other participants. According to Statistics South Africa (2001) the average annual household income in South Africa in 2000 was R45 000 (R3750 per month). Most of the participants had a monthly income of below the average income in SA. P1 and P9 relied solely on the disability grant of the affected boy. Both were unemployed single mothers.

Table 4.2 illustrates the size of the households that were supported by the monthly income.

Table 4.2: Number of people supported by the monthly household income.

Participant No.	Household income	No. people supported
1	R780	2
2	R1600	5
3	R1000	5
4	R2000	3
5	R2780	5
6	R25 000	4
7	R13 000	6
8	R 2780	3
9	R780	6
10	R3000	8

P9 was an unemployed single mother who lived with her unemployed sister and her two children. Their father also lived in the house with them. At the time of the study the six of them relied solely on the disability grant of the affected boy although his mother was looking for work. P10 was the aunt of the affected boy whose mother had died. She and her husband were taking care of him and his younger sister, as well as their own son and daughter and her two children.

4.4 RCWMCH NEUROMUSCULAR CLINIC

Participants were asked various questions regarding the RCWMCH neuromuscular clinic to gauge its accessibility. Table 4.3 (p 39) illustrates some of the important aspects of the clinic as well as comments made by the participants.

Table 4.3: Accessibility of RCWMCH neuromuscular clinic

P No.	Referred by	Clinics attended	How often	Cost	Public transport	Travelling time	Comments
1	Crossroads Medical Centre	Neuromuscular	3 mnths	R22	Yes	1 hr	Difficult using public transport
2	GP	Neuromuscular and cardiac	3 mnths	R35	Yes	45 min	Difficult using public transport
3	Gatesville Medical Centre	Neuromuscular	3 mnths	R30	Yes	1 hr	Public transport difficult
4	Harare Day Hospital	Neuromuscular	3 mnths	R40	Yes	1 hr	Public transport difficult
5	Mitchells plain Day Hospital	Neuromuscular	3 mnths	Free	No	2 hrs	School transport
6	Developmental paediatrician	Neuromuscular	6 mnths	Unknown	No	30 min	None
7	GP	Neuromuscular and cardiac	6 mnths	Unknown	No	30 min	None
8	Local clinic	Neuromuscular	6 mnths	R40	Yes	3 hrs	Bus difficult
9	Manenberg clinic	Neuromuscular and cardiac	6 mnths	R20	Yes	45 min	Taxi doesn't take wheelchair
10	RCWMCH	Neuromuscular	6 mnths	R100	Yes	1 hr	Dial-a-Ride difficult

When participants were questioned regarding the accessibility of the neuromuscular clinic seven of the ten participants commented on the difficulty of using public transport although this was not asked directly. P1, P2, P3, P4, P8, P9 and P10 stated that it was very difficult to use public transport when attending the clinic. Some of the difficulties that participants mentioned included the inaccessibility of buses and taxis and the unwillingness of taxis to stop if the child was in a wheelchair. Many participants commented on this aspect and the frustration that it caused, as frequently they were left without transport which resulted in them missing a clinic visit.

When asked how far the clinic was from the house P4 responded as follows:

“It is so difficult for me to take him there. Sometimes in the morning, you have to take him to the taxi rank and the people at that time are in a hurry to go to work and sometimes, I wait and wait for transport.”

The costs involved are the estimated costs of the participants for each clinic visit. The participants who used public transport commented that these costs were substantial. Two participants (P9 and P10) had tried to contact Dial-a-Ride without success. In one case the telephone was not answered after repeated attempts and the other participant found them to be very unreliable resulting in them missing clinic visits.

The travelling time involved in getting to the clinic was also commented on as participants who used the bus had to walk a moderate distance from the bus stop to the hospital. Some of the participants had to carry their sons this distance and those that had wheelchairs commented that this was also problematic.

Participants also mentioned concern for their safety while using public transport. P1 had been robbed twice. When asked how far the clinic was from home she replied as follows:

“It is very difficult for me because I have been robbed twice on my way to hospital, I have to carry him down to the taxi rank and then I have to stand and wait...”

Three participants (P5, P6 and P7) did not rely on public transport to travel to the clinic. These participants were not aware of the costs involved in getting to the clinic, nor did they experience any of the difficulties that the participants using public transport expressed. This can be related to the socioeconomic level of the participants. As they had a higher income than the other participants these issues were not concerns for them.

Although P5 was not of a higher socioeconomic level, the school that the affected boy attended provided transport to the clinic.

4.5 SATISFACTION WITH COUNSELLING SERVICE

The participants were asked to rate their level of satisfaction with specific aspects of the service they received at the neuromuscular clinic at RCWMCH in categories from “very unsatisfactory” to “highly satisfactory”. Their responses are presented in Table 4.4.

Table 4.4: Frequency of responses in each category. (n=9*)

	Very unsatisfactory	Unsatisfactory	Satisfactory	Very satisfactory	Highly satisfactory
1. The way in which DMD was explained to you.		2	4	3	
2. The counsellor listened to what I had to say.		1	3	2	3
3. The counsellor understood what was worrying me.	1	1	5	2	
4. The counsellor showed a lot of caring.		1	5	3	
5. The counsellor answered all my questions.		1	4	3	1
6. Counselling helped cope with your problems.	1	1	4	2	1
7. There was enough time given at each session.			5	5	
8. The clinic environment was informal.	1	3	4	1	
9. There were no disruptions during the counselling.		1	8		
10. The way the test results were presented to you.	1	1	4	3	
11. Time in the follow up visits for questions to be answered.			7	1	1
12. Time waited to get first appointment.			9		
13. Time waited to see staff at each visit.		1	8		
14. Rate satisfaction with muscle clinic in general.		1	3	4	1
15. Follow up visits gave a lot of support.		1	6	2	

*Participant P10 was not asked to respond to these questions as she was not involved with the affected son at the time of the diagnosis. She and her husband had taken over the care of him and his sister only for the two years prior to the study.

Two participants P5 and P7 felt that DMD was not explained satisfactorily. They felt that they could have been given more information regarding DMD. P7 felt they had to resort to the internet for adequate information.

P7: "I remember going in and all the genetic stuff was explained, but I was the only one who received that, which I don't feel was very thorough...because we had to go looking, we turned to the internet for the information we needed."

P9 found many aspects of the service at RCWMCH unsatisfactory. She rated items 1, 2, 3, 4, 5, 9 and 10 as unsatisfactory. She was the only participant who found items 2, 3, 4, 5, 9 and 10 to be unsatisfactory, but when asked to rate her satisfaction with the RCWMCH neuromuscular clinic in general she said she found it to be satisfactory.

The items that were scored as very unsatisfactory (Items 3, 6, 8 and 10) were all rated by the same participant (P5). P5 felt that nobody really tried to understand what was worrying her and commented that she felt very uncomfortable in the clinic environment.

Three participants, including P5, found the clinic environment uncomfortable. P2 commented:

"It's just that there are so many people there, doctors, physiotherapists and other people. When he is around so many people he gets shy and very embarrassed and he doesn't want to be carried in front of strangers..."

Only one participant P8 felt that the clinic was not satisfactory. She commented as follows:

"All they do there is ask how we are, check his weight and make him blow in this thing...that's all they do, there is nothing they can do. There is no cure for his condition,

and man, I suppose that's all they really can do for him right now. They do the best they can, but it doesn't stop me from getting frustrated."

Most of the participants indicated that they were satisfied or very satisfied with the service they received at the RCWMCH neuromuscular clinic. The questions that were asked in Table 4.4 (p 41) attempted to assess the participants' level of satisfaction with specific aspects of the neuromuscular service at RCWMCH. Participants found it very difficult to rate the questions in a specific category and there was a tendency to expand the answers with explanations. In addition to the specific questions that were asked in Table 4.4 (p 41), participants were asked questions to determine how they experienced the time of diagnosis. Their responses are summarized in Table 4.5 (p 44 and 45).

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Table 4.5: A summary of the participant's experience of the time of diagnosis.

P no.	First symptoms	Reaction to diagnosis	Receive Counselling?	DMD confirmed with DNA
1	Walking funny '98. Became a problem in 2003 age 10yrs.	Felt very bad - thought he would be normal. Pictured him as he was younger.	Remembers being counselled.	Remembers blood being taken, not aware that it is genetic test.
2	When he started walking he seemed "off balance."	"Worst day of my life." Kept thinking other children are worse off.	Yes, asked for detail. "When you are told bad news you don't really know what you are thinking." Did own research.	Doesn't remember. "I don't know...everything happened together."
3	Used to fall a lot became a problem age 7yrs.	(F)Had to accept and make the best of it. (M) Difficult as a mother as it wasn't expected. Hard to accept because he is oldest and only son.	Remembers being counselled.	Remembers blood being taken.
4	Took very long to get up, always tired. Teacher hit him for being lazy (age 10yrs).	Felt very bad - thought he was going to grow up and help mother. Took him to witchdoctor to confirm diagnosis.	Remembers being counselled.	Remembers blood being taken.
5	Walking funny age 6yrs. Always walked on toes.	Thought something could be done to make muscles stronger (Still does).	Says never received any information.	Remembers blood being taken - nobody explained reason for it.

6	Always thought something was odd even as baby. Walked late and couldn't climb.	"As if the sky landed on my head." Had to carry on there are always people worse off.	Yes, helped a lot.	Understood DNA test.
7	Couldn't climb, couldn't run, had no motor coordination, huge muscles.	Numb, shock and disbelief. Floods of tears. Think it will never happen to you. Changes your life. Learn to semi cope with it, don't think you ever cope completely.	Explained everything but didn't feel it was very thorough. Turned to internet for information.	Understood DNA test.
8	Walking funny, limping, waddling.	Strong person, just had to deal with it. Very supportive family.	Yes, but did own research as well.	Understood DNA test
9	Thought something was wrong with his leg. Couldn't walk properly and couldn't run and jump.	Didn't know much about DMD. First time heard of it. Very difficult but he made it easier, very open with him.	Remembers being counselled, did own research too, on internet.	Understood DNA test, knows couldn't find cause and had to do muscle biopsy.
10	Walked with difficulty and fell on his head often. Had TB when younger - that's what caused problem.	Not involved.	Not involved.	Not involved.

*P10 was not involved in the care of the affected boy at the time when the diagnosis was made and was not asked to respond to these questions.

When P4 was told of the diagnosis she took her son to a witchdoctor to confirm the diagnosis. When asked how she reacted when she was told what the diagnosis was P4 responded as follows:

“I was feeling so bad because I thought my children were going to grow up and take care of me... I took him to the witchdoctor also because I didn't understand what was happening to him, you see. I just take him to a witchdoctor to go and answer what is happening to my child.”

Eight of the participants remembered receiving counselling when the diagnosis was made. Only P5 did not remember receiving any counselling although the affected boy's hospital notes clearly reflect that she did receive counselling. Four of the participants (P2, P7, P8, and P9) felt that it was necessary to do their own research after receiving counselling. The reasons given for this were that at the time they did not know enough about DMD to be able to ask questions and they were so emotionally overwhelmed that they did not absorb all the information that was given to them. Information seeking is a normal psychological defence in anxious individuals and can be turned to the parents' advantage (Buchanan et al 1979).

When asked if she remembered receiving counselling P2 replied:

“You know when you get bad news you don't really know what you are thinking, you don't want to hear what is good for you, you just think about the bad parts.”

4.5.1 Discussion of satisfaction with service at RCWMCH

The findings in this study were similar to those of Green and Murton (1995) who found that more than half of the participants in their study were satisfied with the way the diagnosis of DMD was made. In this study four of the nine participants found the way the test results were presented was “satisfactory” and three found it “very satisfactory”, demonstrating that it is possible to deliver bad news in a satisfactory way.

The two participants who were not satisfied with the way in which the test results were presented were P5 and P7. P5 did not remember receiving counselling although it was documented in the hospital notes. Both she and her husband had the lowest level of education among the participants having only completed Gr. 7. Although participant P7 had a high level of education she was on her own when the test results were presented and felt that she did not receive enough information. According to Green and Murton (1995) level of education, presence of the father, and remembering and understanding information are important variables that determine the rate of satisfaction with genetic counselling.

Most participants in this study had very specific concerns regarding the development of their sons, namely excessive falling, difficulty climbing stairs, inability to run and jump and an unusual gait which distinguished them from those with more general developmental delay. Participants in the study by Green and Murton (1995) expressed similar concerns. Another similarity between the studies is that the occasion at which the diagnosis was made seemed to be vividly remembered by most of the participants as was illustrated by their comments regarding their reaction to the diagnosis (Table 4.5 p44 and p45). The earliest and most common reactions to the diagnosis included various stages of the grief process. Although the message that was being delivered to participants was distressing most participants felt that it was done in a satisfactory way. This may be related to the amount of empathy they perceived the consultant to be displaying and was illustrated by the high satisfaction ratings of Items 1-5 of Table 4.4 (p41). Green and Murton (1995) also commented on the necessity of empathy and sensitivity to parents' informational and emotional needs.

The results of the current study are similar to other studies that report a high level of satisfaction with genetic counselling (Mitchie et al 1997; Shiloh et al 1990; Bernhardt et al 2000). However Shiloh et al (1990) report some clients may be disappointed because some of their questions can not be answered or because they may be told information

that they do not want to hear. P9 said that she was dissatisfied with the clinic, but expanded on this by saying that there was nothing more they could do for her son as there is not a cure for DMD, and they do the best they can at the clinic. High satisfaction ratings (or in this case low satisfaction ratings) do not necessarily mean that patients have had good experiences in relation to services, but often reflect attitudes such as “they are doing the best they can.” Williams et al (1998) highlighted that experiences described by users in positive or negative terms do not necessarily correlate with the users’ evaluation of the services which produced those experiences.

4.6 LEVEL OF UNDERSTANDING OF GENETICS

Many studies have been conducted to determine the level of understanding of genetics (De Pina-Neto and Petean 1999; Eggers et al 1999; Mitchie et al 1997b; Somer et al 1988). In this study participants were asked a range of questions that included questions that were adapted from a study done by Somer et al (1988). The responses to selected questions are presented in Table 4.6 (p49).

Table 4.6: Participants understanding of the genetic aspects of DMD.

P No.	Cause of DMD	Inheritance	Recurrence Risk	Male/females	Feelings re TOP
1	Can't walk anymore.	Comes from me (mother).	If another boy he will also be affected.	Only boys.	N/A.
2	Genetic-in the genes.	Comes from me (mother).	Very high chance. Abortion was suggested.	"One of my sons could get married and have a child with problem."	Not an option (believes it is murder).
3	Not sure but it is a family disease. "Doctors did explain but used words we didn't understand."	Not sure. Know that if their daughters have boys there is a chance of them having DMD.	Another boy will have same problem.	Only boys.	N/A.†
4	She was poisoned by child father's girlfriend while pregnant.	Didn't understand.	Was told by doctor that other children wouldn't have DMD.	Not sure.	N/A.†
5	Problem with muscle.	"From me or my mother-but there is nothing wrong with us."	Possible but I didn't want more children.	Boys and girls.	N/A.†
6	Genetic mutation.	"I'm not a carrier but my daughter may be."	50/50.	Only boys.	Not for abortion.
7	Gene passed on by maternal predecessors. Genetic thing caused by genes.	Knows there is a chance of her daughters being carriers - not sure of percentage.	Not sure.	"Manifests only in boys."	Would choose not to terminate.
8	Loss of muscles, muscle weakness.	"Comes from me, my gene, my chromosome" (mother).	Another child would be tested from day 1.	Only boys have it but girls are carriers.	Wouldn't be able to cope with another affected boy (TOP option).
9	"The problem actually comes from father's side, but I am the carrier."	"He is the genetic, and I am the carrier."	If another boy he will also be affected.	Only boys.	N/A.†
10	Mother didn't take him to hospital for TB treatment when he was small.	N/A.*	N/A.*	N/A.*	N/A.*†

* P10 was not asked these questions as she was not involved in the initial counselling.

†These participants were not aware that prenatal diagnosis was available.

4.6.1 Misconceptions regarding the genetic nature of DMD

Only three of the participants were aware of the genetic nature of DMD. Even among these three participants there were various other misconceptions. Among the other participants there were many inaccurate responses.

P3 thought DMD was a family disease as the husband's sister was also born with a disability and was confined to a wheelchair. They were, however, aware that there may be implications for their daughter's children. They also felt that the counsellors used language and terminology which they did not understand.

P4 did not seem to have any understanding of the genetic nature of the condition. When the diagnosis of DMD was made, she took her son to consult a witchdoctor. She was told that while she was pregnant with her son, her husband's girlfriend had poisoned her so that she would not give birth to the first son. According to P4, her son was supposed to die, but because the poison did not work as it should have, he was born with this disability.

When P4 was asked what the witchdoctor had explained the cause of the problem to be P4 explained as follows:

"That lady just put poison in something at the time that I was carrying my child. You know, in our culture we believe that if your boyfriend is getting another girlfriend, she will be fighting that you mustn't get that child. I was coming before her, but now she was going to give me something to kill my son. I did take some medicine from him (witchdoctor) but this medicine takes to long to make my son go back to normal."

P9 believed that the DMD came from the father's side of the family. She and the affected boy's father were never married and, according to P9, they didn't have a good relationship as they were frequently in court regarding maintenance money. She said,

however, that she knew she was a carrier. When probed about this the aunt who was also present at the interview explained as follows:

"This Duchene's comes from his father's side. I always knew it was bad news them being together...you see there are disabled children in his family but they don't have Duchene's...you see the thing is he is the genetic and she is the carrier."(Translated)

When asked about the recurrence risk they replied as follows:

"If she has another baby and it is a girl, she will be normal, but she may be a carrier and one day if that girl has a son he can have Duchene's."(Translated)

Participant P10 was the aunt that had taken over care of the affected boy and his sister. She believed that his condition was due to negligence on the part of his mother. When he was younger he had tuberculosis and was not taken to the hospital soon enough for treatment and this led to his problem. She also said that he had fallen on his head often when he was younger and this may also have been a part of why he was now in a wheelchair.

P10: *"They didn't tell me what the cause of his problem is, I assume it is because his mother didn't take him to hospital when he had TB when he was younger."*

4.6.2 Discussion of the general understanding of genetics.

Most previous studies have found participants to have a good level of understanding or recall of the information. Clarke et al (1996) stated that participants in studies in Europe and America usually show a good level of understanding. Somer et al (1988) found that in Finland 80% of the participants correctly understood the mode of inheritance and 74% had correct knowledge of the recurrence risk of the disorder. Abramovsky et al (1980) reported that 72% of respondents correctly remembered the recurrence risk of the disorder and 56% could recall the mode of inheritance.

Contrary to these findings the participants of this study had a predominantly inadequate level of understanding of the genetic aspect of DMD. Only three of the ten participants knew that a genetic mutation is the cause of the DMD. Five of the participants were aware of the inheritance of DMD, but described it in their own words as “coming from the mother.” The extent to which family members are aware of conditions that run in the family depends not only on communication within the family and the nature of the condition but also the pattern of inheritance that they observe and live in their family (Marteau and Richards 1996). None of the participants used the term “X-linked inheritance”.

There were many misconceptions regarding the recurrence risk of DMD as is illustrated by Table 4.6 (p49). Six of the participants were aware that DMD only manifests in males. All the participants except P5 remembered receiving counselling regarding the diagnosis (Table 4.5; p44 and 45), although most participants still did not have much understanding of the cause of the DMD or the inheritance and recurrence risk. Only one participant P3 verbalized that she had not understood the medical terminology that was used when the diagnosis was explained. This was also found by Davies et al (2002). Genetic terminology is generally less intelligible to lay people and this may lead to limited knowledge and misconceptions (Chapple et al 1997). It is important to note that P3 had expressed that she was satisfied with the way in which DMD was explained, although she later added that she found some of the medical terms confusing and difficult to understand.

The findings in this study are similar to those of De Pina-Neto and Petean (1999) who found that the participants in their study had an inadequate level of understanding. They found adequate levels of understanding were related to social class of the participants. In this study, level of understanding was related to socioeconomic status as well as level of education. The way in which the participants described the disorder was dependant on the socioeconomic level to which they belonged and was interpreted within a personal frame of reference. This has been mentioned by Mitchie et al (1997b) and De Pina-Neto

and Petean (1999) who discussed the relevance of traditional medicine in third world countries. In these countries underdevelopment causes marked social differences, and among people of lower socioeconomic level, there is less penetrance of scientific medicine. Due to the persistence of traditional medicine a large number of people do not know how to explain what happened in their family (De Pina-Neto and Petean 1999). This is clearly illustrated by P4. Individuals often draw on other belief systems which may be held in addition to scientific explanations. The genetic explanation may be accepted on one level, but the personal meaning of the situation comes in a large part from other beliefs (Weil 2000). Participants with little formal education may benefit from additional counselling. Although it is possible that some may still not understand genetic concepts after several sessions, it is nevertheless the counsellor's obligation to try and find ways of communicating this complex information (Somer et al 1988).

4.7 EXPERIENCES OF PARENTS

Participants were asked to identify what they experienced as a major problem in their everyday life. Only then were they asked to identify the practical and emotional problems. Table 4.7 illustrates the various responses of the participants.

Table 4.7: A summary of problems identified by the participants

P No.	Major problem	Practical problem	Emotional problem	Sons problem (parents opinion)	Means of psychological support
1	Doesn't have a job-financial implications	1.Lifting 2.Turning 3.Bathing	Hurts to think he will never walk again.	He wants to play but he can't.	Brother very supportive. "I'm just happy that he isn't sick sick. our life is difficult but there are many others whose lives are more difficult."
2	He wants to go out but he doesn't want to be seen in a wheelchair.	Feel they cope with practical things.	Don't like being asked questions in front of the children. "Adults can ask stupid questions; just as I've come to terms to with it they rub it in my face."	"Acceptance" Appears to take it in his stride but I know he doesn't want to be seen in his wheelchair.	"Always think to myself there are people who are worse off" "God is bigger than this thing." Helps knowing he is happy at school.
3	Toilet, shower and putting to bed. [Lifting]	Lifting.	Could not identify.	Having to ask others to do everything.	Communication within marriage.
4	Worried that she spends too much time at work and not enough with son. Her new husband doesn't want to support son.	House is too small.	"I think to myself. maybe if I didn't get him from his father he wouldn't have this problem."	Difficult to ask for everything.	Feels very alone. as if there is no one to support her. Although the school is supportive and he is happy there.
5	"Sometimes there isn't money for food or electricity, but we make a plan."	Could not identify. "It is a pleasure to help him."	Son is teased by the children in the street.	He is very lonely and wants to play with his brother but can't.	Religion – "I pray for strength everyday."

6	Loss of spontaneity have to plan everything.	Bathing using the hoist.	Worried that younger daughter suffers due to DMD.	Frustration at not being able to get around.	"Being on the MDF committee. And when I think of the difficulties other boys/families with DMD have I am very grateful."
7	Sleep deprivation.	Lifting.	"I find it difficult to talk to people about this, because I don't like people feeling sorry for me." "Don't have many friends due to the DMD and the fuss that needs to be made." Guilt at having passed it on.	Frustration and sense of being a burden. Biggest problem is other's attitudes towards him (strangers).	Relationship with God. Knowing that there are many people who have worse things to deal with- grateful. Muscle clinic is very supportive.
8	Doesn't feel anything is a major problem.	Dressing him is very tiring.	Sometimes ask myself "why" "why him, I see him look at other children and think I can't do that."	Having to ask other to do simple things like pick stuff up.	Supportive family and "Research I did to understand so that I can explain it to others."
9	Lifting - weight is problem.	Lifting.	When he gets hurt.	Having to ask for something and then wait until someone can help him.	Supportive family and being on the MDF committee (Aunt). "He makes it easier for us; he was very young when he was diagnosed, so he never played soccer etc. so it is easier for him to accept."
10	Children that tease him - he gets very upset.	Lack of sleep.	His temper - when he gets angry he behaves badly.	Thinks he will walk again.	"Myself. I get very little support from family." (They want to her to send him away)

Participants were questioned about communication within the family about DMD and the response of the affected boys' siblings; certain responses are presented in Table 4.8.

Table 4.8: Communication about DMD within the family

P No.	Reaction of siblings	What has son been told	What have siblings been told
1	Very protective. Normal relationship.	"Told him he won't be able to walk again, because his muscles are weak, but I will always be here for him."	Explained that his muscles are weak.
2	Act like normal brothers but they understand he is special and needs to be taken care of.	"Knows about DMD." "Was once told at school that he was going to die."	Tried to explain, they understand that he has very weak muscles.
3	Too young to understand, normal relationship.	Told him he won't be able to walk again, because his muscles are weak.	That their brother has weak muscles.
4	Too young.	Only what he has been told at school.	N/A.
5	N/A	Haven't discussed it.	N/A.
6	Sibling stuff, but they get on very well.	Not really, just answers questions and has told him muscles are getting weaker.	Just muscle problem.
7	"Younger sister would prefer him to be invisible" Mom feels this is very shallow. Younger sister grew up knowing that she carries the gene. Older siblings feel family restricted in outings but are very protective.	Told about condition and questions are answered.	Understand and have been told everything.
8	Only child.	Answer questions as honestly as possible, told him he has weak muscles.	N/A
9	Only child.	Told him he is sick and that he has weak muscles. "But he has a one track mind that he will walk again."	N/A.
10	Doesn't understand just accepts that he must be in a wheelchair.	Told him he will become weaker until he won't be there anymore.	N/A.

4.7.1 General problems identified by participants.

When asked to identify the major problem in their everyday life six of the ten participants identified a practical problem. Three participants identified emotional problems and one participant could not identify anything as a major problem.

Seven of the ten participants felt that the biggest problem their son faced everyday was loss of independence. Most parents commented on their son's frustration at not being able to move around as they pleased or having to always ask others to do simple things for them. P7 felt that her son's biggest problem was other people's attitudes towards him:

"One of his biggest problems is attitudes towards him. I've never been shy to take him out. I've always insisted that we keep things as normal as possible, but he is the one saying, I really don't want to go to that shop anymore because everybody looks at me. It is quite noticeable that sometimes he is the only child in the shopping mall in a wheelchair and then I wonder where they all are, I know he's not the only one. Parents just don't take their children out, so they are making the ones who do go out so obvious, and then the public are not used to seeing children in wheelchairs, because he is in a wheelchair people are waiting for him to do something peculiar and he hates it."

Three of the participants mentioned that they often thought of other people who have worse problems than they do and felt very grateful. This made it easier for them to cope with their daily lives.

4.7.2 Discussion of the experiences of parents of sons with DMD.

Other studies report similar findings; Firth et al (1983) identified three major categories of problems: service (23%); practical (62%) and emotional problems (15%). The four most commonly mentioned problems were lifting; housing; inadequate information and the boys' depression or other emotional problems (Firth et al 1983). In this study the

problems the participants experienced were very varied. The practical problems experienced by the participants are categorized in Table 4.9.

Table 4.9: Practical problems experienced by parents. (n =10)

	No problem	Mild problem	Moderate problem	Severe problem	Complete problem
Lifting	2	1	2	2	3
Housing	5		2	2	1
Difficulties using public transport	2	1		2	5
Bathing	4	3	2		1
Lack of sleep due to turning son at night	1	3	2	2	2
Toileting	3	3		3	1
Dressing	5	2	2	1	
Financial	2	1	1	5	1
Feeding	6	1	3		
Social activities	2	2	4	2	

Contrary to the findings in this study, when Buchanan et al (1979) asked parents to identify a major problem they were experiencing 76% of participants identified some psychological problem, while only 16% of participants identified practical issues.

Only one participant P7 admitted that she had experienced guilt at having passed on the DMD. This is often found in the literature (Gagliardi 1991) and a possible reason that only one participant mentioned this aspect may be the lack of understanding of the genetic aspect of DMD. P7:

"I think there is also a lot of guilt attached to it which seems absurd, but from my part, I have to deal the with the fact that unknowingly I passed this on to him, and although I don't accept the guilt, I did not do it knowingly, but I think unconsciously, I do what I do for him because I did pass it on to him and I'm not willing to give him to someone else to care for because it is my thing."

Most parents had difficulty discussing DMD with their sons and said that they had not explained everything but generally answered any questions their son may have had as honestly as possible. Open communication among family members is, in general, associated with more effective coping, including greater emotional support, the sharing of relevant information, and effective informed planning (Weil 2000). Buchanan et al (1979) found that most parents had explained DMD to their sons simply as weak muscles which is similar to the participants in this study of whom six had told their sons that they had weak muscles. Fitzpatrick and Barry (1986) reported that parents in their study had great difficulty communicating within the family regarding DMD. They speculated that lack of discussion was an adaptive device for the functioning of the parents as they were inclined to focus on practical problems such as lifting and housing and tended not to think ahead (Fitzpatrick and Barry 1986). Therefore it may be more appropriate to facilitate communication between the affected boy and person outside the family such as a teacher or a counsellor. Communication about DMD is especially important when parents are aware that their sons are being misinformed or are given incorrect facts. P2 was very alarmed by the fact that her son had been told by a fellow pupil at school that he was going to die before he was 16 years of age. She and her husband found this issue very difficult to address.

Communication within families was found to be difficult by participants but two participants also mentioned that they found it very difficult to communicate with others about DMD (see Table 4.7 emotional problems stated by P2 and P7). Many factors may limit or block communication. The defences of denial and suppression as well as more conscious coping mechanisms such as the desire to avoid stigmatization, pity, “undeserved” sympathy, or a sense of guilt and shame may all be involved (Weil 2000).

Most of the participants reported that the relationship between the affected boy and his siblings was normal. Only P7 reported that there was some tension among the siblings, but also stated that they were very protective towards him. There is very little evidence of the effects on siblings of serious childhood disease and disability (Firth et al 1983).

The findings of Buchanan et al (1979) emphasize the need to attend to the well siblings' reactions to the illness. Parents were often so engrossed in the affected boys' difficulties that the subtle emotional concerns of their other children were missed. P6 voiced concern regarding the effect the DMD was having on her younger daughter.

"I think sometimes, it's not always the sufferer that suffers, like my daughter, sometimes I worry more about her than me, I'm inclined to treat both my children the same and (husband) is sometimes more hard on her (daughter)."

A feeling of guilt towards other children and the inevitability that they would be neglected at times was commented on by the participants of the study by Firth et al (1983). Some parents may feel that they have been unduly severe towards their unaffected children. In most families the siblings of the affected boy shared with the parents at least some knowledge of the disorder and their brother's prognosis. It was found that parents often gave more information to the unaffected children than to the affected boy (Firth et al 1983). This was not the case in this study as most parents had given only basic explanations to the unaffected children, except in the case of P2 and P7 who had older children who were given more information.

The biggest practical problem that the participants experienced was using public transport. This has been discussed previously in section 4.4 (p38). Lifting and financial problems were the next biggest problems. Lifting is a problem that progresses as the affected boy's weight increases. Many of the participants had injured themselves by trying to lift the boys on their own, or having to carry them up stairs. Most participants reported that they had financial problems. This suggests that parents who are already under severe emotional stress are placed under additional stress because of the lack of suitable aids which could overcome many of their difficulties (Firth et al 1983). Only the two participants who were of a higher socioeconomic level found that this was not a problem.

Two participants found social activities to be a severe problem. When asked about social activities P5 responded as follows:

“He doesn't have many friends, because children like to make fun of children like him and the children here in the road laugh at him and tease him.”

P7:

“Socially we don't have many friends, I know that his condition holds people back from inviting us to their homes because they feel perhaps the wheelchair is not going to cope and they don't want to exclude him from the invitation so they don't invite us. If they do then it is a big fuss which he hates. People are quite willing to invite my husband out, maybe just him, or there will be situations where the other kids will go out with him and (son) and I are left at home. Which obviously causes distance and separation.”

Many factors contribute to the potential for social isolation among families in which there is a child with a disability. Demands on time and resources may constrain the social activities of the affected person and other family members. Parents of affected children may experience shame and a sense of stigmatization in social situations. If the appearance or behaviour of the child is affected parents may have to confront inappropriate, hurtful responses from strangers in the course of routine public activities (Weil 2000).

This study like others (Firth et al 1983) depended mainly on the views of mothers of boys with DMD, despite the fact that fathers were invited to participate. In the interviews with there was some indication that fathers had difficulty accepting the diagnosis of DMD, but there is little research available in this area and may be a possibility for future research. It has been found that women usually take the primary role in exchanging information about a genetic condition (Marteau and Richards 1996). This was also the experience in this study.

The conclusion is presented in the next chapter.

CHAPTER 5

CONCLUSION

University of Cape Town

5.1 CONCLUSION

The aims of this study were to determine the level of understanding of genetics of parents who have sons with DMD; to determine their level of satisfaction with the genetic counselling service at RCWMCH; and to investigate the impact that DMD has had on the family.

A qualitative research approach was the most appropriate in this setting where most of the participants did not have a high level of education. By conducting personal interviews as opposed to a survey, more accurate information was able to be elicited as many participants had difficulty responding to certain questions. The level of understanding of the participants could thus be gauged by the researcher.

The findings show that while most participants demonstrated a high level of satisfaction with the service received at RCWMCH neuromuscular clinic, there are some areas of the service which could be enhanced. The participants from the lower socioeconomic group had many problems with the accessibility of the clinic particularly regarding public transport. Participants from the higher socioeconomic group did not express these concerns. The time of diagnosis was clearly remembered by the majority of the participants and, although the information that was being given was distressing, most found that it had been done in a satisfactory manner.

Education and information are an important form of support. A common complaint from the participants was that they were inadequately educated about DMD. However, the time of diagnosis is a period of high stress which makes it difficult to perceive information correctly. It may not be appropriate to give parents full detailed information about all aspects of the disorder at the initial diagnostic session and parents will have many questions that they will not have been able to assimilate at this time.

The participants generally had an inadequate level of understanding of the genetics of DMD. There were various misconceptions regarding the genetic nature of DMD, and the level of understanding was related to socioeconomic status as well as level of education.

Most of the problems experienced by the participants were of a practical nature. Those who were dependant on public transport found it very difficult. Financial problems and difficulties with lifting the affected boy were among the most frequently mentioned by the participants.

The health care professionals involved in the neuromuscular clinic have a daunting task. They are responsible for the diagnosis and management of families in which DMD has been identified. In the past they have also been tasked with having to transmit complex information to people who know little about genetics. However, the service that is delivered at the RCWMCH may be improved by having a genetic counsellor involved with all families in which a diagnosis of DMD is made, to facilitate the information giving process and to assist in providing the necessary psychosocial support.

5.2 RECOMMENDATIONS

- Information giving should not stop at the initial session when the diagnosis is given. It should be followed up on a regular basis, at follow-up or routine clinic appointments possibly by involving a genetic counsellor with all families in which a diagnosis of DMD is made.
- Healthcare professionals need to be aware of the practical barriers confronting these families and refer them to appropriate services which may lessen the burden of the parents, for example the Muscular Dystrophy Foundation of SA.
- Speedy follow-up appointments to allow these issues to be addressed should therefore be routine as recommended by Green and Murton (1996). Parents may benefit from being directed to appropriate sources of information although they

should be carefully guided as information overload may be as frightening as no information (Buchanan et al 1979). Witte (1985) recommended ongoing therapeutic contact for the entire family, possibly at four-month intervals, where family functioning can be assessed and communication of feelings about DMD and its process can be facilitated.

- Disability creates different burdens for each family especially in developing countries like SA. Maslow's (1970) complete hierarchy of needs should be considered when examining family strengths and weaknesses. A family can not be expected to follow up on recommendations for therapeutic treatment for their child or to attend clinic regularly if, for example, they are not able to provide food or heat for their family. The health care professionals involved with these families should be familiar with local resources or services that are available to assist families with basic needs of food, clothing, shelter and financial assistance (Ziolko 1991).

5.3 FUTURE RESEARCH

It will be of great value to:

- Explore how boys affected by DMD experience their disability;
- Compare families from high socioeconomic groups and families from lower socioeconomic groups' ability to adapt to DMD. This was touched on in this research but needs to be expanded to add more value;
- Include participants from all over SA to explore the experiences of a larger population, from a larger geographical area, including urban and rural areas;
- Compare the implications that various life-threatening diseases have on families for example, the impact of haemophilia or CF and DMD;
- Compare the experiences of families living in rural areas to that of families living in urban areas. This study only investigated families living in an urban area; and

- Interview families who do not regularly attend RCWMCH neuromuscular clinic to investigate the reasons for their lack of attendance.

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REFERENCES

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REFERENCES

- Abi Daoud MS, Dooley JM, Gordon KE (2001) Depression in parents of children with Duchenne muscular dystrophy. *Pediatr Neurol*. Vol. 31(1):16-19.
- Abramovsky I, Godmilow L, Hirschhorn K, Smith H Jr. (1980) Analysis of a follow-up study of genetic counseling. *Clin Genet*. Vol. 17(1):1-12.
- Bernhardt B, Biesecker BB, Mastromarino CL (2000) Goals, Benefits, and Outcomes of Genetic Counseling: Client and Genetic Counselor Assessment. *Am J Med Genet*. Vol. 94:189-197.
- Biesecker BB and Peters KF (2001) Process Studies in Genetic Counselling: Peering into the Black Box. *Am J Med Genet*. Vol. 106(3):191-8.
- Bregman A (1980) Living with progressive childhood illness: parental management of neuromuscular disease. *Soc Work Health Care*. Vol. 5:387-408.
- Bridgeman SJ and Prior TW (2005) Experience and Strategy for the Molecular Testing of Duchenne Muscular Dystrophy. *J Mol Diagn*. Vol. 7(3):317-325.
- Buchanan DC, LaBarbera CJ, Roelofs R, Olson W (1979) Reactions of families to children with Duchenne muscular dystrophy. *Gen Hosp Psychiatry*. Vol. 1(3):262-9.
- Chapple A, Campion P, May C (1996) Clinical terminology: anxiety and confusion amongst families undergoing genetic counselling. *Patient Educ Couns*. Vol. 32:81-91.
- Clarke A, Parsons E, Williams A (1996) Outcomes and process in genetic counselling. *Clin Genet*. Vol. 50(6):462-9.
- Davey A, Rostant K, Harrop K, Goldblatt J, O'Leary P (2005) Evaluating Genetic Counselling: Client expectations, Psychological Adjustment and Satisfaction with Service. *J Genet Couns*. Vol. 14(3):197-206.
- Davies R, Davis B, Sibert J (2002) Parents' stories of sensitive and insensitive care by paediatricians in the time leading up to and including diagnostic disclosure of a life-limiting condition in their child. *Child Care Health Dev*. Vol. 29(1):77-82.
- De Pina-Neto JM and Petean EBL (1999) Genetic counselling follow-up – A retrospective study with a quantitative approach. *Genet Mol Biol*. Vol. 22(3):295-307.

- Eggers S, Pavanello RCM, Passos-Beuno MR, Zatz M (1999) Genetic counselling for childless women at risk for Duchenne Muscular Dystrophy. *Am J Med Genet.* Vol. 86:447-453.
- Emery AE (1991) Population frequencies of inherited neuromuscular diseases- a world survey. *Neuromuscul Disord.* Vol. 1(1):19-29.
- Emery AEH (1998) The muscular dystrophies: Clinical review. *BMJ* Vol. 317:991-995.
- Falek A (1984) Sequential aspects of coping and other issues in decision making in genetic counselling. In: Emery A, Pullen I. (Eds) *Psychological aspects of genetic counselling.* Academic Press, London.
- Falvo DR (1991) *Medical and Psychosocial aspects of chronic illness and disability.* Aspen Publishers Inc., Maryland.
- Firth M, Gardner-Medwin D, Hosking G, Wilkinson E (1983) Interviews with parents of boys suffering from Duchenne muscular dystrophy. *Dev Med Child Neurol.* Vol. 25(4):466-71.
- Fitzpatrick C and Barry C (1986) Communication within families about Duchenne muscular dystrophy. *Dev Med Child Neurol.* Vol. 28(5):596-9.
- Fitzpatrick C and Barry C (1990) Cultural differences in family communication about Duchenne muscular dystrophy. *Dev Med Child Neurol.* Vol. 32(11):967-73.
- Fraser FC (1974) Genetic Counselling. *Am J Human Genet.* Vol. 26: 636-661
- Gagliardi BA (1991) The impact of Duchenne Muscular Dystrophy on Families. *Orthop Nurs* Vol. 10 (5): 41-49.
- Giacomini MK and Cook DJ (2000) User's Guides to the Medical Literature. XXIII. Qualitative Research in Health Care. A. Are the Results of the Study Valid? *JAMA.* Vol. 284: 357-362.
- Goliath R (2006) Personal Communication: 8 May 2006. NHLS Head of diagnostic laboratory, Division of Human Genetics, University of Cape Town.
- Green JM and Murton FE (1995) Diagnosis of Duchenne muscular dystrophy: parents' experiences and satisfaction. *Child Care Health Dev.* Vol. 22(2): 113-128.
- Grinnell RM (1988) *Social research and evaluation.* Peacock Publishers, Illinois.

- Guba E and Lincoln Y (1989) *Fourth generation evaluation*. Sage, Newbury Park.
- Harper PS (2004) *Practical Genetic Counselling*. 6th Edition. Oxford University Press, New York.
- Hoffman EP, Fischbeck KH, Brown RH, Johnson M, Medori R, Loike JD, Harris JB et al. (1988) Characterization of dystrophin in muscle-biopsy specimens from patients with Duchenne's or Becker's muscular dystrophy. *N Engl J Med*. Vol. 318(21):1363-8.
- Holloway I (1997) *Basic Concepts for Qualitative Research*. Blackwell Science, Oxford.
- Holloway I and Wheeler S (1996) *Qualitative Research for Nurses*. Blackwell Science, Oxford.
- Holroyd J and Guthrie D (1986) Family stress with chronic childhood illness: cystic fibrosis, neuromuscular disease, and renal disease. *J Clin Psychol*. Vol. 42(4):552-61.
- Kübler-Ross E (1969) *On Death and dying*. Macmillan, New York.
- Lincoln YS and Guba EG (1985) *Naturalistic enquiry*. Sage, Beverly Hills.
- Livneh H, Antonak RF (1997) *Psychosocial adaptation to chronic illness and disability*. Aspen Publishers Inc., Maryland.
- Marteau T and Richards M (1996) *The troubled helix: Social and psychosocial implications of the new human genetics*. Cambridge University Press, Cambridge.
- Maslow AH (1970) *Motivation and personality*. 2nd edition. Harper and Row, New York.
- McDowell I and Newell C (1996) *Measuring health: A guide to rating scales and questionnaires*. 2nd Edition. Oxford University Press, New York.
- McMillan JH and Schumacher S (2001) *Research in Education: a conceptual introduction*. 5th Ed. Longman: New York.
- Mitchie S, Marteau T, Bobrow M (1997a) Genetic counselling: The psychological impact of meeting patients' expectations. *Clin Genet*. Vol. 34:237-241.
- Mitchie S, McDonald V, Marteau T (1997b) Genetic counselling: information given, recall and satisfaction. *Patient Educ Couns*. Vol. 32:101-106.

- Mueller RF and Young ID (2001) *Emery's elements of medical genetics*. Churchill Livingstone, London.
- Nereo NE, Fee RJ, Hinton VJ (2003) Parental stress in mothers of boys with Duchenne muscular dystrophy. *J Pediatr Psychol*. Vol. 28(7):473-484.
- Neuman WL (1999) *Social Research Methods: Qualitative and Quantitative Approaches*, 4th Edition. Allyn and Bacon, Massachusetts.
- Nigro G, Comi LI, Politano L, Bain RJ (1990) The incidence and evolution of cardiomyopathy in Duchenne muscular Dystrophy. *Int J Cardiol*. Vol. 26(3):271-7.
- Pope C, Ziebland S, Mays N (2000) Qualitative research in health care: Analysing qualitative data. *BMJ* Vol. 320:114-116.
- Reid DT and Renwick RM (2001) Relating familial stress to the psychosocial adjustment of adolescents with Duchenne muscular dystrophy. *Int J Rehabil Res*. Vol. 24:83-93.
- Ross E, Deverell A (2004) *Psychosocial approaches to health, illness and disability*. Van Schaik Publishers, Pretoria.
- Sacket DL (1979) Bias in analytical research. *J Chron Dis*. Vol. 32:51-63.
- Shiloh S, Avdor O, Goodman RM (1990) Satisfaction with genetic counseling: Dimensions and measurement. *Am J Med Genet*. Vol. 37:522-529.
- Smith J, Harrer R, Van Langenhowe L (1995) *Rethinking methods in psychology*. Butterworth-Heinemann, Oxford.
- Somer M, Mustonen H, Norio R (1988) Evaluation of genetic counselling: recall of information, post-counselling reproduction, and attitude of the counselees. *Clin Genet*. Vol. 34:352-265.
- Statistics South Africa: Earning and Spending in South Africa. Selected findings and comparisons from the income and expenditure surveys of October 1995 and October 2000. Retrieved on 20 April from <http://www.statssa.gov.za/>
- Sussman M (2002) Duchenne muscular dystrophy. *J Am Acad Orthop Surg*. Vol. 10(2):138-51.
- Thompson RJ, Zenman JL, Fanurik D, Sirotkin-Roses M (1992) The role of parent stress and coping and family functioning in parent and child adjustment to Duchenne muscular Dystrophy. *J Clin Psych*. Vol. 48(1):11-19.

- Webb CL (2005) Parents' perspectives on coping with Duchenne muscular dystrophy. *Child Care Health Dev.* Vol. 31(4):385-396.
- Weil J (2000) *Psychosocial genetic counselling*. Oxford University Press, New York.
- Williams B, Coyle J, Healy D (1998) The meaning of patient satisfaction: An explanation of high reported levels. *Soc Sci Med.* Vol. 47(9):1351-1359.
- Witte RA (1985) The psychosocial impact of a progressive physical handicap and terminal illness (Duchenne muscular dystrophy) on adolescents and their families. *Br J Med Psychol.* Vol. 58(Pt 2):179-87.
- Ziolko ME (1991) Counseling parents of children with disabilities: a review of the literature and implications for practice. *Journal of Rehabilitation.* Vol. 57:29-34.

University of Cape Town

M.Sc in Genetic Counselling Research Project

An investigation into the level of genetic knowledge of parents of sons with Duchenne Muscular Dystrophy and their satisfaction with the genetic counselling service at Red Cross War Memorial Children's Hospital.

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 because I have a son who is affected by Duchenne Muscular Dystrophy (DMD) and attend the neuromuscular clinic at Red Cross War Memorial Children's Hospital (RCWMCH).

2.1 I understand that the objective of this study is to investigate:

- the level of knowledge of the inheritance of DMD and knowledge of the carrier status of female family members;
- the impact that DMD has had on our family; and
- the level of satisfaction we experience with the genetic counselling service at RCWMCH.

2.2 I understand that the interview will take place 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 investigation that will take place in 2006 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 will refer me to a 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 will not be included.

3.2 I understand that the interview will 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 tape and the data stored on the computer will have a numerical code only and my name does not appear anywhere.

4. I have been assured that the recorded and transcribed information discussed at the meeting will only be made available to the researcher's supervisors with my study code number and that they do not know that it refers to my name.

5. I have not been coerced 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 current or future access to medical and genetic services to which I am entitled.

6. 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 will be no medical benefits to me from this study.

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 VOLUNTARILY AGREE TO PARTICIPATE IN THE ABOVE RESEARCH STUDY

Signed at:
(address) on
2006

.....
Participant's signature

.....
Witness

**I HEREBY DECLARE THAT I AGREE TO HAVE MY INTERVIEW
AUDIOTAPE RECORDED**

Signed at:
(address) on
2006

.....
Participant's signature Witness

IMPORTANT INFORMATION

Dear Participant,

Thank you for your participation 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:

Kelly Loggenberg: (021) 406-6373

Email: kloggenb@cormack.uct.ac.za

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 VERSION OF THE INFORMATION AND CONSENT FORM

M.Sc in Genetiese Berading Navorsingsprojek

'n Onderzoek na die kennis van genetika van ouers van seuns met Duchenne Spier Distrofie en hulle tevredenheid met met die genetiese raadgewing diens by die Rooi Kruis Oorloggedenk Kinderhospitaal.

INLIGING EN TOESTEMMING VORM

VERKLARING DEUR DEELNEMER

Ek,

(adres)

bevestig dat:

1. Ek is uitgenooi om aan die bogenoemde navorsings projek wat deur die Divisie van Mensgenetika, Universiteit van Kaapstad geïnisieer is, deel te neem aangesien ek 'n seun het met Duchenne Spier Distrofie en die neuromuskulêre kliniek by Rooi Kruis Oorloggedenk Kinderhospitaal bywoon.
 - 2.1. Ek verstaan dat die doel van hierdie projek is om die volgende te ondersoek:
 - Kennis van die oorerflikheid van Duchenne Spier Distrofie en kennis van die draer status van vroulike familieledes;
 - Die impak wat Duchenne Spier distrofie op ons familie gehad het; en
 - Die vlak van tevredenheid wat ons ondervind met die genetiese raadgewing diens by die Rooi Kruis Oorloggedenk Kinderhospitaal.
 - 2.2. Ek verstaan dat die onderhoud of by my huis of by 'n ander plek van my keuse sal plaasvind en dat dit een of twee besoeke van twee ure elk behels.
 - 2.3. Ek is bewus dat dit 'n eenmalige ondersoek is wat in 2006 sal plaasvind op 'n tyd wat vir my en my gesin gerieflik is.

- 2.4. Ek verstaan dat van die vra my hartseer of ongelukkig mag maak, maar dat die risiko's van die studie minimaal is. Die navorser sal my na 'n genetiese raadgever verwys indien nodig. Sy sal my met respek, aanvaarding en empatie behandel gedurende die onderhoud.
- 3.1. Ek is verseker dat alle inligting vertroulik behandel sal word. Inligting mag vir 'n tesis, publikasies in wetenskaplike tydskrifte en aanbiedings by professionele kongresse gebruik word, maar name sal nie ingesluit word nie.
- 3.2. Ek verstaan dat die onderhoud op band opgeneem sal word sodat die navorser nie te veel hoef te skryf gedurende die onderhoud nie. Die band sal in 'n kluis gestoor word tot dat die navorsing opgeskryf is en sal daarna dadelik vernietig word. Die band en die data op die rekenaar sal slegs 'n numeriese kode op hê en my naam sal nie daarop verskyn nie.
4. Ek is verseker dat die inligting wat opgeneem en getranskribeer is slegs aan die navorser se mentor bekend gemaak sal word, maar dit sal slegs my numeriese studie kode bevat en my naam sal nie daarop verskyn nie.
5. Ek is nie oorreed om aan die die projek deel te neem nie en ek is bewus dat ek mag weier om deel te neem, en ek kan op enige stadium besluit om te onttrek. My onttrekking sal op geen manier my huidige of toekomstige toegang tot die mediese of genetiese dienste, waarop ek geregtig, is beïnvloed nie.
6. het die inligting van die projek in Engels/Afrikaans aan my verduidelik. Ek is vlot is hierdie taal en my vra is ten volle beantwoord.
7. Ek verstaan dat daar geen mediese voordele vir my sal wees as gevolg van hierdie projek nie.

8. Ek is verseker dat my deelname aan hierdie projek nie tot enige addisionele koste vir my familie sal lei nie en dat ek nie finansieel gaan baat daarby nie.

EK VERKLAAR HIERMEE DAT EK VRYWILLIG AAN DIE BOGENOEMDE NAVORSINGS PROJEC DEELNEEM

Geteken te:

(Adres) op2006

.....
Deelnemer se handtekening

.....
Getuie

EK VERKLAAR HIERMEE DAT EK MY ONDERHOUD OP BAND OPGENEEM MAG WORD

Geteken te:

(Adres) op2006

.....
Deelnemer se handtekening

.....
Getuie

BELANRIKE INLIGTING

Geagte deelnemer,

Baie dankie vir u deelname aan hierdie studie. As U gedurende die verloop van die navorsing enige vrae het aangaande:

1. probleme as gevolg van die navorsing, of
2. vrae aangaande inligting oor die projek

kontak my of Prof. Greenberg gerus op die volgende telefoon nommers:

Kelly Loggenberg (021) 406-6373
Email: kloggenb@cormack.uct.ac.za

Prof. Jacquie Greenberg: (021) 406-6299

As u enige vrae het in verband met u reg as 'n deelnemer, kontak Prof. T Zabow, die Voorsitter van die Etiese Hersiening Komitee van die Universiteit van Kaapstad by (021) 406-6492.

INTERVIEW SCHEDULE

Participant No:.....

A. Sociodemographic Information

1. Family history
 - Date of birth of child with DMD
 - Marital status
 - How many children
 - Age of children
2. How many people live in this house and Wendy houses?
3. What is your relationship to them?
4. How old are they?
5. Are they employed?
6. How many rooms are there in your home?
 - Bedrooms
 - Bathrooms
 - Toilet (inside or outside)
 - Kitchen
 - Other
7. Is your home accessible by wheelchair?
 - Stairs
 - Doorways
 - Passages
 - Size of rooms
 - Outside environment from front door to road
8. Which grade/standard did you complete at school?
(To be answered by father and mother)
 - Grade 12 (matric, Std 10)
 - Grade 11
 - Grade 10
 - Grade 9

- Grade 8
- Grade 7
- Other

9. Have you started any further courses/training since leaving school?

- Yes
- No

10. If Yes to question 9, what?

- Trade/apprentice
- Certificate from college
- Diploma (beyond Grade 12)
- Bachelors degree
- Postgraduate diploma/degree
- Other

11. Have you completed it?

- If not, give reasons

12. Do you work?

- Yes
- No

13. If Yes to question 12, what kind of work?

14. Is the work;

- Self-employed
- Full-time employed
- Part-time employed

15. If No to question 12, are you:

- Unemployed
- Permanently unable to work due to caring for your son
- Retired or pensioner
- Other

16. How many people contribute to the family income?

17. How many people does the family income support?

18. What is your current household income per month?

- No income
- Disability grant R780

Salary income:

- R1 – R400
- R401 – R800
- R801 – R1 600
- R1 601 – R3 200
- R3 201 – R6 400
- R6 401 – R12 800
- R12 801 – R25 600
- R25 601 – R51 200
- R52 201 – R102 400
- R102 401 – R204 800
- More than R204 800

B. Red Cross War Memorial Children's Hospital (RCWMCH) neuromuscular clinic.

19. How were you referred to RCWMCH neuromuscular clinic?

20. To which other clinics at RCWMCH have you been referred?

21. How often do you and your son attend the neuromuscular clinic at RCWMCH?

22. How far is it from your home?

23. How do you get to the clinic?

24. How long does it take to get to the clinic?

25. How much does it cost to get there?

26. Do you have medical aid?

- Yes
- No

27. Does your medical aid cover your son's medical needs completely?

- Consultations
- Wheelchairs

- Medication

28. Has it ever happened that your medical aid has become depleted? (what then)

C. Satisfaction with counselling service.

29. When did you first suspect a problem with your son?
- Can you describe the early symptoms that made you wonder if your son had a problem?
30. How long did you have to wait for your first visit to RCWMCH?
31. By who was the diagnosis made?
32. What was your reaction to the diagnosis?
- Describe what that time was like for your family.
33. Did you receive counselling regarding the inheritance of DMD and it's prognosis? (When)
34. Has DMD been confirmed with a DNA test?
35. How long did you have to wait for a result?
36. How did you feel in this initial phase?
37. In the following questions rate your level of satisfaction with the counselling service at RCWMCH (Adapted from Shiloh et al1990).

	Very unsatisfactory	Unsatisfactory	Satisfactory	Very satisfactory	Highly satisfactory
The way in which DMD was explained to you.					
The counsellor listened to what I had to say.					
The counsellor understood what was worrying me.					
The counsellor showed a lot of caring.					
The counsellor answered all my questions.					
Counselling helped cope with your problems.					

There was enough time given at each session.					
The clinic environment was informal.					
There were no disruptions during the counselling.					
The way the test results were presented to you.					
Time in the follow up visits for questions to be answered.					
Time waited to get first appointment.					
Time waited to see staff at each visit.					
Rate satisfaction with muscle clinic in general.					
Follow up visits gave a lot of support.					

Other: _____

D. Level of understanding of genetics (adapted from Somer et al. 1988)

38. What do you understand about the cause of DMD?
39. Is DMD caused by genes
- entirely,
 - partly, or
 - not at all?
40. What do you understand about the inheritance of DMD?
41. What is the chance of having another child with DMD?
42. Does DMD affect both males and females?
43. Who in a family does the affected boy usually inherit the abnormal gene(s) from:
- father,
 - mother,
 - both, or
 - neither?
44. Is it possible to detect DMD before a baby is born?
45. Is it possible to detect DMD by amniocentesis?
46. What are your feelings with regard to TOP if the foetus is positive?

47. What do you know about girls being carriers of the gene that causes DMD?

E. Experiences of parents of boys with DMD.

48. In general, what do you experience as a major problem in your daily lives?

49. In general, what are the practical problems that you experience in your daily lives?

50. In caring for your son; to what extent do you experience the following to be a problem?

	No problem	Mild problem	Moderate problem	Severe problem	Complete problem
Lifting					
Housing					
Difficulties using public transport					
Bathing					
Lack of sleep due to turning son at night					
Incontinence					
Toileting					
Dressing					
Financial					
Feeding					
Social activities					

Other: _____

51. What are the emotional problems that you experience?

- Unpredictability of the disease
- Society's attitude to handicapped people
- Watching your son deteriorate
- Parental isolation
- Other

52. Has DMD had an effect on your marital relationship?

53. In what way have your other children been affected by having a brother with DMD?

54. How have your other children reacted towards their brother?

55. What have you told your son about DMD?
56. What have you told your other children about DMD?
57. In your opinion; what are the difficulties that your son experiences in his daily life?
58. Who/ what provides you with the most support?
59. What has made your experience easier?

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AFRIKAANS VERSION OF THE INTERVIEW SCHEDULE**ONDERHOUD SKEDULE**

Deelnemer nr:

A. Sosiodemografiese Inligting

1. Familie geskiedenis

- Geboortedatum van kind met DSD
- Huwelik status
- Hoeveel kinders
- Ouderdom van kinders

2. Hoeveel mense bly by hierdie adres?

3. Wat is u verhouding met hulle?

4. Hoe oud is hulle?

5. Het hulle werk?

6. Hoeveel kamers is daar in u huis?

- Slaapkamers
- Badkamers
- Toilette (binne of buite)
- Kombuise
- Ander

7. Is u huis toeganklik met 'n rolstoel?

- Trappe
- Deure
- Gange
- Buite omgewing van die voordeur tot die pad

8. Watter standerd het u voltooi op skool?

- Graad 12 (Matriek, Std 10)
- Graad 11
- Graad 10
- Graad 9

- Graad 8
- Graad 7
- Ander

9. Het jy enige verdere kursusse/opleiding gedoen na u skool verlaat het?

- Ja
- Nee

10. Indien JA vir vraag 9, wat?

- Ambag
- Sertifikaat van kollege
- Diploma (na Graad 12)
- Baccalureurs graad
- Nagraadse opleiding of diploma
- Ander

11. Het u dit voltooi?

- Indien nie, gee redes

12. Werk u?

- Ja
- Nee

13. Indien Ja vir vraag 12, watter tipe werk?

14. Is dit:

- U eie besigheid
- Voltydse werk
- Deeltydse werk

15. Indien Nee vir vraag 12, is u:

- Werkloos
- Kan permanent nie werk nie omdat u seun oppas
- Afgetree of pensionaris
- Ander

16. Hoeveel mense dra by tot die gesamentelike inkomste?

17. Hoeveel mense word deur hierdie inkomste onderhou?

18. Wat is u huidige gesamentelike huishoudelike inkomste per maand?

- Geen inkomste

- Ongeskikheids toelaag R780

Salaris inkomste:

- R1 – R400
- R401 – R800
- R801 – R1 600
- R1 601 – R3 200
- R3 201 – R6 400
- R6 401 – R12 800
- R12 801 – R25 600
- R25 601 – R51 200
- R52 201 – R102 400
- R102 401 – R204 800
- Meer as R204 800

B. Rooi Kruis Oorlog Gedenk Kinder Hospitaal neuromuskulere kliniek.

19. Hoe was u verwys na RCWMCH neuromuskulere kliniek?
20. Na watter ander klinieke by RCWMCH was julle verwys?
21. Hoe gereeld woon u en u kind die neuromuskulere kliniek by Rooikruis oorloggedenk kinder hospitaal by?
22. Hoe ver is dit van u huis af?
23. Hoe kom julle by die kliniek?
24. Hoe lank neem dit om by die kliniek te kom?
25. Hoeveel kos dit om daar te kom?
26. Het u mediesefonds?
 - Ja
 - Nee
27. Dek u mediesefonds u seun se mediese onkoste volledig?
 - Konsultasies
 - Rolstoele
 - Medikasie
28. Het dit al ooit gebeur dat u mediesefonds uitgeput word? (wat dan)

C. Tevredenheid met die beradingsdiens.

29. Wanneer het u vir die eerste keer vermoed daar is n problem met u seun?
- Kan jy die vroeë simptome wat jou lat wonder het of daar 'n probleem was, beskryf?
30. Hoe lank moes u wag vir u eerste besoek aan RCWMCH?
31. Deur wie was die diagnose gemaak?
32. Hoe het u gereageer op die diagnose?
- Beskryf hoe die familia daardie tyd eraar het.
33. Het u berading aangaande die oorerwing van DSD en die verloop daarvan ontvang? (wanneer)
34. Is DSD met 'n DNS toets bevestig?
35. Hoe lank moes u wag vir 'n resultaat?
36. Hoe het u gevoel gedurende hierdie begin fase?
37. Dui in die volgende vrae u vlak van tevredenheid met die huidige raadgewingsdiens by die RCWMCH aan. (Aangepas van Shiloh et al 1990).

	Baie ontevrede	Ontevrede	Tevrede	Baie tevrede	Hoogs tevrede
Die manier waarop DMD aan u verduidelik was.					
Die raadgewer het geluister na wat ek te se gehad het.					
Die raadgewer het verstaan wat my pla.					
Die raadgewer het baie meegevoel getoon.					
Die raadgewer het al my vrae beantwoord.					
Raadgewing het my gehelp om met my probleme te 'cope'.					
Daar was genoeg tyd by elke sessie.					
Die kliniek omgewing was informeel.					
Daar was geen onderbrekings gedurende die raadgewing.					
Die manier waarop die resultate aan u gegee is.					
Tyd in die opvolg besoeke om u vrae te beantwoord.					
Tyd gewag vir eerste afspraak.					

Tyd gewag om personeel te sien tydens elke besoek.					
Dui tevredenheid aan met die neuromuskulere kliniek in die algemeen.					
Opvolg besoeke het heelwat ondersteuning gebied.					

Ander: _____

D. Vlak van verstaan van genetika (aangepas van Somer *et.al.* 1988).

38. Wat verstaan u is die oorsaak van DSD?
39. Is DSD veroorsaak deur gene
- Heeltemaal
 - Gedeeltelik, of
 - Geensins?
40. Wat verstaan u van die oorerwing van DSD?
41. Wat is die kans om nog n kind met DSD te hê?
42. Affekteer DSD beide mans en vrouens?
43. Wie in 'n familie dra gewoonlik die abnormale geen oor:
- Vader
 - Moeder
 - Albei, of
 - Nie een nie?
44. Is dit moontelik om DSD voor geboorte te diagnoseer?
45. Is dit moontelik om DSD deur middel van amniosentese te diagnoseer?
46. Hoe voel u oor terminasie van swangerskap indien die fetus positief toets?
47. Wat weet u van meisies wat draers is van die geen wat DSD veroorsaak?

D. Ervaring van ouers van seuns met Duchenne Spier Distrofie (DSD).

48. Wat ervaar u, in die algemeen, as 'n groot probleem in julle daaglikse lewe?

49. Wat is die praktiese probleme wat u in die algemeen in u daaglikse lewe ervaar?

50. In die versorging van u seun; tot watter mate is die volgende vir u 'n probleem?

	Geen probleem	Effense probleem	Matige probleem	Ernstige probleem	Abolute probleem
Optel					
Behuising					
Probleme met publieke vervoer					
Bad					
Slaap tekort (redes)					
Inkontinent (gebrek aan blaas beheer)					
Toilet gebruik					
Aantrek					
Finansieel					
Voeding					
Sosiale aktiwiteite					

Ander: _____

51. Wat is die emosionele probleme/bevrediging wat u ervaar?

- Onvoorspelbaarheid van die siekte
- Gemeenskap se houding teenoor gestremde mense
- Om te kyk hoe jou seun agteruitgan
- Ouerlike isolasie

52. Het DSD 'n effek op jou huwelik gehad?

53. Op watter manier is jou ander kinders geraak deur n broer te hê met DSD?

54. Hoe reageer jou ander kinders teenoor hulle broer?

55. Wat het jy jou seun vertel van DSD?

56. Wat het jy jou ander kinders van DSD vertel?

57. In u opinie, wat is die moeilikhede wat u seun op 'n daaglikse basis ervaar?

58. Wie of wat gee u die meeste ondersteuning?

59. Wat maak u ervaring makliker?

University of Cape Town

UNIVERSITY OF CAPE TOWN



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20 December 2005

REC REF: 360/2005

Prof J Greenberg
Human Genetics

Dear Prof Greenberg

PROJECT TITLE: AN INVESTIGATION INTO THE LEVEL OF GENETIC KNOWLEDGE OF PARENTS OF SONS WITH DUCHENNE MUSCULAR DYSTROPHY AND THEIR SATISFACTION WITH THE GENETIC COUNSELLING SERVICE AT RED CROSS WAR MEMORIAL CHILDREN'S HOSPITAL

Thank you for your letter to the Research Ethics Committee dated 07 December 2005

It is a pleasure to inform you that the Ethics Committee has **formally approved** the above-mentioned study on the 20 December 2005

Your comments to the queries raised are noted with thanks.

Please quote the REC. REF in all your correspondence.

Yours sincerely

PROFESSOR J. ZABOY
CHAIRPERSON, HSE HUMAN ETHICS

lempdt