



The role of a genetic counsellor in a support group for Huntington disease

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Dedication

I would like to dedicate this writing to my husband, Philip and son, Caleb. You have been the greatest support I could have ever asked for and I would like to honour you for your endless love, assistance, encouragement and patience throughout my studies.

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Abstract

Genetic counselling students are encouraged to become involved with support groups for genetic conditions. However, their roles within these groups are not well defined and poorly understood and ethical concerns have been raised regarding the appropriateness of such involvement.

A limited number of international studies have examined the role of a genetic counsellor in a support group. These studies report broadly on the matter but lack the personal response that qualitative data produces. South African literature is even more limited and no studies (both locally and internationally) have attempted to describe the role of a genetic counsellor in a support group for Huntington disease.

This study examines the role of a genetic counsellor in a Huntington Disease support group, specifically the Huntington's Association of South Africa (HASA). A qualitative research approach was used to interview 17 people who have been involved with HASA in the past seven years. In-person and telephonic interviews were conducted with five genetic counsellors and one psychologist. Two separate focus groups were conducted with a total of 11 support group members and one genetic counsellor (who was also interviewed in-person).

The interviews and focus groups were audio recorded and transcribed by a combination of an online software program named Sonix Transcription and manual transcription by the researcher herself. Thematic analysis was done and the results were grouped according to the following five themes that emerged from the data: 1) Is there a role?, 2) Information provider, 3) Emotional support, 4) Practical helper, and 5) Community member.

It was found that there is a role for a genetic counsellor in a HASA support group and that the relationship that is formed through such involvement can be mutually beneficial to both the support group members and the genetic counsellor. The genetic counsellor's role was found to be wide and included specialist and practical roles, as indicated by above mentioned themes two to five.

These findings cannot be extrapolated to apply to all support groups due to the unique characteristics and function of different support groups. This was evident in this case as the two branches in SA (Western Cape and Gauteng) seem to function very differently. The research can, however, be used as guideline for involvement with other support groups. This

study's findings made a unique contribution in that it documented in detail the genetic counsellor's involvement in support groups.

The study found that all the participants agreed that HASA should have access to a genetic counsellor, thus it is recommended that future studies should explore the role of a genetic counsellor in support groups for other genetic conditions, as those groups could also find it beneficial.

List of terms and abbreviations

AD	Autosomal dominant
AOO	Age of onset
CAG	Cytosine-adenine-guanine
CT	Cape Town
CTG	Cytosine-thymine-guanine
DGE(s)	Data gathering event(s)
FG	Focus group
GP	Gauteng
HASA	Huntington's Association of South Africa
HD	Huntington Disease
HDL2	Huntington disease-like 2
HPCSA	Health Practitioners' Council of South Africa
Htt	Huntingtin protein
HTT	Huntingtin gene
IA(s)	Intermediate allele(s)
mHtt	Mutant form of huntingtin protein
NHLS	National Health Laboratory Service
PT	Predictive testing
PTP	Predictive testing process
RP	Reduced penetrance
SG(s)	Support group(s)
UCT	University of Cape Town
WITS	University of the Witwatersrand

Clarification of phrases and terms

“Clinical team”	The doctors, registrars, nurses and genetic counsellors working at the department of Human Genetics, UCT
“Counselling/Counseling”	The UCT Harvard referencing style has been used for this dissertation as it is recommended for UCT academic purposes. It should be noted that “counselling” is spelt with a double “l” in the main text as it is generally spelt this way in South Africa; yet, in references, it is spelt as “counseling” (single “l”) to abide by the correct spelling of the United States and the United Kingdom.
“Counsellor”	Used interchangeably with the term “genetic counsellor”.
“Data gathering event(s)”	Global term referring to either individual interviews (in-person or telephonic) or focus groups conducted as methods of data gathering.
“Genetic counsellor”	Includes both trainee counsellors (studying or doing their internship) and registered genetic counsellors.
“HASA members”	All HASA members (affected, unaffected, at risk, family members, friends) excluding genetic counsellors and psychologists involved. Used interchangeably with the terms “group members” and “support group members”.
“Participant category”	Refer to the three categories of participants: HASA members, genetic counsellors and psychologists.
“Participants”	All participants of the study including the HASA members, genetic counsellors and psychologist.
“The clinic”	GSH/UCT’s neurogenetic clinic that runs once a month at the department of human genetics at UCT.

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Chapter One

Literature review, Rationale, Aims & Objectives

1.1 CHAPTER INTRODUCTION

In this chapter empirical literature applicable to the study is reviewed. Literature was searched for using PubMed, Ebscohost, Sabinet African Journals and ScienceDirect research databases. The following search phrases were used: *Huntington Disease, South Africa, support group, genetic counselling, relationship with support group* as well as permutations of these terms.

The literature is presented according to the following as it relates to the study:

- Genetic counselling as a medical service
- The role of support groups
- The partnership between genetic counsellors and support groups
- Huntington disease as genetic condition
- Support group for Huntington disease

Chapter one concludes with the rationale for the study stemming from the literature review as background.

1.2 GENETIC COUNSELLING

Genetic counselling is a service that helps people understand the implications of inherited genetic conditions. It is provided by qualified health care professionals that include genetic counsellors, medical geneticists and genetic nurses.

Genetic counselling plays a unique role within the field of medicine as it provides patients with both emotional support and education regarding genetic conditions. The Genetic Counseling Definition Task Force of the National Society of Genetic Counselors (NSGC) defines the field as follows:

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

- 1) Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
- 2) Education about inheritance, testing, management, prevention, resources and research.
- 3) Counselling to promote informed choices and adaptation to the risk or condition.”

(Resta et al., 2006:77)

The abovementioned definition broadly describes the different roles that counsellors play within their scope of practise.

A study conducted in South Africa investigated the work that local genetic counsellors do (Kromberg, Wessels & Krause, 2013:753). It was reported that, as professionals, they are adaptable and play several significant roles including counselling on 57 different genetic disorders, educating patients, conducting research, marketing and fulfilling administrative functions. Other roles include being involved with SGs and certain social activities (Kromberg, Wessels & Krause, 2013:754).

A similar study conducted in Europe and the UK, lists the roles of genetic counsellors in more detail as: “family history taking, pedigree drawing, risk assessment, discussion of natural history of the condition, psycho-social impact of the diagnosis, provision of patient education, discussion of options, addressing ethical issues, making a psycho-social assessment and providing psycho-social support” (Skirton et al., 2015:452-458). Additional responsibilities such as providing professional and public education and guiding new-born screening programmes are also listed as appropriate activities for genetic counsellors to conduct.

Disclosing and discussing genetic information and its personal and familial implications requires a certain level of privacy to adequately protect and support patients. For this reason, the abovementioned roles of and services by genetic counsellors usually take place in an intimate setting with only the genetic counsellor, patient and (optional) support partner present. This private setting is the “traditional space” in which genetic counselling takes place.

In addition to receiving genetic counselling, patients are often encouraged to join lay support groups (SGs) where additional support from people with a similar condition can be received. Genetic counsellors (professional and in-training) are also encouraged to get involved with lay

support groups as an extension to their scope of practice (Kromberg, Wessels & Krause, 2013:754), to learn first-hand about the conditions that students encounter at clinic and to consider what the role of a genetic counsellor is in a parent/patient lay support group (SG).

1.3 SUPPORT GROUPS

A SG is often defined as a group of people (led by a professional or lay person) “who provide each other with moral support, information and advice relating to a shared characteristic or experience” (Plumridge et al., 2012:256). When an individual is affected by chronic illness, there are many practical and emotional difficulties that family members may have to cope with (Plumridge et al., 2012:256) and becoming part of a SG can help people on their journey.

Support groups come in various shapes and sizes. In South Africa there are well-known SGs such as CANSA (www.cansa.org.za) and PinkDrive (www.pinkdrive.co.za) offering support for people affected by breast cancer, and the South African Depression and Anxiety Group (SADAG) offering support for people with depression and anxiety. Genetic support groups can be a valuable resource for much-needed support among people who have a hereditary condition in common (Mann, 1999:549).

A genetic SG can benefit people when meeting others who share a similar situation and who can provide empathy, emotional support and first-hand experience. (Plumridge et al., 2012:256). Additional benefits include the sharing of practical information, moral support and advocacy (Mann, 1999:549) and the formation of friendships that can help with the isolation that can be felt when dealing with a genetic condition (Plumridge et al., 2012:256). In addition, it has been found that membership of a genetic SG improves knowledge of the condition (Plumridge et al., 2012:256).

Different type of SGs exist including in-person SGs, online SGs and Whatsapp SGs. The nature of the groups differs depending on the needs of the group members. A study conducted in the United Kingdom (UK) explored the role that different types of genetic SGs play in helping families cope emotionally and communicate risks to children in cases where conditions are inherited (Plumridge et al., 2012:255). Despite different groups having different foci, the main aims of genetic SGs were found to be similar, including psychosocial support network, social gathering, education and awareness raising for members, and raising public awareness and funds for research and support (Plumridge et al., 2012:258).

There are also groups that focus on the youth that is affected by HD, including active participation and contribution to an SG's website by the youth themselves (Williams et al., 2013). A study focusing on teens showed that involvement in a HD SG was seen as being beneficial, with online social networks aiding this by providing information on the condition and providing a platform for teens to express themselves (Williams et al., 2013).

The Genetic Alliance South Africa (GA-SA) (www.geneticalliance.org.za) and Rare Diseases South Africa (www.rarediseases.co.za) are non-profit organisations that help unite patient support groups, healthcare professionals and other stakeholders involved with congenital disorders' care and prevention. Their websites offer information on relevant resources, treatment, supportive care and contact details of various SGs for the public to contact and get involved with.

The GA-SA was established in 1973 as the Southern African Inherited Disorders Association (SAISA), after it became evident to founders Professor Trefor Jenkins and Professor Jennifer Kromberg that systems for support were needed for people affected by inherited disorders (GA-SA, 2019a). More than 20 patient support groups formed part of it when it started and similar organisations exist worldwide, e.g. the Genetic Alliance based in Washington DC (www.geneticalliance.org).

Unique is an international organization that provides support in various ways (including networking) to people who are impacted by chromosome or genomic disorders (GA-SA, 2019b). Some of their members are South African and there is also a South African representative.

1.4 PARTNERSHIP BETWEEN GENETIC COUNSELLORS AND SUPPORT GROUPS

A limited number of research studies have been published on the partnership between genetic counsellors and support groups, but current existing literature is discussed below.

When genetic counsellors become involved in SGs, the setting differs significantly from the typical genetic counselling environment. SG meetings mostly take place in group format instead of in a traditional intimate counselling environment, are often much less formal and are based upon peer support and resources rather than professional counselling.

A quantitative study conducted in 1988 in New York did a survey on the partnership of professionals with genetic support groups (Black & Weiss, 1988:21-33). The participants of the study consisted of affected individuals, their family members and professionals collaborating with SGs. They reported that genetic counsellors work directly with SGs as guest lecturers, consultants and advisory board members. Not only do counsellors provide members with information and assistance about genetic aspects of disorders, they also provide information and assistance regarding problems of daily life.

The following detailed services by genetic counsellors were reported:

- 1) "Education about genetic aspects of the disorder
- 2) Help with adjusting to the personal or emotional meaning of the disorder
- 3) Help with concerns that might arise when informing extended family of the genetic nature of the disorder
- 4) Help with concerns arising in informing one's children about their reproductive risks
- 5) Help with decision making about genetic risks to offspring
- 6) Help with any other genetic concerns of members and their families"

(Black & Weiss, 1988:21-33)

A similar, more recent quantitative study conducted in 2006 in Cincinnati explored the partnership between genetic counsellors and genetic advocacy groups from a genetic counselling perspective. Similar themes to the 1988 study were identified but included additional aspects that include serving on a group advisory board, volunteering for practical help at events (such as fundraising, child care during meetings, working at summer camp), acting as liaisons between group members and external parties and referring people to the SGs (Williams, 2006:16). The study also found that genetic counsellors contribute their own special skills such as web development, networking, grant writing, and public policy/advocacy skills.

1.5 HUNTINGTON DISEASE

Huntington disease (HD) is an inherited neurodegenerative disorder that affects patients physically, mentally and cognitively. The condition is life-limiting and influences individuals and families physically, psychologically and socially (Meissen, Maguin & Woodruff, 1987:245). It is considered a rare disease; due to the genetic nature of the condition, incidence estimates

may vary considerably depending on the population under study (Pringsheim et al., 2012:1084).

The incidence of HD is estimated to be 5-10 per 100,000 in North and South America, Europe and Australia, with a lower prevalence in Asia and Africa (Goldman, 2015:299). In western societies the incidence of HD is estimated to be 1: 100,000 (Rosenblatt & Frank, 2015:S1). South African incidence has previously been reported as 2: 100,000, but a lack of updated data may be hindering an accurate figure (Scholefield & Greenberg, 2007:590). Evidence does, however, suggest that the prevalence of HD will continue to rise due to new mutations which are suspected to account for 10% of diagnosed cases (Pringsheim et al., 2012:1084).

HD symptoms are caused by progressive breakdown of nerve cells in the brain and include disturbances in movement (involuntary non-repetitive dance-like movements known as Huntington's chorea), dementia and a variety of psychiatric difficulties such as depression, agitation, irritability, apathy, anxiety, delusions, and hallucinations (Kowalski et al., 2015:157). The condition is currently untreatable, and management is focused on controlling symptoms (Kowalski et al., 2015:157). The course of HD is irreversible and ultimately fatal (Rosenblatt & Frank, 2015:S1).

HD typically goes undetected for the first decades of life with symptoms usually presenting between age 30 and 50 (Meissen, Maguin & Woodruff, 1987:245). By the time symptoms appear, affected individuals often already have their own children who are each at 50% risk of inheriting HD due to its autosomal dominant (AD) pattern of inheritance (Charles et al., 2017:2). Despite generally having an adult presentation, age of onset (AOO) is variable and symptoms occasionally start in childhood (juvenile onset) (Dipple & Evans, 1998:286). The duration of the condition from AOO to death ranges between 15 and 20 years and gives rise to a need for long-term management of the condition (Dipple & Evans, 1998:286).

The HD phenotype is usually caused by a cytosine-adenine-guanine (CAG) trinucleotide repeat expansion within exon one of the Huntingtin (*HTT*) gene located on chromosome four (Pringsheim et al., 2012:1083). The increased number of CAG repeats create the toxic presence of a mutant form of the protein huntingtin (Htt) which serves an important yet incompletely understood function for nerve growth and development (Kowalski et al., 2015:157).

A CAG trinucleotide repeat of up to 26 is considered normal, but whenever the trinucleotide repeat number is increased, a neuron-killing, mutant form of huntingtin protein (mHtt) is

created (Kowalski et al., 2015:157). The threshold for 100% penetrance is ≥ 40 CAG repeats and is associated with disease expression (Goldman, 2015:300).

There are two other ranges in which a CAG repeat can fall, and it correlates with disease expression and age of onset (AOO) of the condition. HD generally develops in the third or fourth decade, but AOO can vary widely depending on the number of CAG repeats present. These two ranges are described as follows:

Reduced penetrance (RP)

The repeat range of 36–39 CAG is seen as a “grey area.” It demonstrates reduced age-dependent penetrance with some individuals becoming affected, but usually at a later age than the average AOO (Goldman, 2015:300). In this CAG range symptoms can be milder with a slower disease progression (Semaka & Hayden, 2014:303).

Intermediate allele (IA)

Intermediate alleles (IAs), or premutations, have been identified to have 27-35 CAG repeats and fall below the threshold required for disease expression (Semaka & Hayden, 2014:303).

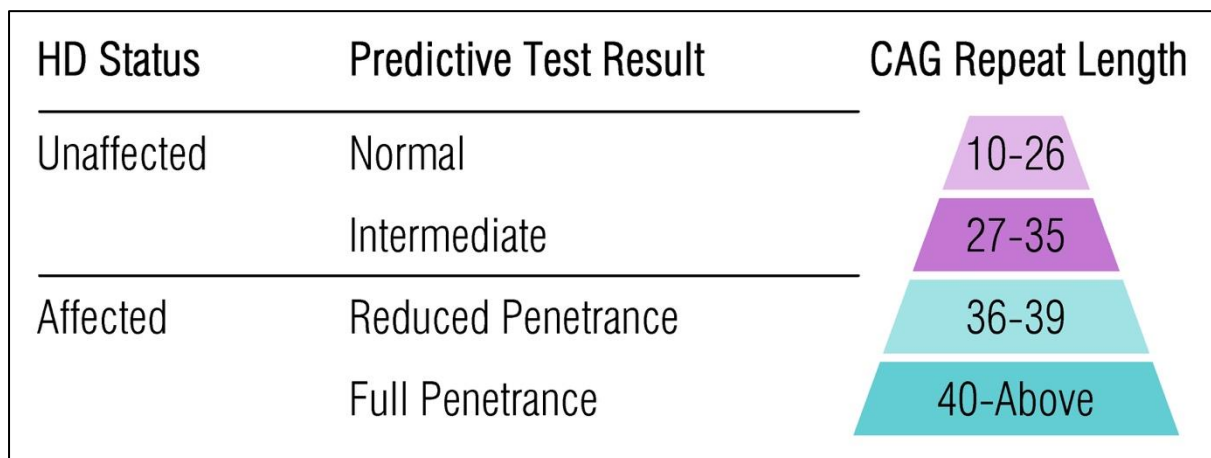


Figure 1: CAG repeat lengths and ranges in HD
 (Based on <https://en.hdbuzz.net/027>)

The IA range was initially thought to be asymptomatic, yet meiotically unstable so that germline expansion could occur (Goldman, 2015:300). However, a controversial study has reported a behavioural/psychiatric phenotype without motor or cognitive symptoms where subjects showed greater irritability, anxiety, depression, obsessive thinking and apathy when compared to control subjects (Goldman, 2015:300).

As mentioned above, an inverse correlation of 60-70% has been found to exist between AOO and the number of CAG repeats (Goldman, 2015:300). Consequently, a patient with 70 or 80 repeats may present with HD during childhood (juvenile onset) while a patient with 40 repeats may only present when he or she is older (Rosenblatt & Frank, 2015:S1).

When HD is passed down from generation to generation, the phenomenon of anticipation can sometimes be seen where earlier AOO is observed in following generations. The earlier AOO is due to germline expansion of the CAG repeats in the HD gene (Semaka & Hayden, 2014:303). Anticipation can also cause the offspring of unaffected individuals with IAs to develop HD later in life (Semaka & Hayden, 2014:303).

Germline expansion is much more likely to occur during paternal transmission than maternal transmission (Goldman, 2015:300). Seldom, and mostly during maternal transmission, a small contraction is observed (Goldman, 2015:300).

IAs are commonly seen in families in which a de novo (new) mutation has occurred, but are also accidentally discovered on the unaffected side of families with a history of HD (Semaka & Hayden, 2014:305).

HDL2 disease

Mutations in the *HTT* gene are the cause of HD in the greatest majority of families worldwide (Krause & Greenberg, 2008:193). However, a similar neurodegenerative disorder caused by a CAG/cytosine-thymine-guanine (CTG) trinucleotide repeat expansion in the Junctophilin-3 (*JPH3*) gene on chromosome 16, have been identified in families with a similar phenotype as HD (Krause & Greenberg, 2008:193). This condition is named Huntington disease-like 2 (HDL2) disorder and it is reported to have only been observed in individuals of Black Ancestry (Bardien et al., 2007:2084). It is thought that HDL2 is due to a founder effect or recurrent mutations on a predisposing haplotype (Bardien et al., 2007:2084) and more than half the cases reported worldwide are from South Africa (Krause & Greenberg, 2008:193). The discovery of HDL2 is especially significant to the South African population as South Africans with a Black Ancestry are almost as likely to have HDL2 as HD (Krause & Greenberg, 2008:193).

HDL2 has been reported to be clinically and pathologically indistinguishable from HD, however, features of Parkinson disease may predominate in some families (Krause & Greenberg, 2008:193). A study conducted on a South African family with HDL2 presented with progressive dementia and a movement disorder similar to HD. However, two of the patients

presented with clinical symptoms that differ considerably from HD, where one patient had myoclonus and the other had Parkinsonism (Bardien et al., 2007:2083). Images taken by brain magnetic resonance imaging (MRI) scans also showed features atypical for HD. The study concluded that the phenotype of HDL2 is broad and can differ from that of typical HD. The diagnosis of HDL2 should consequently be considered in a wide spectrum of neuropsychiatric and abnormal movement presentations.

Impact of HD

HD symptoms can be classified into three main domains: (1) physical symptoms, (2) cognitive symptoms and (3) psychiatric symptoms.

Motor impairments are the most striking symptoms of HD (Rosenblatt & Frank, 2015:S2). Huntington's chorea is excessive, involuntary, non-repetitive dance-like movements (Kowalski et al., 2015:157) and is considered the hallmark symptom of HD. It is most commonly associated with adult-onset HD (Rosenblatt & Frank, 2015:S2). The impairment of voluntary movement is seen more often in early-onset (juvenile) HD and includes symptoms such as dysarthria, dystonia, bradykinesia, rigidity, myoclonus, tics and tremors (Kowalski et al., 2015:158). Both the occurrence of involuntary movement and the impairment of voluntary movement can be very debilitating (Rosenblatt & Frank, 2015:S2). The progressive nature of the condition makes it difficult for patients to perform everyday routines as the condition results in losing abilities gradually over time (Kowalski et al., 2015:158). Purposeful actions such as walking, swallowing and talking become progressively challenging (Dipple & Evans, 1998:286) with individuals ultimately requiring complete assistance in activities of daily living (Rosenblatt & Frank, 2015:S7).

Cognitive dysfunction is always present as HD progresses, with subtle mental changes appearing up to 15 years before onset of motor symptoms (Rosenblatt & Frank, 2015:S2-3).

Cognitive impairment results in reduced mental speed, and impaired ability to organise information (expressed as a decline in concentration and mental flexibility) (Kowalski et al., 2015:158). Additional cognitive symptoms include deficiency in working memory and visuomotor performance and difficulty with attention, multitasking, problem solving, judgment and planning (Rosenblatt & Frank, 2015:S2-3). Perception is altered in several ways such as the loss of ability to recognise facial expression and experiencing altered perception of time, space and smell (Kowalski et al., 2015:158). Individuals experience a deficit of self-awareness and lose the sense that something is wrong with them (Kowalski et al., 2015:158).

Impulsive behaviour is also typically observed through activities such as stealing, gambling, inappropriate sexual activities and irritability (Kowalski et al., 2015:158). In addition, implicit memory deteriorates progressively – leading to the loss of well-practiced behaviours such as riding a bike, playing an instrument or driving a car (Kowalski et al., 2015:158).

Psychiatric symptoms such as depression is common in HD and an increase in suicidal behaviour is observed (Rosenblatt & Frank, 2015:S3). Other common emotional disorders in HD include major psychiatric disorders, psychosis, generalised anxiety (Dipple & Evans, 1998:286) and mania (Kowalski et al., 2015:159).

Various personality changes such as apathy, irritability, disinhibition, obsessiveness, impulsiveness and perseveration are typical symptoms of HD (Rosenblatt & Frank, 2015:S3). Violent temper outbursts due to loss of impulse control, confusion, and feeling overwhelmed are also common, and while the patient may quickly forget about the incident, it may leave family members upset and shaken (Kowalski et al., 2015:157).

Psychosocial impact and needs

The effects of HD can be physically, cognitively and psychologically overwhelming for both the individual and family (Meissen, Maguin & Woodruff, 1987:245). It has been reported that physical effects of HD can be overwhelming, but that the most devastating problem reported is the psychosocial impact of the illness (Meissen, Maguin & Woodruff, 1987:245).

HD patients are challenged by the loss of ability to perform everyday routine tasks as the disease progression takes its toll on the functioning of the individual (Kowalski et al., 2015:157). During the decade or two from AOO to death, the individual becomes increasingly dependent on others for their basic care. This duration in time gives rise to a need for long-term support to both those affected and their carers (Dipple & Evans, 1998:286).

Caregivers of individuals with a chronic disease such as HD experience stress due to the constant demands associated with patient care (Charles et al., 2017:2). The challenging nature of HD ideally requires support from both public and charitable agencies to help deal with challenges and social isolation (Charles et al., 2017:2). Functional support systems and networks of sympathetic, educated professionals are consequently important for the psychosocial wellbeing of the individual and the family (Meissen, Maguin & Woodruff, 1987:245).

Genetic counselling and testing

Due to the hereditary nature of the condition, genetic counselling plays a significant role for both patients and families in the management of HD. The counselling process consists of two main aspects that are inseparable from one another: psychosocial support and guidance (Semaka & Hayden, 2014:306) and the use of education as clinical tool (Nance, 2017:75). The main topics of counselling for HD include pre-test counselling, risk assessment, post-test counselling, prenatal counselling and testing and offering family members predictive testing (PT) (Semaka & Hayden, 2014:306).

Predictive and prenatal testing for HD used to be offered by linkage analysis, but since 1993 direct mutation testing for HD has been available (Creighton et al., 2003:462). In South Africa diagnostic testing for HD and HDL2 is offered by both academic laboratories, namely the Divisions of Human Genetics at UCT National Health Laboratory Service (NHLS) and Wits NHLS (Krause & Greenberg, 2008:193). Testing-options available include diagnostic testing, predictive testing and prenatal testing.

Diagnostic testing is available to symptomatic individuals where clinicians suspect the diagnosis of HD while PT is available to at-risk family members who would like to know their status. The South African PT process follows a protocol based on international guidelines that were published in 1994 and revised in 2013 and 2015 (Nance, 2017:80-81). The protocol helps navigate a person through the PT process including both genetic counselling and a psychiatric evaluation to determine the individual's readiness for testing and to prepare the patient for the results thereof (Goldman, Huey & Thorne, 2017:435).

Prenatal testing for HD is a controversial matter due to the condition's predominantly adult AOO and became available concurrent with the predictive test for HD (Goldman, Huey & Thorne, 2017:436-437).

1.6 SUPPORT GROUP FOR HUNTINGTON DISEASE

The International Huntington Association (IHA) is an umbrella organisation formed by HD associations over the world and they represent more than 250 000 individual members worldwide. IHA aim to connect people internationally in order to form collaborations in striving towards better care, treatment and research for HD (IHA, 2019).

Huntington's Association of South Africa (HASA) is a local organisation that operates in the best interest of people affected by HD in South Africa. Their mission is to stay informed of the latest developments in finding a cure for HD and to spread this information to their community through online channels and support groups (*Huntington's Association of South Africa, 2018*). HASA has an online Facebook support group as well as two physical support groups: one in Cape Town (CT) in the Western Cape and one in Pretoria in Gauteng (GP).

HASA Cape Town holds monthly support group meetings which take place every second Thursday of the month. Meetings are held at Abbots College, Claremont from 19:00 until 20:30. The group consists of people symptomatic with HD, and people who are at risk of developing the condition or who have tested positive but are pre-symptomatic, and their friends and family members.

1.7 RATIONALE FOR THE STUDY

From the literature above it is seen that some broad, international studies have been conducted to explore the role of genetic counsellors in SGs. No such study has, however, been conducted in our local, South African setting and no SG has been studied in detail.

The researcher became involved with HASA CT in the year prior to the conduction of the study. Her involvement with the SG lead to her questioning what role(s) she as a trainee genetic counsellor could play in the group – now, and as professional in future. Very few studies on SGs specifically relate to SGs for genetic conditions or the detail on the support they offer (Plumridge et al., 2012:256). Hence, this lack of South African and international literature on the topic motivated the researcher to design the study in pursuit of answering the following research question:

What role(s) can a genetic counsellor play in a support group for Huntington disease?

1.8 AIMS

The study aims to examine the views and opinions of professionals and members of the public involved with HASA in exploring the role of a genetic counsellor in the SG.

1.9 OBJECTIVES

- To explore what information regarding genetic aspects of the disorder patients would like to receive/discuss
- To determine what possible assistance a genetic counsellor could offer patients in adjusting to or living with the personal and emotional meaning of HD
- To learn how a genetic counsellor could help with concerns that might arise when informing extended family of the genetic nature of HD
- To determine what assistance a genetic counsellor could offer patients regarding their concerns of informing their children about their reproductive risks
- To explore the possible emotional support a genetic counsellor could offer in the group
- To discover what other genetic concerns counsellors could help with in the group.

Chapter Two

Methodology

2.1 INTRODUCTION

In this chapter the methodological design and framework that were used for the study are outlined. It starts off by explaining the researcher's involvement with the Huntington's Association South Africa's Cape Town branch (HASA CT) before commencement of the research study. It goes on to describe the study population and research setting, participant recruitment and data collection as well as the methods used for data analysis. Furthermore, reports on the validity and reliability of the study as well as ethical matters that were considered are also noted.

2.2 RESEARCH BACKGROUND

The researcher became involved with HASA CT during her first year of her master's degree in genetic counselling at UCT. Upon recommendation of her academic supervisors to join a SG of her choice, the researcher started attending the monthly HASA meetings in 2017.

The researcher's choice of SG was influenced by her background in Psychology and her interest in the psychological wellbeing of individuals confronted with a significant genetic condition. Her motivation for choosing HASA was to learn first-hand about HD and patient experience of a neurodegenerative condition.

While attending the SG in 2017, the researcher got to know many of the HASA members as well as the two psychologists who take turns in facilitating the group. HASA members consist of individuals diagnosed with, or at risk for HD as well as friends, family and carers. The researcher experienced the group to be very welcoming towards her and sometimes asked her questions regarding the clinical services at Groote Schuur hospital and UCT in the neurogenetics clinic.

During the researcher's initial involvement, one genetic counsellor with long-term involvement in the group visited the group, but most often it was only the researcher present from UCT's clinical team. After attending a couple of meetings, the researcher started questioning the role

of professionals in a SG. She was confronted with the question of how she as aspiring genetic counsellor could contribute to the wellbeing of the group members. There were also questions raised by the clinical team at UCT about the appropriateness of involvement and whether the presence of a counsellor could have a negative effect on the group. These questions inspired the researcher to approach the organisers of the group and ask permission to conduct the study. The request was greeted with optimism and verbal agreement was provided to conduct the study.

2.3 RESEARCH DESIGN

The study explores personal views and opinions of individuals involved with HASA. It requires participants to draw on their own thoughts of what role(s) a genetic counsellor can play in a SG and a qualitative methodology was consequently chosen as research framework.

Qualitative research is known for allowing researchers to interpret personal views and opinions and in so doing make use of individual frames of reference and experience of reality (Taylor, Bogdan & DeVault, 2015:7). Such a research paradigm is based on the perception that people's lived experiences provide valuable sources of information as people articulate their own experiences (MacFarlane, 2014:199). By using a qualitative approach, participants can voice their views without having to place the data in predefined categories.

Studies of a qualitative nature are a "natural" methodological choice in genetic counselling, because genetic counselling training and clinical practice involves interacting with people around sensitive and complex topics such as the significant impact that HD has on individuals and families (MacFarlane, 2014:199). The skills that one develops through counselling patients, couples and families are similar to the type of skills needed to conduct individual and group interviews (MacFarlane, 2014:199). The study consequently makes use of individual interviews and focus groups that were conducted over a four-month period.

2.4 RESEARCH SETTING

As discussed in the literature review, HASA CT holds monthly support group meetings on the second Thursday of each month. The group meets in person under the management of the chairperson and is facilitated by two psychologists that take turns in leading the group.

By the time the researcher started conducting the study, HASA CT had already existed for approximately 7 years and were a well-established SG functioning in the manner described above. During her visits the researcher observed the SG's attendance to fluctuate between 5 and 15 people per meeting, although the organiser reported that numbers can range from 10-30 attendees per meeting.

The attendees consisted of three groups of people:

- HASA members (individuals with HD or at risk for HD, as well as friends, family and carers)
- Two psychologists who act as group facilitators
- Genetic counsellors (professional and in-training)

Meetings start at 19:00 with group members “catching up” over a quick cup of coffee and biscuits. It proceeds with all attendees sitting in a circle introducing themselves briefly to the group, mentioning the minutes of the previous meeting and setting the agenda for the current meeting. The group then either stays seated in a single circle with members taking part in the same conversation or splits up into two groups: one group for the affected members and one group for the caregivers. During “single group” meetings the two psychologists act as co-facilitators and during “split group” meetings each psychologist facilitates one of the groups. If there is a genetic counsellor present during a “split group” meeting, she usually takes part in the caregiver's group.

The meetings alternate monthly between a “single group” and a “split group” in order to alternate and enhance the type of emotional support members get from the group. Under the leadership of the facilitator(s) the group discusses relevant topics or concerns in an open and flexible manner without a predetermined structure as to how the meeting should proceed. At approximately 20:30 the facilitator brings the group's attention to the nearing end and facilitate the closing-off of the meeting.

2.5 RESEARCH POPULATION

The participants of the study are people who have been involved with a HASA SG – either in CT or GP. Although the researcher was involved with HASA CT, she extended the scope of the study to include members involved with HASA GP to make sure that data saturation could be reached. The population consists of three distinct groups within HASA: group members, genetic counsellors and psychologists as group facilitators.

Inclusion criteria stipulated that participants needed to have attended at least one HASA SG in South Africa in the past 7 years (the approximate amount of time that HASA CT had existed by the time recruitment started). Participants further needed to be at least 18 years of age and be able to give informed consent. Participants had to be either Afrikaans or English speaking since these are the languages the researcher can communicate fluently in.

Exclusion criteria stipulated that affected individuals in an advanced stage of HD were not eligible to take part in the study. This stipulation was due to the cognitive and communicative difficulties that the condition causes as well as possible impaired ability to give informed consent.

2.6 PARTICIPANT RECRUITMENT

The participant sample consisted of unequal numbers of HASA members and genetic counsellors and only one psychologist represented her “category”. However, the proportion of HASA members to genetic counsellors to psychologists were representative of real-life involvement, where significantly more HASA members are involved with the group than genetic counsellors, and SGs in general seldomly have psychologists involved.

Even though the population was small (total of 17 participants), it represents a large population relative to the total amount of HASA members or professionals involved with HASA.

Recruitment for the study was done through two separate processes – one process for recruitment of the SG members and the psychologist, and one process for the recruitment of genetic counsellors.

Recruitment of SG members

SG members were informed of the study by the researcher herself whom many of the members were already familiar with due to her prior involvement with HASA CT. After obtaining ethical approval for the study, the researcher distributed information on the study at two of the monthly SG meetings. The distributed documentation included information on the study and consent to be contacted by the researcher for participation (Appendix A). Completed forms were handed back to the researcher by the end of the SG meetings. After receipt of these, the researcher contacted the willing participants either via email, telephone or in person at a SG meeting, explained the research in detail and allowed questions from the participants.

Recruitment of psychologists

The researcher initially did not plan on interviewing the psychologists, but due to introducing the study at the SG meetings the psychologists were aware thereof. One of the psychologists approached the researcher and volunteered to take part in the study. After discussion with her supervisor, the researcher decided it could be beneficial to the study to gain a viewpoint that is separate from the HASA members and genetic counsellors. The researcher consequently provided the psychologist with the same documentation as the SG members (Appendix A). A telephonic interview with the psychologist was scheduled.

Recruitment of genetic counsellors

The researcher asked the organiser of HASA CT to contact the GCs who had attended the group in the previous 7 years in CT and those that she knew of in GP. The organiser informed the GCs of the research study and gave the researcher the email addresses of those who were willing to be contacted regarding possible participation in the study. The researcher then sent the same documentation (Appendix A) that was distributed at the SGs to the genetic counsellors and proceeded to make individual interview appointments with those who showed interest in the study.

2.7 DATA COLLECTION

The study made use of individual interviews and focus groups (FGs) as methods of gathering data, collectively referred to as data gathering events (DGEs). Nine DGEs took place that consisted of two FGs, three in-person interviews and four telephonic interviews with a total of 17 participants. The DGEs with participants (P) are summarised in Table 1 below:

Data collection event	Number of participants involved	Category of participant	Participants involved
Focus group 1:	8 (+ 1 non-participant)	7 HASA members 1 genetic counsellor	P3, P4, P5, P6, P7, P8, P9, P10
Focus group 2:	3	HASA members	P12, P13, P14
In-person interviews:	3	Genetic counsellors	P1, P2, P10
Telephonic interviews:	4	2 genetic counsellors 1 HASA member 1 Psychologist	P11, P15, P16, P17

Table 1: Summary of DGEs and participants involved

Before the conduction of the FGs and interviews, participants were handed consent forms to read, ask questions about and sign. The consent forms explained the study and stipulated that participants gave permission for the DGEs to be audio recorded (Appendix B). Participants were also given anonymous socio-demographic information sheets to complete (Appendix C).

Each DGE was audio recorded in order to allow the researcher to be able to listen to the events and transcribe them afterwards. The researcher took limited notes during the DGEs so as not to disturb the focus from what was being discussed. However, after each DGE the researcher made ample notes and audio recordings of detail regarding the event to preserve as much of her observations as possible.

Interviews

A total of seven individual interviews were conducted. Three genetic counsellors were interviewed in-person in a private room on UCT's medical campus. Four telephonic interviews were conducted including two interviews with genetic counsellors, one interview with a HASA psychologist and one interview with a HASA member who were not able to attend either of the two FGs. The interviews were semi-structured and followed an interview guide tailored to suit an interview with either a genetic counsellor (Appendix D), a psychologist (Appendix E) or a HASA member (Appendix F). The participants of the individual interviews are visually depicted by the graph in Figure 2 below.

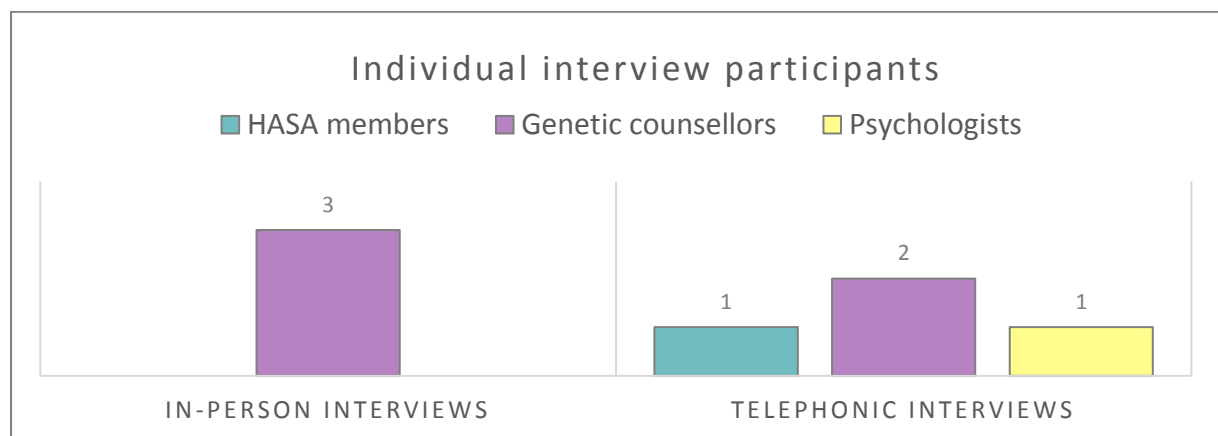


Figure 2: Graph representing participants who took part in individual interviews

Focus groups

Focus groups were used to interview the majority of the HASA members and were warranted for two reasons:

Firstly, group members are used to communicating and responding to one another's conversations and ideas in group format. At SG meetings members sit in a circle with a clear view of each other while conversation unfolds between individuals in the group. A focus group functions similarly to a SG meeting – a method of communication that group members are familiar and comfortable with.

Secondly, the dynamics of a focus group mimics a “brain storming session” and allow for participants to build on one another's expressed opinions and ideas – thus allowing for potentially richer data to emerge than what would have come from individual interviews (Bloor et al., 2001:7). A semi-structured interview guide was used for the FGs (Appendix F).

Two focus groups were conducted. The participants of the focus groups are visually depicted by the graph in Figure 3 below.

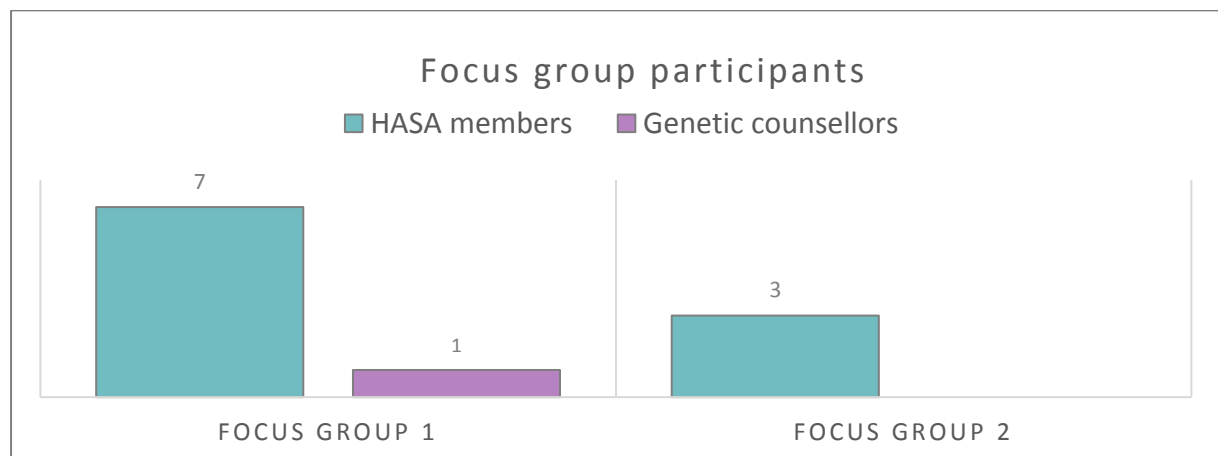


Figure 3: Graph representing participants who took part in the focus groups

FG1 was conducted at Abbott's College, Claremont on a Thursday evening after the SG meeting had come to an end. After the HASA members that did not want to take part in the research had left, the remaining members stayed behind for FG1 to take place.

It was initially anticipated that three people were going to take part in FG1 since these participants had shown interest in the study at a previous SG meeting and had stipulated that they would be joining the FG. However, during the course of the SG meeting earlier the evening, the study was mentioned and four more HASA members indicated that they would

also like to take part in FG1. The researcher enquired if the four additional individuals would be willing to take part in a subsequent FG at a different time, but all four indicated that they preferred to take part that evening due to not being certain if they would be able to attend a following SG meeting. The researcher did not want to risk losing the willing participants and agreed that all seven HASA members were welcome to take part in FG1.

It was intended that only HASA members take part in the FGs, but a genetic counsellor that attended the SG meeting also joined FG1. This happened due to the natural flow of the group of people from SG meeting into FG1. The researcher wanted to conserve the natural, unforced feeling of group communication and consequently allowed for the counsellor to take part in FG1 despite being scheduled for an individual interview later that week. This participant thus took part in two of the nine DGEs, a FG and an individual interview.

A family member of one of the participants were also present at FG1 because she was waiting for the person to take her home. This individual did not, however, actively take part in the research due to being severely affected by HD and not meeting the inclusion criteria of being able to give informed consent for the study. To show respect for, and acknowledgement toward the individual who was already sitting in the SG circle, she was included in the conversation. The researcher felt it imperative that the individual not feel marginalised and excluded from the group in which she finds acceptance and community. However, in order to protect her autonomy as individual who cannot give informed consent, questions were not directed at her, yet acknowledgements were made when she volunteered information such as her name. Despite acknowledgement of the individual's presence and contribution, her interaction was restricted by her severely affected speech and cognitive disease progression. The researcher referred to this individual as a "non-participant" in the transcription of the audio recording.

FG1 consequently consisted of eight individuals of which seven were HASA members and one was a genetic counsellor. The non-participant was not included in this number. FG1 took approximately an hour to conduct.

FG2 was conducted at one of the HASA members' residence on a Saturday morning. The group consisted of three HASA members and took approximately 50 minutes to conduct.

2.8 DATA ANALYSIS

The six-phase system of thematic analysis

Thematic data analysis that allowed for themes and patterns to be identified in the data were used to provide a rich account of the opinions expressed during the DGEs (Wagner, Kawulich & Garner, 2012). The analysis was done according to a six phase analysis system (Braun & Clarke, 2006) as depicted by Figure 4 below.



Figure 4: Six phases of thematic analysis (summarised)

(www.researchgate.net/figure/Summary-of-the-six-phases-of-thematic-analysis_fig2_305144308, based on Braun and Clarke, 2006:35)

Phase 1: Familiarisation with the data

The raw data obtained from the audio recordings was transcribed in Microsoft Word documents by a combination of an online software program named Sonix Transcription (producing text from the recorded audio which the researcher refined afterwards) and manual

transcription by the researcher herself. There was no need for translation of transcripts as all DGEs took place in English. All transcripts were checked against the audio recordings by the researcher to make sure that no mistakes or inconsistencies were present.

The data familiarisation phase concluded with the researcher re-reading the transcripts and marking ideas for possible codes to be identified (Braun & Clarke, 2006:17).

Phase 2: Generating initial codes

The coding process was done to help organise the data into meaningful sets of information. The data collection followed a “data-driven” approach where themes were revealed through the data and not through pre-determined theory (Braun & Clarke, 2006:18).

The coding was done concurrently with data collection. The researcher manually highlighted text line by line and made notes as far as she went, allowing for an inductive flow of themes to emerge. The entire data set was worked through methodically, giving undivided and equivalent attention to each data item (Braun & Clarke, 2006:18). The researcher identified interesting characteristics in the data items that formed the foundation of recurring patterns/themes throughout the data set (Braun & Clarke, 2006:18).

Phase 3: Searching for themes

The researcher used the coded data to focus the analysis on a broader level of themes and consequently grouped relevant coded data extracts within initial identified themes/categories (Braun & Clarke, 2006:19). Manually created mind-maps were used to visually represent how different themes and sub-themes could be used together to make structured sense of the coded data. The relationship between the initial themes were also explored visually with the use of mind-maps.

Phase 4: Reviewing themes

Themes were re-examined at two levels: at the level of the coded data extracts and within the entire data set (Braun & Clarke, 2006:19). The reviewed themes were discussed twice with the supervisor and reworked yet again to represent the data as logically and comprehensively as possible.

Phase 5: Defining and naming themes

After creating a satisfactory mind-map of the data, the themes were “defined and re-defined” in order to simplify the essence of each theme (Braun & Clarke, 2006:22). It was also

determined what the essence of each theme was and how the sub-themes complimented the systematic representation of the main themes.

Phase 6: Producing the report

The writing-up of the thematic analysis was done to tell the story of the data in a concise, coherent, logical, nonrepetitive and interesting manner (Braun & Clarke, 2006:23). Evidence for the themes were provided by using vivid examples and extracts from the data.

A participant code (P-code) was assigned to each participant to preserve the identity of the individual. All identifiable information was removed from the transcripts to maintain confidentiality, anonymity and to avoid bias during data analysis.

Data saturation

The researcher aimed to interview between 15 and 25 individuals from the three participant categories (HASA members, genetic counsellors and psychologists). After conducting and analysing the nine DGEs (with a total of 17 participants), it was decided that additional recruitment of participants was not necessary. Data saturation had been reached as no new data emerged from the last transcripts (MacFarlane, 2014:200).

2.9 TRUSTWORTHINESS

The ultimate aim for a qualitative research study is to be useful. This “usefulness” is measured by the trustworthiness of the study, which is defined as the degree to which the results can be believed (MacFarlane, 2014:53) and is measured by the following four criteria:

- 1) Credibility (how believable the results are from the participant’s perspective)
- 2) Transferability (the ability to repeat the study and get similar results using other respondents)
- 3) Dependability (how reliably the research is conducted in an ever-changing context)
- 4) Confirmability (the researcher’s ability to recognise her positional viewpoint, biases and preconceptions)

(Taylor, Bogdan & DeVault, 2015:243)

The researcher honoured the abovementioned principles by making use of the following during the study:

- 1) Following a systematic, structured approach to the data collection and transcription
- 2) Re-checking the verbatim transcripts to make sure the written text accurately reflects the audio recordings
- 3) A systematic, analytic approach to coding of the data
- 4) The use of direct quotes from the transcripts to prove the statements made in the research findings
- 5) Triangulation of results using data from three different participant “categories” (HASA members, genetic counsellors and a psychologist) each with a unique viewpoint of the role of a genetic counsellor
- 6) Self-examination and reflection of the researcher to make sure that her biases do not influence the data
- 7) Close discussion of data and themes with supervisor

2.10 ETHICAL CONSIDERATIONS

Ethical approval

Ethical approval was given for this study by the Medical Research Ethics Committee of UCT with the following reference number: HREC REF: 420/2018 (Appendix G).

Informed consent

Providing informed consent for participation in a research study is a process involving communication between the researcher and the potential participants; it is not just the signing of a form (MacFarlane, 2014:84).

Informed consent refers to the need for a potential participant to have enough information about a study to be able to make an informed decision about participation (MacFarlane, 2014:84). Not only do participants need to have enough information, but they need to understand the information, be of legal age to give consent (18 years and older) and they need to be cognitively competent without impaired mental status (MacFarlane, 2014:84). Because some of the members of the HASA community has HD that leads to cognitive decline, it was

especially important to make sure that potential participants were able to give true informed consent.

There was no objective clinical measure created to assess whether a participant was able to give informed consent or not. However, the researcher used her discretion based on observation of, and communicating with the HASA members over time to ensure that all willing participants were able to communicate in a group and understood the risks and benefits of the study.

Before the DGEs started, all willing participants were provided with adequate time to read the consent form (Appendix B) and to ask questions. The researcher highlighted the main aspects of the study as participants read through the consent forms. They then decided if they wanted to continue with participation or not.

For the two FGs the researcher verbally set group guidelines for the FGs that stipulated that FG members were to abide by the same rules as they do at the HASA meetings namely:

- Respecting one another's opinions when it differs from their own
- Allowing one another the opportunity to talk and express themselves freely
- Respecting one another's privacy and not disclosing one another's opinions to people outside the group

The researcher highlighted that although she would take all measures in her control to keep the identities of the participants anonymous, she could not guarantee that other people in the FG would not break confidentiality. She gave group members an additional opportunity to excuse themselves from the research assuring them that they would not be discriminated against in any way should they wish to not continue with the research. All prospective participants, however, agreed with the guidelines and willingly continued with their participation.

Privacy and confidentiality

The privacy and confidentiality of the participants were respected and upheld by the following methods:

- All confidential and identifying documentation were stored separate from the data
- Audio recordings of the interviews and focus groups were removed from the recording device after uploading it to the computer
- The researcher herself did the translation of the audio recordings

- The raw data was stored on a password protected computer that only the researcher had access to
- The data was de-identified and participants were given participant codes (P-codes)
- Data was only shared with the supervisor in order to help with the identification of codes and themes
- The data from the research is backed-up on an external device and will be deleted after conclusion of the study

Risks and benefits to participants

The potential harm of the research was that some of the research questions were of a sensitive nature and could have evoked an emotional response in the participants. The questions were designed so as to not ask too personal details, yet it was acknowledged that emotions could still have been triggered by discussion of these matters.

This risk to the participants was minimised by the way the researcher interacted with the participants. The researcher used her theoretical knowledge gained through the theory learnt in the Practices of Genetic Counselling and Principles of Genetic Counselling courses that she has completed. She also used her counselling skills set in her background in psychology and genetic counselling to treat the participants with empathy and sensitivity and assured participants beforehand that they could only answer the questions that they wanted to answer. This was especially ideal in the FGs as there was no expectation for anyone to answer specific questions and participants volunteered information/answers as they wanted to, similar to taking part in a SG. The researcher also mentioned to the participants that she could refer them to counselling with a professional such as one of the psychologists involved with HASA, should they feel it necessary to debrief after the DGE.

The benefit of the research to the participants is twofold. Firstly, it may be good for the participants to “tell their story” by expressing their opinions and emotions in a safe space. Secondly, the research aims to help genetic counsellors to benefit the HASA group, and by sharing their views and opinions the participants had a direct influence on this. Once the study is completed, the researcher will present her findings at a HASA CT support group meeting. The feedback that she will provide to the participants (and the other members of the support group) will include the major findings of the study as well as recommendations to enhance the functioning of the support group.

2.11 SELF-REFLECTION AND -POSITION IN RESEARCH STUDY

My involvement and interest in this research study stretch years into my past and my outlook on life of wanting to help others. Ever since my teenage years, I was drawn toward people going through a difficult time; wanting to lighten their burden.

In my mid-twenties I studied Psychology with the aim of becoming a counselling psychologist. I volunteered as group facilitator teaching life-skills to recovering drug addicts. I found the group-approach of sharing stories, experiences and advice to be greatly beneficial and complimentary to the African outlook of communal counselling – finding “help” through conversation with lay people in society.

As my interest developed into counselling involving medical conditions, I found inspiration and compassion in hearing the stories of others – in person and in reading online blogs. I recognised the benefit of people finding healing through the involvement of other human beings. After being encouraged as a first year Genetic Counselling Masters student to become involved with a SG, I did research on the local genetic SGs in CT. I decided on HASA as my SG of choice due to my interest in the unique psychosocial challenges that people with HD face.

I attended the SG a few times and I enjoyed getting to know the members and hearing their stories. However, I was confronted with individuals of our clinical team’s apprehension regarding the appropriateness of such involvement. Ethical questions were raised regarding whether we as counsellors could be beneficial to a SG or whether our involvement threatens the autonomy, privacy and unique functioning of such a group. I questioned my involvement as I reflected upon these concerns raised and I embarked upon a literature search regarding genetic counsellors’ involvement with SGs. When I was met with a lack of research on the topic, I decided to embark upon this study as my research project in partial fulfilment of my degree.

My prior positive experience with HASA played a role in my outlook on the study. From the beginning of the study I realised that I thought a genetic counsellor has a significant role in a SG and that this personal bias threatened the trustworthiness of the study. I consequently followed a methodical approach to data gathering, transcription, coding and thematic analysis. I made use of direct quotes, self-examination and self-reflection. I also discussed the themes and my personal outlook with my peers and supervisors and it helped me gain insight into distinguishing between my own opinions and those expressed through the data.

By following the measures described above, I tried my best to ensure the integrity of the study. I do, however, recognise that a qualitative study such as this can never be truly objective, and that this shortcoming should be acknowledged to be able to truly be transparent regarding the research findings.

Upon reflection I realise that the integrity of the study could have been compromised due to P10 (genetic counsellor) taking part in FG1. The mere fact that the counsellor was present could have influenced (and possibly elevated) the views of the SG members regarding the appropriateness of a counsellor in a SG.

Chapter Three

Results and discussion

3.1 INTRODUCTION

In this chapter the findings of the study are documented. The discussion starts with the sociodemographic information of the participants followed by the results and discussions. The results and discussions are reported under five themes that were identified during the data analysis and include excerpts of the verbatim-transcribed audio recorded at data gathering events (DGEs). Participant codes (P-codes) are used to distinguish between participants' contribution to the data without revealing individuals' true identities.

3.2 PARTICIPANT DEMOGRAPHICS

Seventeen individuals were interviewed of whom eleven were HASA members, five were genetic counsellors and one was a psychologist. One participant (P15, a counsellor) was from HASA Gauteng (GP) and the other 16 participants were from HASA Cape Town (CT). Of the seventeen individuals, eight took part in focus group 1 (FG1), three took part in focus group 2 (FG2), three were interviewed in-person and four had telephonic interviews. All participants were interviewed only once except for P10 who took part in both FG1 and Interview 3. Participants' involvement in the various DGEs is depicted by the graph in Figure 5 below.

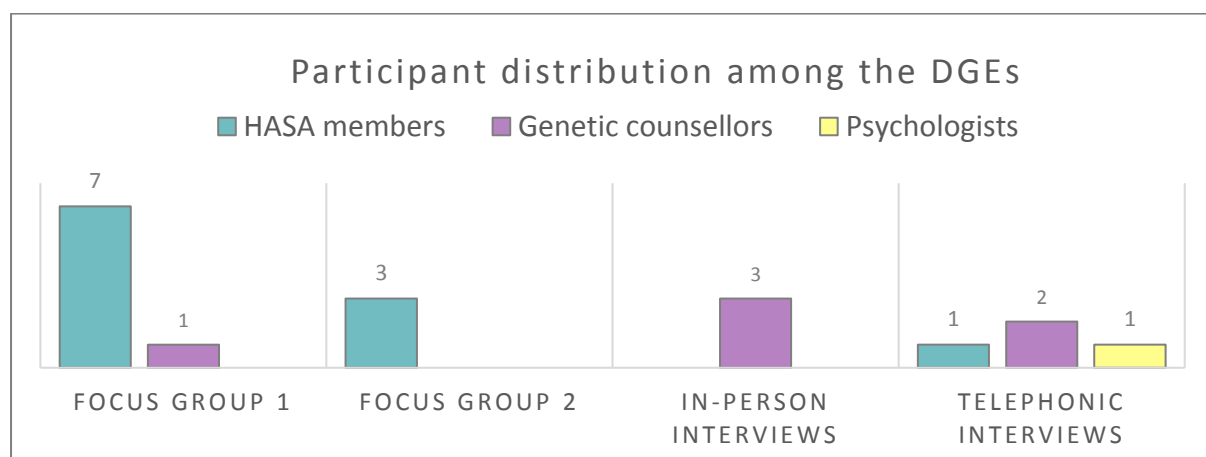


Figure 5: Graph depicting participant distribution among the various DGEs

Participants' time of involvement with HASA varied from two SG meetings attended, to regular attendance over the past seven years (the approximate length of time HASA CT has existed). A distinction was made between members with a short-term involvement (attendance of a few SG meetings) and those with long-term involvement (regular, ongoing involvement for a significant amount of time).

The graph in Figure 6 below shows the breakdown.

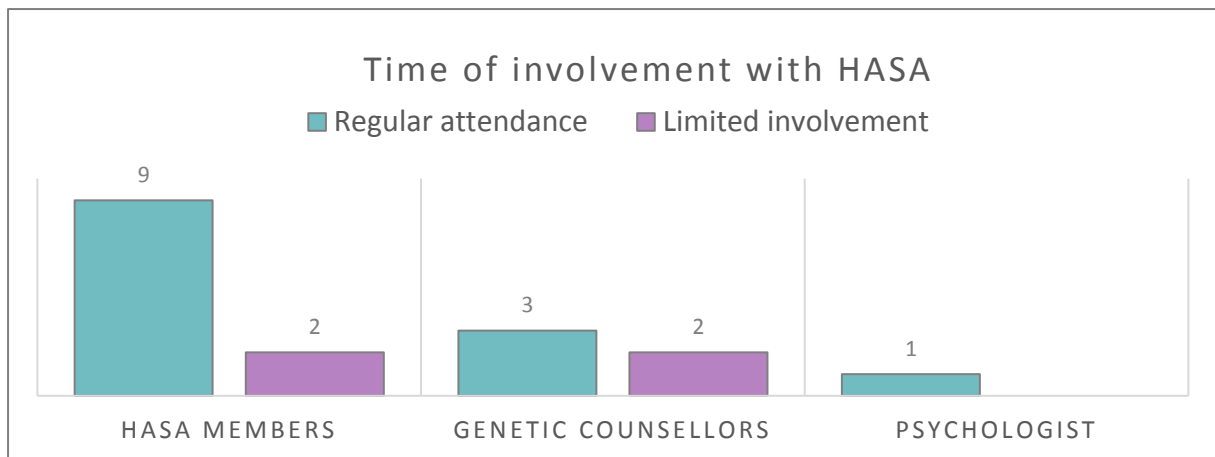


Figure 6: Graph representing the time of involvement with a HASA SG

A summary of participants' sociodemographic information is reflected in Table 2 below. It shows the participant, whether the interview was in person or telephonic, if the participant was a genetic counsellor, SG member or the psychologist, and how long they had been involved with the group.

P-code	Data gathering event number	Nature of interview	Relationship to HASA	Experience with HD	Time of involvement	HASA branch
P1	Interview 1	In-person	Genetic counsellor	Clinical work	Long-term involvement	CT
P2	Interview 2	In-person	Genetic counsellor	Clinical work	Short-term involvement	CT
P3	Focus group 1	In-person	HASA member	Family member affected	Long-term involvement	CT
P4	Focus group 1	In-person	HASA member	At risk for HD	Short-term involvement	CT
P5	Focus group 1	In-person	HASA member	At risk for HD	Short-term involvement	CT
P6	Focus group 1	In-person	HASA member	Gene positive, symptomatic	Long-term involvement	CT
P7	Focus group 1	In-person	HASA member	Family member affected	Long-term involvement	CT
P8	Focus group 1	In-person	HASA member	Family member affected	Long-term involvement	CT
P9	Focus group 1	In-person	HASA member	Gene positive, symptomatic	Long-term involvement	CT
P10	Focus group 1	In-person	Genetic counsellor	Clinical work	Short-term involvement	CT
	Interview 3					
P11	Interview 4	Telephonic	Genetic counsellor	Clinical work	Long-term involvement	CT
P12	Focus group 2	In-person	HASA member	Family member affected	Long-term involvement	CT
P13	Focus group 2	In-person	HASA member	Gene positive, asymptomatic	Long-term involvement	CT
P14	Focus group 2	In-person	HASA member	Gene positive, symptomatic	Long-term involvement	CT
P15	Interview 5	Telephonic	Genetic counsellor	Clinical	Long-term involvement	GP
P16	Interview 6	Telephonic	HASA member	Family member affected	Long-term involvement	CT
P17	Interview 7	Telephonic	Psychologist	Clinical	Long-term involvement	CT

Table 2: Sociodemographic information of participants

Additional participant characteristics are depicted in Table 3 below.

Characteristics	N	%
Gender (N = 17)		
Male	5	29.4
Female	12	70.6
Age groups (N = 17)		
18-25 years	1	5.9
26-35 years	6	35.3
36-45 years	2	11.8
46-55 years	5	29.4
56-65 years	2	11.8
66+ years	1	5.9
Ethnicity (N = 17)		
Caucasian	15	88.2
Mixed Ancestry	2	11.8
African	0	0.0
Current education level (N = 17)		
Matric (Grade 12)	1	5.9
Diploma	5	29.4
University degree	11	64.7
Relationship status (N = 17)		
Single	1	5.9
Stable relationship	2	11.8
Married	13	76.5
Widowed	1	5.9
Divorced/separated	0	0.0

Table 3: Summarised characteristics of participants

3.2 INTRODUCTION TO THEMES

Through in-depth analysis and careful study of the transcripts, five main themes and several sub-themes were identified, as illustrated by the graphic in Figure 8 below.

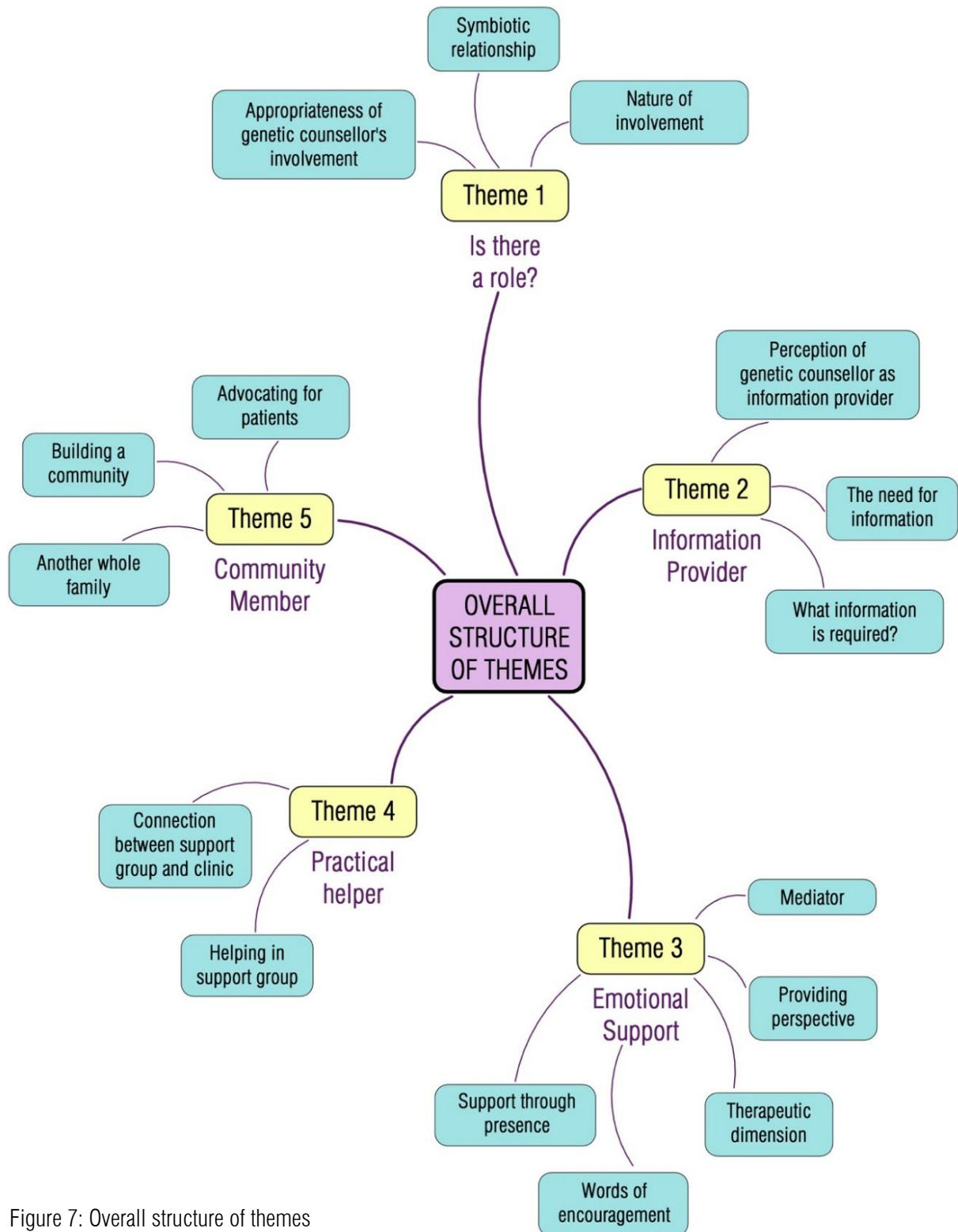


Figure 7: Overall structure of themes

The five themes identified represent the responses of all three participant categories (HASA members, genetic counsellors and the psychologist). The themes were developed as they unfolded from the data and do not separate the data according to category of participants. Each theme does, however, start with a symbolic diagram depicting which participant category was more prominently quoted in that specific theme. Where more than one participant category quoted the same concept, the most significant quote based on its content was used rather than attempting to balance the number of quotations between the three participant categories.

As explanation of how the diagrams were used in the write-up, three example-diagrams are featured below. The size of the diagram circles is a visual representation of the contribution that each participant category made to the specific theme. Example-diagram 1 illustrates equal contribution while Example-diagram 2 shows that the counsellors were quoted most frequently, followed by the HASA members and followed by a small contribution by the psychologist. Example-diagram 3 illustrates that only the counsellors and HASA members were quoted of which the HASA members made the greatest contribution.

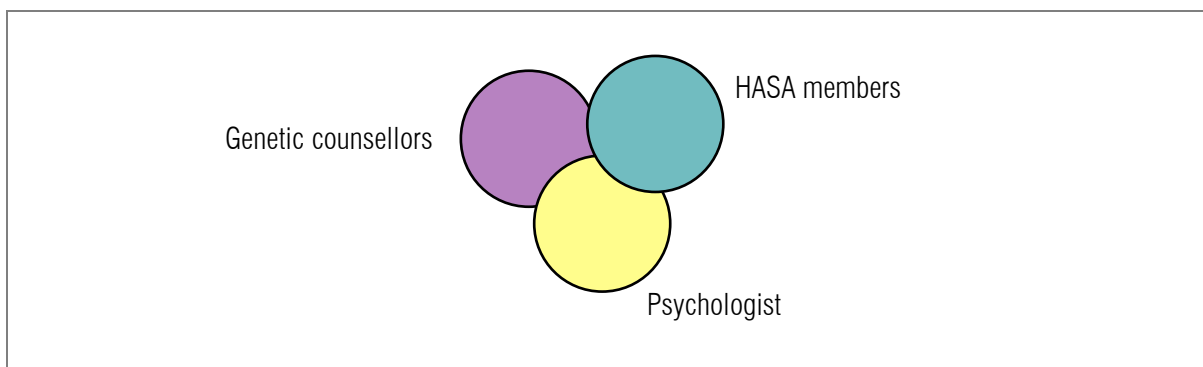


Figure 8: Example-diagram 1

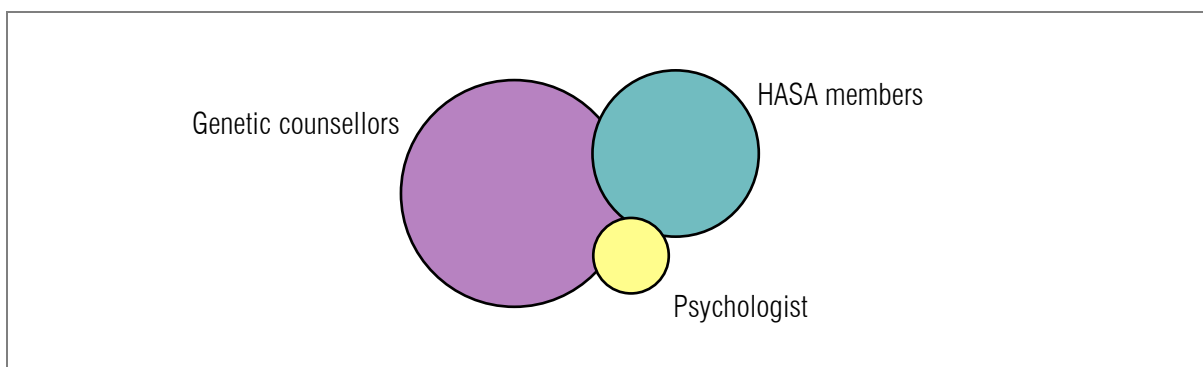


Figure 9: Example-diagram 2

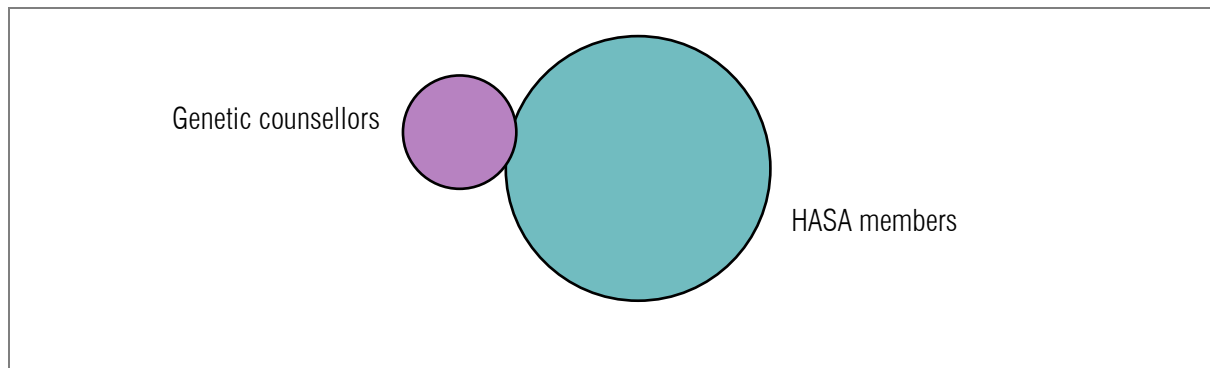


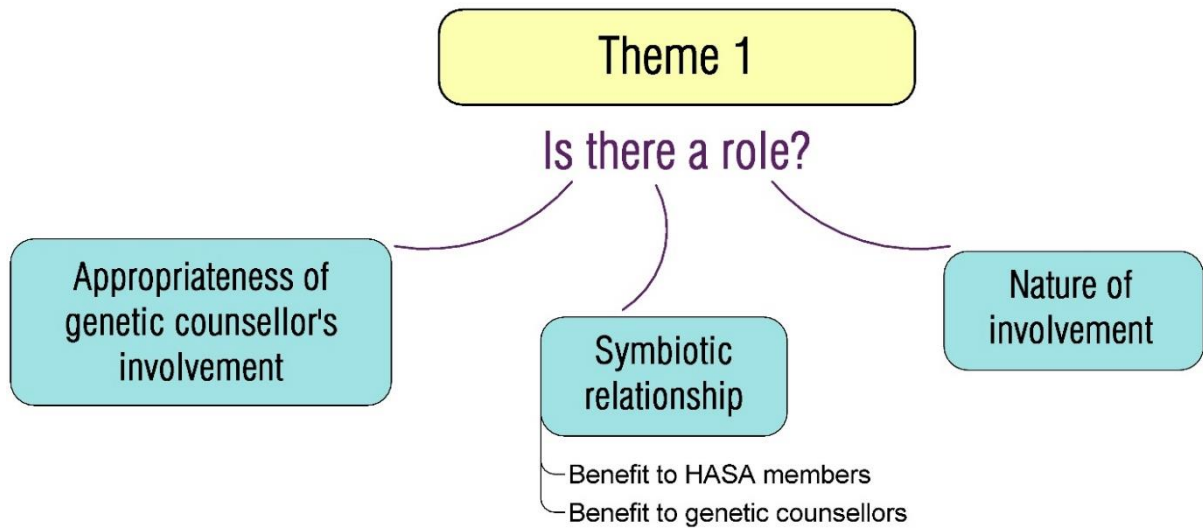
Figure 10: Example-diagram 3

3.3 THEME 1: IS THERE A ROLE?

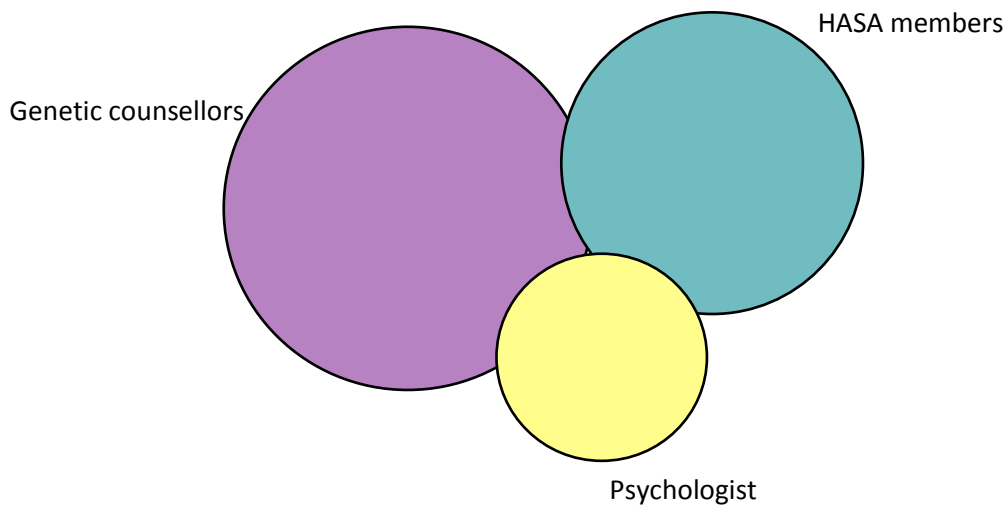
“It was actually a very positive thing that they said, uhm from that experience basically just saying that they felt that it was a great asset to have people there that they could ask questions, but also I think it was more the gesture of knowing that people that maybe weren’t directly impacted actually cared enough to come and support and show an interest.” – P11, counsellor (Interview 4)

The research study aims to answer the question of what the role of a genetic counsellor can be in a SG for HD. Before this question can be answered it is important to know whether there actually is a role for a counsellor in a SG. In theme 1 this question is addressed under the following three sub-themes:

- Appropriateness of a genetic counsellor’s involvement
- Symbiotic relationship
- Nature of involvement



Theme 1 represents the opinions of genetic counsellors and HASA members most strongly, followed by a smaller, yet significant input from the psychologist.



Appropriateness of a genetic counsellor's involvement

As mentioned before, student- and professional genetic counsellors are encouraged to become involved with SGs to learn more about the SG context and to engage with the partnership that exists between professionals and SGs. This encouragement is, however, accompanied by ethical concerns regarding the preservation of SG members' privacy, confidentiality and their opportunity to communicate in a safe space with people who share a similar context. Being aware of these concerns, the researcher explored these matters during the DGEs in determining the appropriateness of involvement.

Participant 4 explained that concerns were often raised by the clinical team during her involvement with HASA.

"Privacy was a big concern for some people from the clinical side, worrying that maybe that we as genetic counsellors being involved in the support group might hinder them from opening up; maybe it could be that the professional boundary would have been crossed."
– P11, counsellor (Interview 4)

Participant 2 explained that she shared the clinical team's burden and that her concern for privacy of the HASA members attending the clinic ultimately caused her to stop attending meetings.

"I just felt worried that perhaps people that attended clinic potentially wouldn't be feeling as free to be able to talk if they knew that I was there listening and they potentially couldn't share their true feelings, uhm, was a big concern so because of that we stopped attending."
– P2, counsellor (Interview 2)

The concern of the clinical team echoes the literature in which it is described that "respect for patient confidentiality is an essential feature of good medical practice" (Lucassen & Parker, 2003:93). The sub-speciality of genetics within the medical field has a special awareness of patient confidentiality due to the "diversity of potential harms, the wide variety of risks of occurrence of such harms and the variability of available interventions" (Lucassen & Parker, 2003:93) that is relevant to conditions that run in families.

Despite this concern voiced by the literature, clinical team and Participant 2 as quoted above, no other participants in the study expressed sharing the same privacy-related burden. Both the HASA members and the psychologist expressed that they did not feel that a counsellor's presence could be harmful to the members or the group.

"I have had not even little inklings of that it was a bad idea for you guys to be there or that the confidentiality was, I definitely didn't feel like that at all." – P17, psychologist (Interview 9)

"I don't think that to be, we think that you could do some harm to us, not at all. I think you can only have a positive effect." – P16, HASA member (Interview 6)

The opinion was raised that the burden regarding confidentiality was carried only by the clinical team and that the SG members did not share in this burden.

"In my experience I felt like it was more a hype from the medical, or clinical team, or concern from their side which I didn't feel was as much a concern from the patient's perspective, or the support group's perspective." – P11, counsellor (Interview 4)

"I know that the genetic counsellors... make a big thing about confidentiality and ethics and all that. I don't think any of us put half as much, ehm, emphasis on confidentiality and ethics the way you do. So I can't see any negative, ehm, effects on anybody. Somebody who doesn't want anybody to know anything will not pick up his hand and, and ask a question." – P16, HASA member (Interview 6)

Participant 11 expressed that she understood the reasoning behind the concern, but that she did not view the matter in the same way.

"There could be that breach in confidentiality, I suppose. It could be almost like a barrier for them opening up about certain complaints about the clinic. I didn't find that much, or I didn't feel like that was a concern." – P11, counsellor (Interview 4)

A possible reason for members not being concerned about encroachment on their privacy may be the view that the counsellor is a medical professional and that comes with a presumed "professional-patient" confidentiality.

"I don't believe anybody feels that this 'stranger' is, is not welcome or glad you go, because you're medical, you're medics in the medical field and that's all confidential anyways. I don't think anyone feels threatened, that's what I am trying to say, threatened by somebody new that's come and said I'm a genetic counsellor." – P12, HASA member (FG2)

It was, however, voiced that counsellors should be aware of the principle of patient confidentiality and that this should be preserved in the SG setting.

"But I think it is very important to, as a genetic counsellor, or to learn the skills during the process of being involved in the support group, about the breach in confidentiality issue." – P11, counsellor (Interview 4)

On responding to the question of whether a counsellor's presence could hinder SG members from opening-up in the group, most participants indicated that this was not what they had experienced.

"Sometimes I think it could stop people from talking. But most of the time people who want to talk will talk and people who don't want, don't talk." – P1, counsellor (Interview 1)

The psychologist expressed a similar viewpoint and explained that members sometimes open up and sometimes not, but that this "level of openness" relates to human nature and not the presence or absence of a counsellor in the SG.

"They don't always open up. There's sometimes weeks where it happens, but I find often it's very surface level chat... kind of avoiding the heavy stuff. But I don't think... that's because, you know, you're there or anything like that. I think that's just the nature of human [interaction] in a public space." – P17, psychologist (Interview 9)

In fact, all the genetic counsellors expressed that they have always been welcomed to the group by the members, even when they had not attended regularly.

"I've missed a lot in the last two years and when I go back there, all is very happy to see me." – P1, counsellor (Interview 1)

Participant 11 explained that the feeling of acceptance she found in the group transcended her physical involvement and that some relationships lasted even after her involvement with HASA had come to an end.

"They were very, very open and welcoming and almost became friends in a way, I'm even now in contact with some of the people and they often send me links like articles or new research on Huntington's disease or drugs or that type of thing. So I think it was a good, positive experience for both me and for them." – P11, counsellor (Interview 4)

During Participant 11's involvement she and her fellow counsellor conducted an informal anonymous survey to determine how HASA members felt about the presence of a genetic counsellor in their group.

"At one support group we actually did do, we brought it up in the group, and we did an anonymous survey just to ask if like us being at the support group was an issue for people, or if they felt that they couldn't open up about things even like their clinical experience or testing process or whatever." – P11, counsellor (Interview 4)

She explained that the feedback from the survey was positive and in favour of having a counsellor present.

“The feedback and the remarks we got back was actually very, very positive. I remember one of the guys from the support group said it's almost like a breath of fresh air having someone from a different perspective come to the support group, but also that they were so overwhelmed and touched that people that really didn't have any direct family member that was suffering from the condition actually cared enough to join in the group, come and provide information and support and all of that.” – P11, counsellor (Interview 4)

Participant 11 explained that the group facilitator explored the issue further at one meeting where no counsellors were present and that the members yet again gave favourable feedback regarding counsellors' involvement.

“There was one support group where neither [genetic counsellors] were at the support group, uhm and it was just the facilitator there who is completely separate from like our clinical team and basically she also brought it up again to just see if there were any issues with us being there because we were concerned because it was brought up at our clinical meetings that maybe we were crossing the professional boundary, but again it was very positive feedback and uhm ja, almost if they like, really valued us being there.” – P11, counsellor (Interview 4)

Symbiotic relationship

Throughout the data collection, participants indicated the existence of a mutually beneficial relationship between the counsellors and the members.

This opinion is supported by the findings of a study involving geneticists engaged with SGs. In the study it is stated that “health care practitioners learn as much from the group members as the group members learn from the health care providers” (Lin et al., 2003:89).

The benefit to SG members having counsellors present is seen in the many roles that counsellors play in the SG. These benefits are explored and described in Themes two to five as if forms the bulk of the research results and include benefits gained from receiving information, emotional support, practical help and assistance in growing the HASA community.

The results are complimented by the gratitude that SG members expressed towards professionals caring and giving their time to be involved with HASA.

“Like I said earlier, I don't think that like I honestly like. Just gratitude, just thank you, just complete gratitude, and the fact that you all come because you want to come, not because anyone, you come to help, you know you give assistance, you give practical advice or

specific genetic advice, so just a big grateful thank you from me, honestly.” – P13, HASA member (FG2)

“I feel like they're just so grateful to us for giving them some time and giving kind of, being able to give some kind of professional input and advice.” – P17, psychologist (Interview 9)

One HASA member expressed her gratitude towards the researcher's involvement with HASA as counsellor, but also specifically for conducting this research study.

“Thank you so much for, you know, coming to the support group and taking an interest. And also just doing your study on that to see, ehm, you know what... because even after you, ehm, there'll be something, there'll be some case study on support groups and so-and-so, that's really nice, thanks for doing that.” – P16, HASA member (Interview 6)

The benefit to the counsellors was expressed through a description of how attending the SG helps professionals shed insight into patients' lives with HD.

“I used to go to the meetings and started enjoying getting to know the patients. It gave me good insight into their, their other life, their real life.” – P1, counsellor (Interview 1)

The opinion was also expressed that counsellors benefitted by learning what patients truly want and need.

“They are helping you so much [to] understand the stages they go through and what they really need, what they need to know more about, also what they actually want when they come to the clinic.” – P11, counsellor (Interview 4)

“[It's] also a huge opportunity for a genetic counsellor to learn what patients actually want, I think that's a really big one.” – P16, HASA member (Interview 6)

In a study examining the participation of clinical geneticists in genetic advocacy groups, the benefit to the professional is also highlighted. Geneticists admitted that by being involved with SGs they get the opportunity to meet many people with interesting rare diseases (Lin et al., 2003:89).

There also exist less altruistic reasons for professionals to be involved with SGs. Participant 1 mentioned that she hopes to get referrals to her private practice by the exposure she gets through the SG.

“On a purely personal, well on a purely selfish level to get more ahm referrals, uhm through their family members and so on, which we haven’t as yet seen before.” – P1, counsellor (Interview 1)

This less altruistic motivation is similar to that of the geneticists in the previously mentioned study that described a SG as an ideal forum for research as it provides practical access to potential research participants and sometimes even funding for such research (Lin et al., 2003:89). Additional egocentric motivation for attendance was listed as ego/reputation, intellectual curiosity and duty.

Nature of involvement

The nature of involved with HASA was a topic that often came up during the DGEs. Opinions varied widely regarding when and how a counsellor can or should be involved.

Such practical considerations for SG meetings are described as logistical considerations that should be made according to the membership and needs of individual SGs (Goh, Lane & Bruckner, 2007:304).

Participant 16 explained that it would be useful if a counsellor could be present at every meeting so that there is always someone to explain the genetics correctly.

“Ideally I would like them to attend, ehm, each time. Ehm, each, each time there is a, you know, if it’s possible each time, because even the, the people who come to the support group, they might come and there’s nobody, and someone else in the support group starts explaining and it’s not quite correct... So I think almost each time we have a support group.” – P16, HASA member (Interview 6)

Another member expressed that when genetic questions come up and a counsellor is not present, members can always wait for a following SG meeting to have their questions answered by a counsellor.

“Some of the people don’t always come every month, so I suppose if you’ve said to them next month we will, you know, if you’ve brought it up next month we will have someone here.” – P17, psychologist (Interview 9)

Participant 7 explained that if permanent involvement is not an option, occasional visits and a formal talk at times would be sufficient.

“Even if you attend occasionally it would be nice to know that there’s a counsellor that attends, you know, and now and again maybe you can have [a] talk too.” – P7, HASA member (FG1)

A similar opinion was expressed by Participant 16 when she explained that she would prefer if there is consistency in one or two counsellors attending, but if that was not an option, having any counsellor attend is better than not having someone present.

“You know, it’s always nice if there’s consistency, but, if we can’t have consistency, we could rather have fifty people coming than nobody coming at all.” – P16, HASA member (Interview 6)

Some participants raised the practical point that at some SG meetings, the genetics of HD does not come up as conversational topic and that during those groups it may feel like the role of a genetic counsellor is redundant.

“The trouble is if there isn’t a genetic question asked, if nobody brings up a genetic question, you know it might just be a behavioural question. If nobody brings up a genetic question then there’s nothing for the genetic counsellor to get involved in.” – P12, HASA member (FG2)

It was, however, acknowledged that it’s not easy to determine during which SG meetings a genetic topic will come up and when not, due to the fact that SG meetings differ from meeting to meeting.

“I mean it’s hard to say how often, you know, it would be needed, ‘cause I suppose we also have different groups where it’s, ehm, you know the full group and then the split group and then the speaker and a full group and the split group and a speaker.” – P17, psychologist (Interview 9)

Participant 1 who attended the group for several years felt it was important for counsellors to be committed to the group in order to build relationship with the members.

“Not just come in once and then go away for the next year and then pitch up again... I really think that continuity would be good, you know to have say somebody old and in two days, in two, two sessions and so somebody new and then also pop in every now and again like I do.” – P1, counsellor (Interview 1)

Participant 13 also mentioned that regular attendance helps build relationship with the group, but that it should not be seen as a forced involvement and counsellors as visitors are also welcome.

"I think it's good to [attend regularly], because it's the relationship with someone. But I don't think it has to be like such a formal, 'you come', you know. I think, you come and if someone wants to come bring them along." – P13, HASA member (FG2)

With regard to the level of structure of a counsellor's involvement, some participants showed a preference for a structured approach while others showed preference for more informal involvement.

Participant 2, who stopped attending SG meetings, expressed that part of the problem for her was a lack of structure and specific expectation of what her role should be. She explained if there was opportunity for a more structured role she would attend SG meetings again.

"We'd be more than happy to be involved and do like topics you know talks or arrive for a certain portion of a meeting uhm but maybe on an invitational, more structured manner uhm but we stopped attending in terms of the patients just talking sort of sessions." – P2, counsellor (Interview 2)

She suggested as an alternative that a counsellor could attend regularly, but only for a part of the SG meeting allowing the HASA members privacy for the rest of the meeting.

"For the first half an hour we'll have you there but the second half an hour would be just patients talking so they would have their own private space." – P2, counsellor (Interview 2)

There were additional suggestions of how involvement can be more formalised such as doing structured presentations based on questions members had previously asked.

"Perhaps doing these like structured presentations, where you know, coming in and saying what is everybody's issues, you know what don't you understand about the condition, tell us and then next month we can have a short presentation on that or whatever the case is, trying to answer the understanding questions, because we're not always going to have answers for the emotional stuff anyway." – P10, counsellor (FG1)

"Occasionally you can do a structured talk like, being a genetic counsellor you can counsel on HD." – P5, HASA member (FG1)

It can also be that at the beginning or the end of the group, a short period of time can be set aside to speak swiftly on a genetic issue of HD.

"You know like an opening prayer, almost, like the genetic counsellor is going to talk about x,y,z, quick, 10 minutes, just to impart some knowledge, you know something like that." – P12, HASA member (FG2)

“Maybe they can be a bit, when I say formal, I think towards the end, is there anyone who would like to ask anything about the genetics, for the counsellors, and you know maybe there will be, maybe there won’t be. That’s ja, that’s fine.” – P12, HASA member (FG2)

Despite suggestions on how counsellor involvement can become more structured, many of the participants indicated that they preferred a less formal approach where counsellors attend meetings and help address issues as they come up.

“[I] think more informal. You know, I think people get to know the genetic counsellors, you know of the time, you know like [the counsellors], and they came quite regularly. And people would always, if something, you know and they were like, oh let’s ask [the counsellors] what do you think? But I do think that, just, just to be there like, like I said, learning from genetic counsellors as well, you know.” – P13, HASA member (FG2)

“I do think that we if we are there, then we really can be an asset to them if that’s something that they wanted, to answer questions. Not that we would just be in that capacity, but that if we are there, like a happy coincidence, then we could be an asset to them.” – P10, counsellor (Interview 4)

The many options regarding the ideal nature of involvement of a counsellor is aptly expressed by Participant 7’s reflection:

“Mmm, maybe time will tel. Maybe you got to try come occasionally and then maybe people will say we need you more often.” – P7, HASA member (FG1)

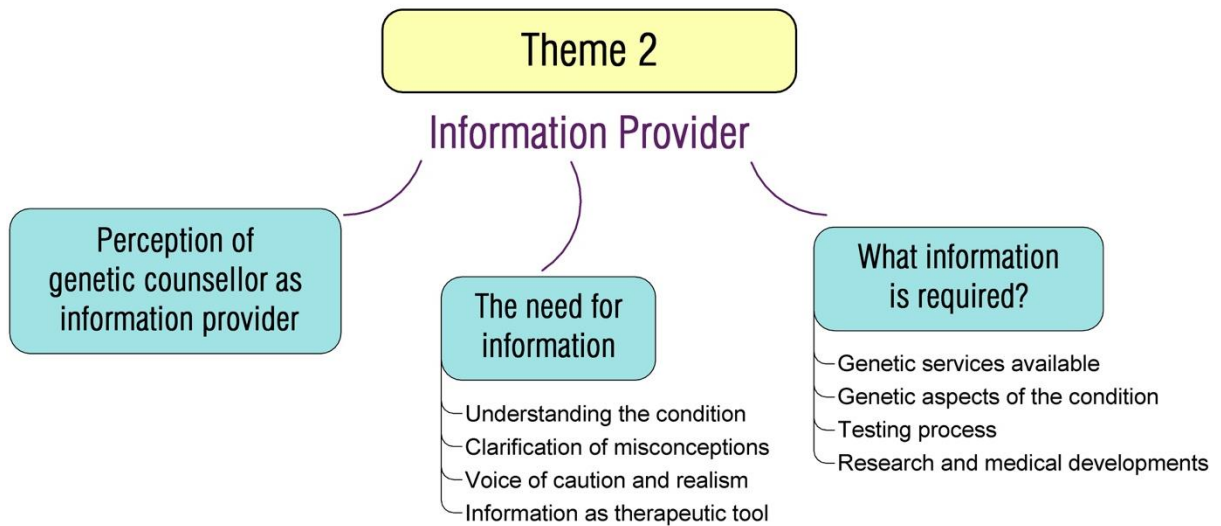
3.4 THEME 2: INFORMATION PROVIDER

*“Nobody can explain [genetic information] better than the genetic counsellor, because you know so much about the condition and you also know so much about counselling.”
– P16, HASA member (Interview 6)*

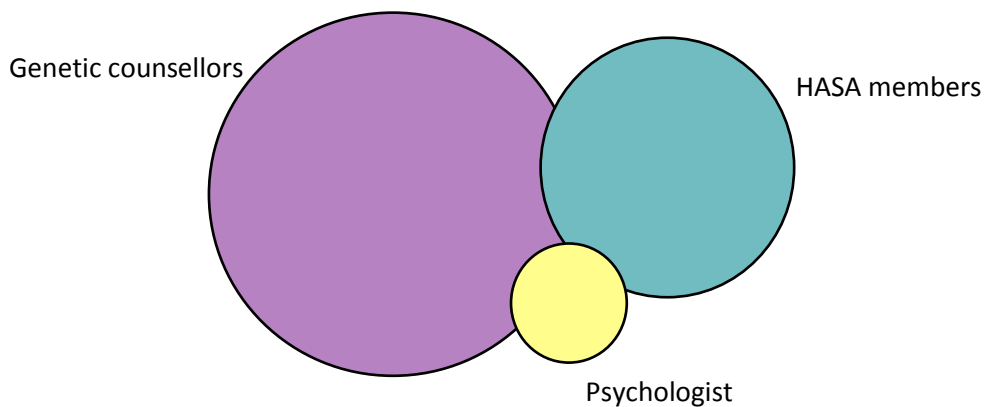
The role of a genetic counsellor as information provider was raised during most of the DGEs and stresses the importance of information in a genetically complex condition such as HD. A study in the USA showed that 76% of HD families noted an information and referral service is important to address their needs (Meissen, Maguin & Woodruff, 1987:248).

This theme is described under the following subthemes:

- Perception of genetic counsellor as information provider
- The need for information
- What information is required?



Theme 2 represents the opinions of genetic counsellors most strongly, followed by those of the HASA members and a small, yet significant contribution from the psychologist.



Perception of counsellor as information provider

With a medically complex condition such as HD, it is valuable to have an expert present to explain concepts and provide information. The unique knowledge and experience of genetic counsellors make them ideal people to provide necessary information on HD. Being qualified practitioners in the field of genetics, they have the credibility to give accurate and detailed information.

“You are able to explain what it's all about a lot better than, than any of us sitting there... I think a genetic counsellor is in a unique position to explain one niche that none of us, ehm, can explain... you know so much about, ehm, the condition and you also know so much about counselling. I mean you are highly qualified.” – P16, HASA member (Interview 6)

SG members view counsellors as information-providers and members know they can direct questions to the genetic counsellor at a SG.

“They're there and if there's something relevant, we'll ask them.” – P13, HASA member (FG2)

Several participants mentioned that the role of a counsellor is to be there for the group when questions arise. Participant 10 explained her role as an information provider and advisor to be in the background until her knowledge is needed.

“[To] be available should there be questions that arose, that people didn't know how to access services, or you know had questions about genetics so I was there just as a advisory role in the background.” – P2, counsellor (Interview 2)

Participant 11 explained that questions would often arise in meetings and that it was her role to answer and address them.

“Very often there were a lot of questions... my role was kind of to answer and address any questions that they had.” - P11, counsellor (Interview 4)

Participant 10 hypothesised that the provision of information by a professional is so significant that SG attendance could increase should it be general knowledge that a genetic counsellor is available to answer questions.

“I think some people would have come more often if they knew that... if they had the opportunity to have their questions answered as well.” – P10, counsellor (FG1)

When there is too big a focus on the genetic counsellor as information provider, it may happen that it is the only role some members feel counsellors can play in the group. One participant

explained that if no genetic topic is raised in a meeting, then there is no role for the genetic counsellor to play.

“The trouble is if there isn’t a genetic question asked, if nobody brings up a genetic question... then there’s nothing for the genetic counsellor to get involved in.” – P12, HASA member (FG2)

Need for information

Understanding the condition

The need for information is ingrained in the need to understand and make sense of the condition and was mentioned by multiple participants. Participant 11 described how she realised during her involvement with HASA that there was a big need for someone to help people understand the condition.

“I realised it was actually quite a big need for us to be there. Just in terms of for them understanding the condition.” – P11, counsellor (Interview 4)

It is not only important for people with HD to understand the condition, but it is also important for their family members. Participant 1 explained that the basic information need is the genetic risks to family members who are often the majority of the attendees.

“I think, the basic need is to understand more about the condition and to understand the risks to the family and so on. They don’t often talk about that direct risk to the family but it’s really families that are there.” – P1, counsellor (Interview 1)

In addition to blood relatives, friends and caregivers also need to understand the condition. Participant 1 expressed a unique need for caregivers to understand the progression of the disease to help them plan accordingly and ahead of time.

“Just understanding that where this condition is going to go and helping them to think about the future and how we’re gonna cope when the patients get really bad and so on, so ya just from a point of view of my experience working with people with Huntington’s this is very valuable for the other people who are with them experiencing this at this moment.” – P1, counsellor (Interview 1)

The need to understand the condition includes the importance of understanding the complex medical information of HD. Clear, accurate information can help individuals understand HD and its implications.

“I would think that the most important and obvious need would be the need for accurate information.” – P10, counsellor (Interview 4)

The need for information is addressed in traditional genetic counselling and aims to empower patients in making informed decisions. Participant 3 explained that SG members grasp the value of information within HASA as many of them have had genetic counselling in the past:

“It’s valuable information and we all can attest to the fact that everybody here has had genetic counselling and we’ve all found it valuable.” – P3, HASA member (FG1)

It is evident that despite being involved with HASA, past genetic counselling and vast amounts of information available online and via other resources, a need remains for someone to explain genetic information at SG meetings. Participant 10 explains the benefit of having a clinical representative present in the following quote:

“To be able to have someone there, that clinical representative, that, you know, can talk about genetic issues or answer questions.” – P10, counsellor (Interview 4)

The amount of information that is needed differs from meeting to meeting depending on the structure of the session and who is attending on that specific day. When the group consists of people who have been involved with HD for a long time, there is usually less of a need for information as many already know the detail and implications of the condition. The meetings when “newcomers” join the group are especially when information is needed.

“A lot of the time there would be new people that would come and ask questions about like the genetics behind it, how’s it inherited, and who’s at risk in the family and how they go about testing.” – P11, counsellor (Interview 4)

It is, however, difficult to determine when exactly information is going to be needed since attendants come and go as they wish:

“There’s definitely gonna be somebody at some stage [who needs information] and we never know when.” – P16, HASA member (Interview 6)

New members are not, however, the only people who need information. The complexity of the condition causes people to ask questions over and over again until they can make sense of the information for themselves. One of the counsellors explained that she came across certain members who would ask the same questions repeatedly:

“Any time that there’s a genetic [discussion] going on, they always, they always ask us umm to answer and explain again and we’ve had one or two members that ask us continuously.” – P1, counsellor (Interview 1)

One of the counsellors shared her experience with an HD family where the patient was diagnosed by a neurologist without receiving genetic counselling to explain and place the condition into context. The counsellor explained that these situations also require proper information and understanding to “fill the void” of understanding.

“That in those kinds of situations then there’s a lot of stress and anxiety put on the family without actually having a clear understanding of what’s happening, and they may just go to Google and try and figure out what this means for them and for the rest of their lives.” – P10, counsellor (Interview 4)

Participants explained the usefulness of having a genetic counsellor explain information at a SG meeting. Participant 3 explained that she did extensive research when a family member was diagnosed with HD and that it would have benefitted her if she knew there was a counsellor at the SG meetings who could have helped her to understand the vast amount of information she was trying to understand:

“I spent hours, hours online. I read medical journals, I was reading to understand CAG counts and, and I would have given anything to have somebody to sit me down and say this is what it means... and had that been an option in this group, and had the group known that there is, oh we’ve got genetic counsellors come to us sometimes, we can put you in contact with them, I, I would have taken that opportunity and done it.” – P3, HASA member (FG1)

An example of how information can help place detail regarding HD as condition into context was described by one of the participants. She explained that a family member was diagnosed with HD but did not receive genetic counselling with her diagnosis. Without proper knowledge of how her condition could affect her family, she did not understand the importance of telling her children about their risk of being affected and they consequently had children without being aware of their risks. The participant believes that had this family member received genetic counselling, she would have understood the importance of sharing the information with her children and in so doing she would have empowered them to make an informed decision when they decided to have their own children.

This type of information within a SG is valuable and can help affected or at-risk individuals to place the condition into context and think more broadly than just themselves.

Clarification of misconceptions

HD is a complex condition and details of it is often misunderstood. Misconceptions are often due to a lack of understanding of the genetics of HD. Participant 10 explained that when such misconceptions surface in a SG it can be corrected by concise and accurate information provided by a genetic counsellor:

“There have been situations where the genetics have come up and we've been in a position to help clarify something or to try and help with the understanding of something; that's been great.” – P10, counsellor (Interview 4)

Participants are not always aware of their misunderstanding and it is often up to the genetic counsellor to intervene in conversations when incorrect information is being shared. Participant 1 explained that questions were not necessarily directed to her, but she often had to intervene in conversations to clarify misunderstood information:

“They don't always ask us directly. They will sometimes be discussing it in a group and then we've got to say that this is actually what's going on. Not what they think, but we try and get them on the right track.” – P1, counsellor (Interview 1)

Participant 16 explained that she had seen that many people, even educated ones, had difficulty understanding the inheritance pattern of HD even after reading up information online:

“There's a lot of misconception, like, ehm, I think listening again at Mr X, whose wife had Huntington's... ehm, his daughter is quite educated, ehm, they went on the internet and they looked at everything and yet, he had no idea exactly how it works. He didn't understand that it can't skip a generation. You need to have a positive gene in order to pass it on.” – P16, HASA member (Interview 6)

Participant 16 further explained that another frequently seen misconception involves the autosomal dominant inheritance pattern of the condition. People sometimes misunderstand that each child has his or her own risk of 50 percent of inheriting the condition and that not half of the children might inherit the condition.

“Another misconception people have [is] ‘oh you got a 50 percent chance’, so immediately they do the math; ‘I've got eight children so four children, no more than four children will get it.’ They don't understand that each person, each of the eight persons has a 50 percent chance. So all eight of them can get that, that 50 percent of the positive, the 50 percent chance and they'll have the positive gene.” – P16, HASA member (Interview 6)

Further misunderstanding regarding pattern of inheritance is seen when people assume an autosomal recessive pattern of inheritance where both parents need to be carriers in order for

a child to be affected. Participant 16 explained that it would be beneficial if all HASA members could understand the way HD is inherited:

“Just all those misconceptions on how it works and that it's not the same like, ehm, not all, it's not the same like maybe another disease. Ehm, it's not the same like cystic fibrosis where both people must have the gene, kind of a thing. Say you, if that can be cleared up with everybody, ehm, that will be a good thing.” – P16, HASA member (Interview 6)

Participant 16 further explains that there is danger in members understanding only part of the genetic information and that education and clarification of misconceptions are consequently of extreme importance:

“Sometimes a little knowledge is more dangerous than no knowledge at all.” – P16, HASA member (Interview 6)

Participant 10 explains that she feels content in her role as information provider to help people understand when they are unsure if they understand the information correctly:

“Being a person not to counsel, but perhaps to just provide that information where people are unsure about certain things.” – P10, counsellor (FG1)

Voice of caution and realism

The fact that HD is currently untreatable is devastating. It is thus understandable that people become elated when there seems to be medical breakthroughs regarding treatment of the condition. Participant 1 explained that HASA members get very excited when they hear about new treatment options and clinical trials becoming available. Although she understood their excitement and the importance of having hope, she felt that one of her roles was to provide a measure of realism, a voice of caution, to the group. She often warned members that even though there is progress in finding a cure, medical technology and -research might not be there yet:

“We also probably provide a voice of caution in terms of all the, all the studies that are being done, the clinical trials because they really, really get very excited about all the clinical trials and we've all and sometimes we feel a bit bad about it, but sometimes we have to say that this may not be everything that you hope it's going to be.” – P1, counsellor (Interview 1)

Participant 1 also explained that she sometimes provides a measure of medical realism to patients who make use of alternative measures to remind them of the actual medical science behind the condition:

“Because and sometimes we even have to act as a little bit of a, a break, because we get sometimes get people, this sound weird, from, from the ahm alternative side of things. And then they have their own ideas and they kind of, they get caught up in this sort of thing and they forget the, the actual medical thing behind what is going on.” - P1, counsellor (Interview 1)

Participant 1 explained that she also felt that due to her medical knowledge she needs to intervene when she feels patients are doing things (such as using medical cannabis) that may not be good for them and rather direct them to proper medical resources.

“Also the person who’s got the medical knowledge and so we can stop them from doing things that may not be good for them. Uhm stop them from going off in the wrong tangent if necessary.” - P1, counsellor (Interview 1)

Information as therapeutic tool

In addition to information being required to understand the condition and its implications, the use of information as therapeutic tool was mentioned several times during the interviews. Participant 10 hypothesised that information could possibly help people deal better with the diagnosis of HD.

“But perhaps... that information helps you to understand and perhaps cope better.” - P10, counsellor (Focus group 1)

Participant 7 had a similar viewpoint as he described that for him receiving information is synonym to receiving therapy and that he feels the line between the two concepts could be blurred.

“I think the information giving is therapy. Maybe the lines between the two are blurred in a good way, I think.” – P7, HASA member (FG1)

Participant 5 elaborated on this concept by describing that we find ourselves living in the information age where people have become accustomed to using information as a way of moving forward. By having one piece of information it allows one to focus on the next thing, and the next, and thus moving forward in the process.

“We live in an information age, having information, it’s quite a need it helps you get on, like you focus on the next thing, because you have this information.” – P5, HASA member (FG1)

Participant 12 explained that part of the therapeutic benefit of having information lies within the saying that knowledge is power. She explained that having information leads to empowerment and understanding:

“The more knowledge you have, you know knowledge is power, you know the more knowledge you have, the more understanding you have.” – P12, HASA member (FG2)

Another example of information as therapeutic tool is seen in members using the gathering of information as coping mechanism. Participant 3 described how she spent months reading up information online to understand and deal with the condition diagnosed in her family:

“In the first few months, I was, I’m an information seeker, that’s how I deal, I spent hours, hours on line.” – P3, HASA member (FG1)

She explained that she would have benefitted greatly if there was a counsellor at the HASA SG who could have helped her make sense of the information she read.

“I would have given anything to have somebody to sit me down and say ‘this is what it means’... and had there been an option in this group and had the group known that there is, ‘oh we’ve got genetic counsellors come to us sometimes, we can put you in contact with them’, I, I would have taken that opportunity and done it.” – P3, HASA member (FG1)

What information is required?

The information that HASA members require from counsellors are the typical information that would be provided in the traditional genetic counselling setting. The type of information includes information to help newcomers orientate themselves in a newly-found world of HD, understand the condition and know about the resources available to them.

Mmm... To be honest, I feel like I’ve, I’ve, mmm... Ja... I suppose it’s just those discussions around risk or family testing and, which is obviously a really sensitive topic, that I feel that that’s... ja, really important to have someone there just to help to answer the right type of questions and ask the right type of questions around that. – P17, psychologist (Interview 9)

Genetic services available

Many of the individuals who attend the HASA SG are family members of affected people and have not necessarily received genetic counselling themselves. Not all HASA members are aware of the services available to HD families and there is consequently a need for a such information. Participant 2 explained that a lack of knowledge regarding services was especially seen in the early days of the SG's existence:

"Not everyone knew about the genetic services at Groote Schuur, not everyone knew what predictive testing was about, not everyone knew how that happened, uhm the processes."
- P2, counsellor (Interview 2)

As members became aware of services available to them, they would ask the counsellors at the SG to clarify some information about the clinic to them:

"They would... come to asking... about clinic, so there's a lot of practical questions like how to be seen at the clinic, which doctor to be seen if they need a referral." - P11, counsellor (Interview 4)

Genetic aspects of the condition

In addition to popular questions regarding available services, another important form of information needed involves the genetic aspects of HD. As described in the literature review, genetic information regarding HD is complex and it is common for people to not understand it. Participant 10 explained that she experienced first-hand in the SG that some people did not understand genetic information:

"There were questions that came up about people saying that they didn't really understand the genetics." – P10, counsellor (Interview 4)

The genetic aspects of the condition that needed explanation ranged from more basic concepts such as inheritance pattern to complex genetic detail. Participant 10 explained receiving basic questions on inheritance and implications for family members:

"How was the [condition] inherited, what it means for their family, common misconceptions about the condition and then, you know that kind of thing." – P10, counsellor (Interview 4)

Participant 2 explained she came across a need for information on the less commonly seen juvenile onset of HD:

"Not everyone knew about juvenile onset Huntington's." - P2, counsellor (Interview 2)

More complex information required on the genetic aspects of HD was reported as genetic anticipation, CAG repeat categories, difference in AOO and phenotypic variability as explained by Participant 10:

“They didn't understand that the whole, you know, the concept of genetic anticipation and what it means to have more CAG repeats and does it mean that it's going to be an early age of onset or a more severe phenotype.” – P10, counsellor (Interview 4)

Participant 16 also described the need for understanding CAG repeats and its relation to AOO in creative lay-man's language:

“Also, you know, about the, how long the stutter is; I call it a stutter, I don't know what you call it, you know, the repeats, how the repeats is, and also people think it maybe started in the 30s, so everyone else will start in their 30s.” – P16, HASA member (Interview 6)

Testing process

Information on testing procedures were explained to be another need of HASA members. Participant 11 experienced that it was often the new members who enquired about testing:

“A lot of the time there would be new people that would come and ask questions about... how they go about testing.” - P11, counsellor (Interview 4)

Information on the two types of testing procedures were also needed when members expressed their dissatisfaction when a symptomatic individual were not offered the predictive testing process. Participant 1 expressed the challenge for members to accept when a diagnostic test is done instead of following the PT process:

“They actually asked me about, they said that they felt they hadn't, we don't counsel people that we perhaps have got a clinical diagnosis properly they feel like that we just throw it at them and we should rather give them a counselling session like we do with predictive test. And to explain why that's not always possible was quite an interesting thing for them to understand why we don't necessarily take those sort of, because they, they are affected already we don't do the whole predictive testing.” – P1, counsellor (Interview 1)

Research and medical developments

As mentioned before there is currently no cure for HD and people are anxiously awaiting a miracle-breakthrough in treating the disease. Participant 5 expressed that he would like to receive medicinal and treatment information:

“In the future you can also come and counsel us on a, on the future of genetic counselling of Huntington’s and where the treatment’s going, and what the Americans are doing and what the British are doing, and what the South Koreans are doing, what the Japanese are doing and what the Chinese are doing and how it comes together to improve the medicine in this country.” – P5, HASA member (FG1)

Participant 13, on the other hand, did not feel that it is the counsellor’s responsibility to be on top of the medicinal developments:

“I think the biggest knowledge we, we really all need is the medication, you know the updating on the medication, but that’s not your [role].” – P13, HASA member (FG2)

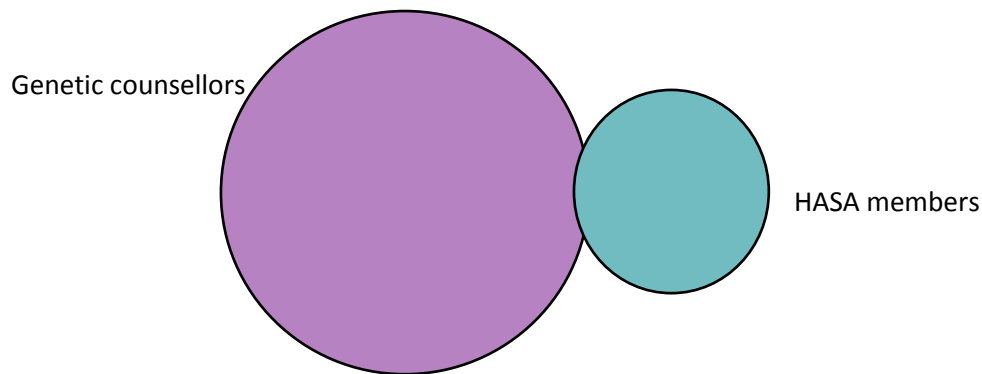
Participant 1 explained that though some members would like to receive medical information from counsellors, they tend to direct their questions toward the neurologist when he attends the group:

“Anything that’s related to the medical condition they ask us but we do find when the neurologist come in that uhm, they defer to them so they may whatever they say is more important to what, what we’ve got to say.” – P1, counsellor (Interview 1)

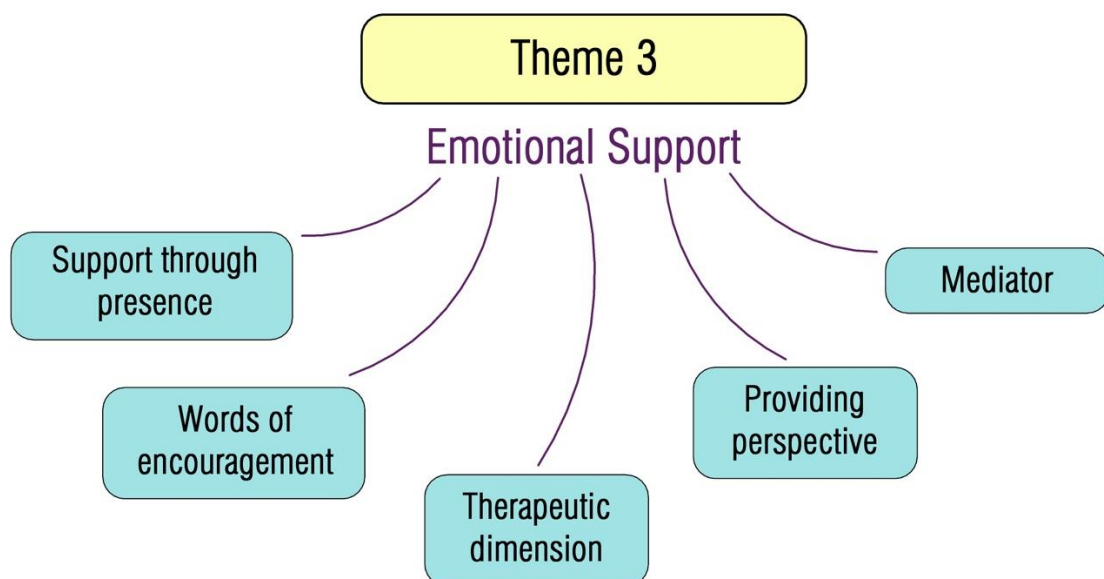
As can be seen from the data above, a prominent role of a genetic counsellor in the HASA SG was expressed to be that of information provider. HASA members primarily see a genetic counsellor as someone who can provide them with much needed information on their condition including detailed genetic explanations, family risks, information on support services etc. In fact, it was described that due to their expertise and knowledge of the subject matter, no-one else is perceived to be more suited to provide the SG with information and clarification on genetic matters as they arise during SG meetings.

3.5 THEME 3: EMOTIONAL SUPPORT

“To provide support, that really is the goal of this support group. I couldn't offer up my own experiences, and I couldn't offer that type of support, but to be able to be like, ‘you're doing a great job’ [was good].” – P10, counsellor (Interview 4)



Theme 3 represents the opinions of genetic counsellors most strongly, followed by those of the HASA members. There was a lack of input from the psychologist regarding this theme.



The data revealed multiple times that the presence of a counsellor provides an element of emotional support within the HASA SG. Many opinions were expressed on the matter and are described under the following sub-themes:

- Support through presence
- Words of encouragement
- Therapeutic stance
- Providing perspective
- Mediator/advocate

Support through presence

The presence of a counsellor in a SG meeting was described as a way of providing “silent support”. Participation in SG meetings are voluntary for counsellors, so the knowledge that personal time is being sacrificed for the benefit of the group conveys a message of caring. Without having to say a word in a SG meeting the mere fact that a counsellor is present shows support for the members of the group. Participant 10 explained that she hoped her presence portrayed the message of care and support even in times when she was just sitting silently in the group:

“But just to know that we are there if they need us and if they don't need us, that we will just be listening.” – P10, counsellor (Interview 4)

Participant 11 explained that through her presence she wanted patients to know that they are not alone in this journey and that there are professionals who care enough to extend their help beyond the clinical sphere:

“To just have a supportive role, just for them to know that they're not alone, that there are people out there who care enough even if they're not directly impacted by the disease themselves.” – P11, counsellor (Interview 4)

Even within an atmosphere of silence, the presence of a counsellor can aid in creating a safe, supportive space for group members. Participant 1 described how she experienced herself as being an emotional “safety net” for the group – someone who cares, but who is a bit removed from the situation and not entwined with family dynamics:

“I think there is sort of a feeling of safeness that we create when we're in the group. That there's somebody beyond just the affected people and their families there apart from the

psychologist that's there I think we do create a bit of a, of a safety net for them as well.” – P1, counsellor (Interview 1)

Words of encouragement

Emotional support by means of encouragement was reported as one of the roles of a counsellor. Counsellors are in the unique position to provide an encouraging word to HASA members since they know about the HD struggles their patients have expressed at clinic. Participant 10 described that even though she could not offer the group a personal experience with HD, she could offer an encouraging word that hopefully could allow someone to take a step forward:

“I couldn't offer up my own experiences, and I couldn't offer that type of support, but to be able to be like, ‘you're doing a great job’ because any person off the street could see that that person is doing a great job.” – P10, counsellor (Interview 4)

In addition to providing direct words of encouragement, sometimes patients can benefit from a mere acknowledgement of their feelings. Participant 10 described the important role of acknowledging the difficult decisions there are to be made with HD in the family and that sometimes acknowledgement can be very helpful in providing emotional support:

“I think just in terms of, like, being able to provide some [additional] emotional support, [such as] ‘the fact that you don't want to know whether you're HD positive or not, it's okay’.” – P10, counsellor (Interview 4)

Therapeutic dimension

A SG facilitator is in the ideal position to enable dialogue between people and working with relational dynamics that are expressed within a group. A counsellor can help manage the communication as they are trained to navigate interpersonal interaction. The role of facilitating and enabling dialogue a role can be interpreted as adding a “therapeutic dimension” to the working of a SG:

“Because we do understand these patients I think they feel a little bit better when we are, when we are taking that role of, of kind of trying to hold a group a little bit more.” – P1, counsellor (Interview 1)

“The holding, the holding, role, you know where you are looking after the group as a whole and saying to them... from the point of view of knowledge we’ve got and, and the ability to work with people that we’ve got, I think it’s a, it is a valuable place to be.” – P1, counsellor (Interview 1)

“So very often before they’d come they would be very overwhelmed, uhm very upset about certain situations so that type of thing, so ja definitely the role of like psychosocial support, helping people to deal with those emotions and ja understand them a little bit better, was definitely a strong role.” – P11, counsellor (Interview 4)

The role of a counsellor as bringing a therapeutic dimension to the group was acknowledged in more than half of the data collection events. Participants 1 and 6 who both spent three years involved with HASA CT most strongly advocated for the therapeutic role of a counsellor in a SG. They both described incidents where they acted in a therapeutic capacity within the SG.

“[There] have been times when I’ve had to take on a therapeutic role when people have got upset about for instance what goes on in the clinic.” – P1, counsellor (Interview 1)

“Uhm, once or twice I had to explore that with them and help them to, to kind of understand why things are the way they are and also just sometimes picking up on, on emotional issues that have come up and be and to sort of pin point maybe this is going on or and helping them to, to work through that.” – P1, counsellor (Interview 1)

Participant 10 explained that she has seen the benefit of therapeutic intervention from a counsellor, but she feels that it is not the most important role for a counsellor as interaction with fellow SG members serves as a type of therapy by itself.

“There’s a lot to say out there about the fact that the therapeutic, just the being around people that are in similar situations. And, and that process of sharing information amongst people in some of those situations can be very therapeutic for you.” – P10, counsellor (Interview 4)

“There have been situations where they [genetic counsellors] have, I guess steered a conversation and help people to think differently, so that’s been their role. But there hasn’t been, I don’t know, from my experience there hasn’t been a lot of a need for therapeutic support because they tend to get that kind of support from the other people.” – P10, counsellor (Interview 4)

Participant 10 elaborates that a counsellor could play a therapeutic role, but that it depends on the relationship the counsellor has with the group.

“I do think there might be an avenue for that [therapeutic role], particularly if a genetic counsellor is more in a facilitator role or or has quite a good relationship with the people, if they’re constantly going and they have a good relationship with the people that are there to, and have establish some sort of like presence within the group and maybe just start trying to challenge people and trying to help in a more therapeutic way could be beneficial.”
– P10, counsellor (Interview 4)

Participant 11 described how the creation of the “split-group” concept created the space for more therapeutic involvement to take place:

“It was really, really powerful because a lot of the carers or the family members were able to talk and discuss things that they maybe couldn’t discuss with their family member in the room or in the support group. So, feelings of frustration and all the behaviours that they thought were unusual but obviously talking amongst other carers and other family members realising that it was actually very normal. Uhm and then as a genetic counsellor, also stepping in and saying those are normal traits, this is normal things that go on. So yeah, definitely I agree that the role of psychosocial support, those skills are very critical as well in a support group especially when you have opportunity to use them like when the groups are split-up, uhm that type of thing.” – P11, counsellor (Interview 4)

Providing perspective

Three participants described how they viewed a counsellor as being able to provide perspective in the group. Participant 8 asked that the counsellor provide them with perspective by advising them on how to think positive:

“How to think positive. We know it’s all negative, but how, how can we put the negative into a positive. How can we deal with it and just give up or just say there’s no more hope, there’s no more point [unclear words], so how do we deal with it, how do we subside, and how do we approach it, because sometimes you can’t fight fire with fire or, how do you control your emotions if anything happens, let’s put it that way.” – P8, HASA member (FG1)

Mediator

There are times that counsellors act as mediators within the group such as managing conflict between group members and providing insight into the situation.

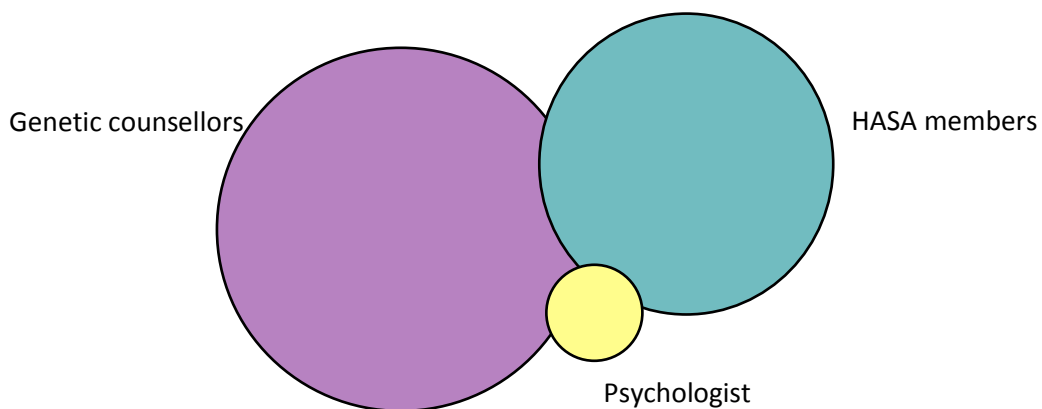
“Occasionally when people are just losing it a bit with each other or something I’ve been able to say, hang on maybe this is what’s going in in your life.” – P1, counsellor (Interview 1)

“There has been certainly times where people get so angry with UCT and GSH and then well this is why I like to understand the people a little bit better.” – P1, counsellor (Interview 1)

As can be seen from the data above, another prominent role of a genetic counsellor in the HASA SG was expressed to be that of emotional supporter. There are multiple ways in which genetic counsellors fulfil this role including providing words of encouragement, acting as mediator in the group, providing members with perspective to simply showing support by their mere presence.

3.6 THEME 4: PRACTICAL HELPER

“Just little practical things... genuine practical advice that could help you, ehm, it is be so invaluable.” – P16, HASA member (Interview 6)



Theme 4 represents the opinions of genetic counsellors most strongly, followed by those of the HASA members and a small, yet significant contribution from the psychologist.

Theme 4

Practical helper

Connection between support group and clinic

- Face of the clinic
- Advice on how the clinic works
- Administrative duties
- Mediator between patient and clinic
- Access to genetic counsellor
- Referring patients to HASA
- Referring patients to other services

Helping in support group

- Facilitation
- Help run support group
- Fundraising and extras

The first two themes describe counsellors in roles that they traditionally play in the clinic albeit slightly differently due to the more public setting of the SG. It is therefore anticipated that SG members see a genetic counsellor as someone who provides information and emotional support. In theme 3, however, the role of a genetic counsellor as practical helper introduces a “new dimension” of involvement as seen in a SG.

The nature of HD as condition is overwhelming and infiltrates several aspects of the individual and family life. Participants described that due to the emotionally and physically engulfing nature of HD, it is very useful to receive help with simple, practical things. Participant 16 explained that small, practical things can make a huge difference for someone who is overwhelmed by the impact of HD:

“Sometimes that practical help is also so invaluable because people are so overwhelmed with, ehm, having this condition or having a family member have the condition, that everything becomes so much harder.” – P16, HASA member (Interview 6)

This “practical help” that a genetic counsellor can offer are discussed under the following two sub-themes:

- Connection between support group and clinic
- Helping in support group

Connection between support group and clinic

The genetic counsellors who attend the SG are often still in training at UCT and form part of the clinical team who sees patients at the neurogenetics clinic. When these counsellors get involved with HASA an assumed connection between the clinic and HASA comes into existence. Participant 11 explained that when she answered questions about the clinic, doctors and referral, it was like a bridge being formed between the patients and the clinic:

“They would... come to asking certain questions... about clinic, so there’s a lot of practical questions like how to be seen at the clinic, which doctor to be seen if they need a referral, so it was almost like a bridge between patients and clinic to kind of like help them with anything that’s needed to be done.” - P11, counsellor (Interview 4)

Face of the clinic

The genetic counsellor unintentionally becomes the “face” of the genetics clinic. Participant 10 explained that if she was in the patients’ shoes it would have been great for her to see a familiar face at the SG:

“When I put myself in their shoes, it’s quite great to be able to see someone that you may recognise as someone from a place that you often go to, you know like from genetics there, and being interested in getting involved. I think it really does change their perception somewhat.” – P10, counsellor (Interview 4)

Participant 7 agreed with this statement. He explained that being familiar with a genetic counsellor at HASA makes it less intimidating for family members to reach out to the clinic. The genetic counsellor “humanises” the clinic when a patient can put a “face” to it. This “familiarity” helps others reach out to the clinic for their own questions and predictive testing without feeling so intimidated:

“Or a family member wants to find out more, but they’re kind of intimidated to go knock on the door at Groote Schuur, but they’re here because the support group, oh well these people I’ve seen you, much easier.” – P7, HASA member (FG1)

Advice on how clinic works

This sub-theme links with theme one that describes a genetic counsellor at a SG as information provider where it was found amongst other things that HASA members like to receive information on how the clinic works. Participant 1 explained that she found it useful on the part of the HASA members when she spoke about the clinic and how it operated. It gave

the members the opportunity to ask questions and make sense of the workings of the clinic for themselves:

“Uhm and it was really useful for them when we could speak about the clinic and stuff here and tell them how that ran.” – P1, counsellor (Interview 1)

Administrative duties

It was explained that a condition such as HD makes everything harder – even a small day to day task may feel like a mountain to conquer. Participant 16 explained that it is very helpful when a genetic counsellor at HASA assists patients administratively in assisting them to make appointments at the clinic:

“[It] seems like such a small thing, ehm, that must be done... but you cannot imagine what a big thing it is for somebody who’s so overwhelmed and how much, how much help that is to the person to know that, ehm, ehm, they don’t have to jump through so many hoops to get, to get an appointment.” – P16, HASA member (Interview 6)

Another administrative role a genetic counsellor can play, is helping HASA members with paperwork. Participant 2 reported that as genetic counsellor she had provided practical help to members in the past by filling out forms for financial assistance and facilitating HASA members in attaining Medic Alert bracelets:

“[I] have been filling in their, their forms for financial assistance for some of our patients that needed some financial assistance uhm as well as sometimes talking about Medical Alert bracelets.” – Participant 2, counsellor (Interview 2)

Mediator between patient and clinic

Patients and family members have experienced some frustration with the clinic and the system by which it operates in the past. Participants described that a genetic counsellor at HASA is an ideal person to act as mediator between the group and the clinic. With the permission of the group, a counsellor can give feedback to the clinical team and in so doing help resolve problems or provide better patient care. It can serve as a two-way channel where HASA members can send suggestions to the clinic and the clinic can inform HASA members of any changes etc.

“Sometimes they had feedback that they also wanted us to feed back to the clinic if it was to do with scheduling or appointments or whatever the case might be.” – P11, counsellor (Interview 4)

“Often we would bring whatever they had discussed at the support group, maybe issues associated with the clinic, and we would bring that sometimes to our clinical meetings to discuss some issues that they had.” – P11, counsellor (Interview 4)

Access to a genetic counsellor

One of the great benefits that HASA members have when a genetic counsellor attends the clinic is access to a genetic counsellor. Certain questions can be answered by the counsellor which could possibly reduce/void/solve the need for the patient to visit the clinic or a private genetic counsellor.

“‘Cause I also sometimes feel like people's access to [genetic counselling] are [you. always getting access to a genetic counsellor, ‘cause then it is nice to have, you know, I suppose the support group in which case it's all volunteer based, so you're lucky to get it, but if it's not easily accessible I think it's amazing for people to have.

Participant 7 insinuated that the genetic counselling they received at the clinic helped him to deal with HD in his family. It seemingly created a stable level of emotional support. Once him and his family were told they did not have to receive additional counselling he was shocked and disappointed. He explained that having a genetic counsellor at the SG is valuable as it can act as a “substitute” for not receiving counselling at GSH anymore:

“In fact it's also nice, because otherwise we only get to go every 6 months to Groote Schuur and the last time they said so well we don't think you kind of need to keep coming, and I was like what, really?” – P7, HASA member (FG1)

Referring patients to HASA

Three participants mentioned that the counsellor can refer patients they see at clinic to the SG for the benefit of the individual, but also to help grow the HASA community. Participant 2 explained that when the group was just started, the genetics team helped build it by referring patients and family members to HASA:

“We helped to recruit members to the group, uhm I suppose that is a need that was outside of the actual group setting uhm, you know informing people about the support group giving out their contact information uhm and that was a need that uhm, I don’t think ya, I don’t think was explicitly expressed at the group we built.” – P2, counsellor (Interview 2)

From a HASA member’s point of view, Participant 3 explained that when a counsellor refers a patient to a group that she has first-hand experience with, it adds a “personal element” to the patient-counsellor relationship:

“Then there’s a personal element to the person that’s dealing with you, they’ve actually been there, so they know. I think it will be different to you if you’re speaking to somebody... the fact that you’ve come to these meetings already, to then meet someone with HD, you’re like ah, there’s a support group meeting that I’ve attended in Claremont, whoow [laugh].” – P3, HASA member (FG1)

This links with Participant 10’s opinion that as a counsellor she prefers to visit a SG before she refers her patients to it. By familiarising herself with a group, she is able to refer her patients, knowing personally how the group operates and what benefit it can provide to her patients:

“I feel like I can’t always refer my own patients to support groups without knowing. You know, I now have first-hand experience of what’s being done and how the support groups can help, how they function actually, to be able to make that kind of recommendation.” – P10, counsellor

Referring patients to other services

Not only can counsellors refer patients to HASA, but they are in the ideal position to refer patients to other services in a wholistic approach to deal with HD. When a counsellor attends a SG, they learn first-hand about the needs, possibilities and potential solutions of a variety of patients. This information can be used to direct patients that a counsellor sees in clinic to reach out to similar resources whether they attend HASA or not:

“Because there is this broad and holistic view and we have to work within teams, I can say maybe you should go and see this sort of person or we see that this has helped with some patients” – P1, counsellor (Interview 1)

Participant 11 explained that it is valuable to know first-hand what support patients need in order to refer them to appropriate sources and service providers. She explains that sometimes patients benefit most from services that can help them in a practical manner:

“Sometimes they don’t just want to come and get all this medical information; it’s sometimes the practical things like be referred to a dietician to help them to not choke or to eat properly, uhm ja, basic things like that; just being aware of what they go through and what support they might need or what referrals we might need to make, that type of thing.”
– P11, counsellor (Interview 4)

Helping out in support group

This sub-theme explains how participants viewed the way in which a genetic counsellor can help out practically in the running of the SG.

Facilitation

Currently the HASA SG is facilitated by two psychologists who have been involved for a couple of years each. As therapists they have the knowledge and experience to lead a group in their discussions and communication. The topic was raised several times that a genetic counsellor is also an ideal individual to facilitate such a SG due to their unique skillsets. Genetic counsellors are trained to manage and facilitate discussion among people since patients often bring loved-ones to their counselling sessions. Participant 11 explained that she feels group facilitation is an important role that a genetic counsellor can play in a SG:

“I think just from how genetic counsellors are trained, we are trained to facilitate conversation even if it’s just like for example in a consultation when you have a family, you have to kind of facilitate the talking and not allow for arguments, for one to dominate it over the other, facilitate those skills, to manage people and to facilitate discussion and communication and to ask the right questions, to get people to open up and really explore issues. I think from that perspective there is a great, great room for genetic counsellors to be the facilitator of a group, bring up issues, help establish communication within the group, make sure one person isn’t dominating the conversation, and if there’s any conflict that is just managed appropriately. So I think facilitation is quite a big one.” – P11, counsellor (Interview 4)

Not only do counsellors feel that they are suited to facilitate the group, but two of the HASA members brought the topic up as well:

“I think you’re perfectly capable of being, doing that role, if you needed to.” – P12, HASA member (FG2)

Participant 12 further explained that even if there is a dedicated facilitator in the group, the genetic counsellor can act as substitute when the facilitator cannot attend a meeting:

“I recon if [the facilitator] couldn’t be there they could say would you mind being the facilitator. I don’t think [the group] would mind at all.” – P12, HASA member (FG2)

Help run SG

In addition to fulfilling a facilitator’s role, a counsellor can also help out practically within the group such as helping with the set-up of chairs and refreshments. Counsellors can also play the very useful role of taking meeting minutes in the group

“The role of a genetic counsellor can also be very practical. So sometimes we would like take minutes for the group or write down main points, that type of thing. So ja in that way we were helping with the running of the support group.” – P11, counsellor (Interview 4)

Fundraising and extras

Participants 12 and 13 commented that the flexibility in the role of a counsellor at a SG allows for the counsellor to step up and fulfil a role that opens up:

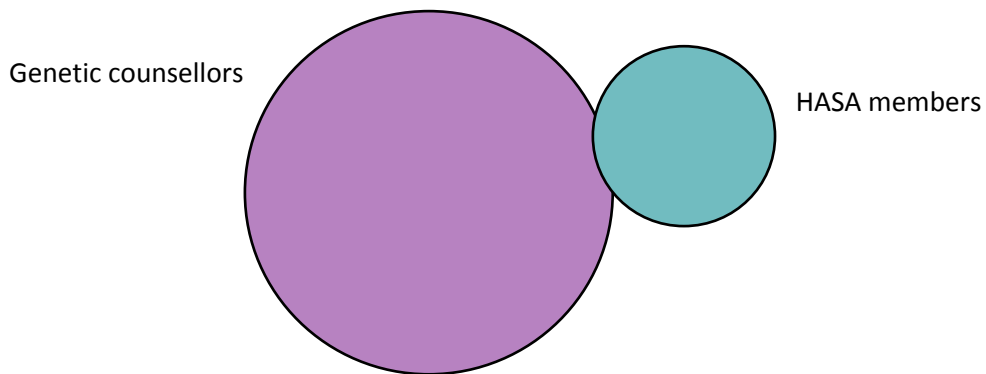
“Flexibility, you know. I think find the flexibility in that role.” – P13, HASA member (FG2)

Participant 12 further explains that except for providing genetic information and support, there is no expectation of what a counsellor specifically has to do. The counsellor, is however, welcome to bring ideas for involvement to the group that the counsellor feels strongly about. For example, if the counsellor is passionate about fund raising she is welcome to do so for the group:

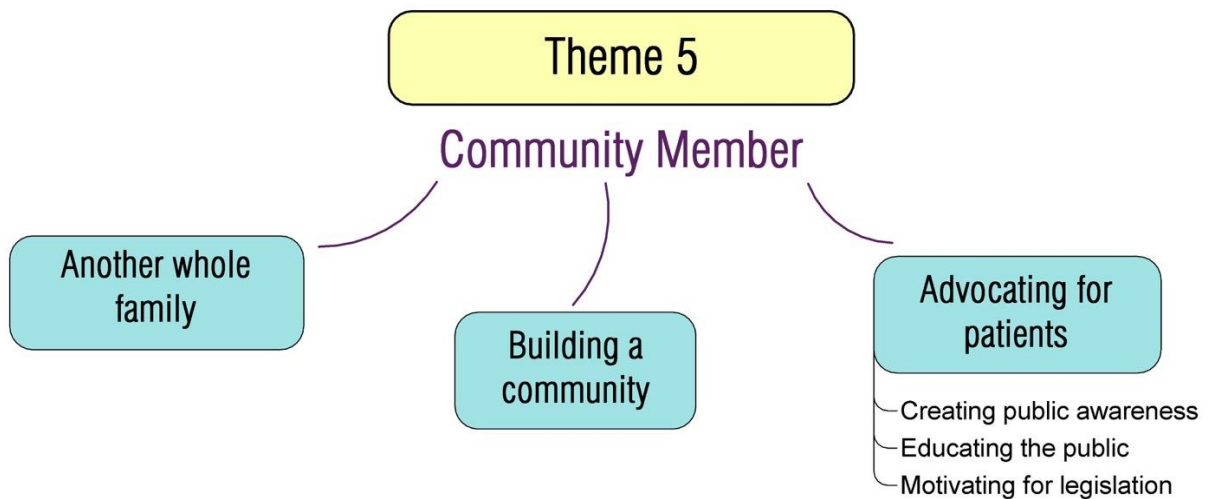
“We don’t expect you to do anything specific unless you say I really would like to do that...I really would like to do a fundraiser. This is my idea.” – P12, HASA member (FG2)

3.7 THEME 5: COMMUNITY MEMBER

“It’s like a whole new friendship circle... it’s like another whole family you know... it does feel like a second family.” – P13, HASA member (Focus group 2)



Theme 5 represents the opinions of genetic counsellors most strongly, followed by those of the HASA members. There was no input from the psychologist regarding this theme.



A less repeated, yet significant theme was raised by participants as they described a sense of community that they experience being part of the HASA SG. By becoming part of the group, people become part of the HASA community - a space of safety and communality, for people with the common factor of HD in their lives. Many of the HASA members have been attending

for years and strong relationships have formed over time. When a genetic counsellor becomes part of the HASA SG, she becomes part of this community.

Participant 10 explained that it is important for her to be regarded as part of the HASA community and not merely as a genetic counsellor. She describes that as a human being she sometimes just wants to be part of the group – supporting the community by her mere presence and genuine regard for people’s wellbeing:

“Sometimes I can just be there as... anyone else that’s going to sit there, hear their stories, learn from them and provide emotional support. I don’t want to go there and be regarded as just a genetic counsellor.” – P10, counsellor (Interview 4)

This sense of authenticity was present in all the counsellors who were interviewed as many of them sincerely expressed their care for the HD community.

The theme of a genetic counsellor as community member is further described under the following subthemes:

- Another whole family
- Building a community
- Advocating for patients

Another whole family

Being part of the HASA community has been described as having a new friendship circle and even having another family. Participant 12 described that she found friendship within the HASA community that has been a great source of support to her over the years:

“It’s another whole friendship circle we’ve managed to get uhm. I look forward to it every month, I must admit.” – P12, HASA member (FG2)

Participant 13 agreed with Participant 12’s statement and elaborated that for her the HASA community has surpassed mere friendship; to her it has become her family. She further acknowledged the good of having genetic counsellors as part of this friendship, this family:

“It was like [Participant 12] was saying, you know, it’s like a whole new friendship circle. But to me it’s like another whole family you know like ja, it does feel like a second family, I agree with [Participant 12] with that, you know. A genetic counsellor, ja, it’s good.” – P13, HASA member (FG2)

Building a community

A significant role of counsellor as community member lies within the counsellor helping to build the HASA community. As mentioned in theme 3, a genetic counsellor is in an ideal position to refer patients seen at clinic to visit the HASA SG. Participant 10 expressed that by referring patients to the group, she is not only helping the individual, but she is also helping to build the HASA community. She explained that for her it is important to help the HASA community grow to help sustain the group for the benefit of many.

“it’s important because if people stopped attending, you know the support group falls apart.” - P10, counsellor (Interview 4)

Participant 10 elaborates that it is good for the group’s growth if others hear about it and see that it is being supported and recommended by genetic counsellors – in a way validating the benefit of the group:

“But it is a good way to build up... people seeing that [the support group] is being supported.” - P10, counsellor (Interview 4)

Advocating for patients

An important role of a counsellor as community member, is acting in the best interest of the HD community. The condition is rare, and patients are often confronted with stigmatisation and ignorance from the public. A professional, such as a genetic counsellor, who works with HD is in the ideal position to advocate for HD patients. Participant 11 explains the importance of advocating on behalf of the HD community:

“There’s a huge need especially with like a condition such as Huntington’s that is much more rare, very stigmatised, and people can often make judgment calls on a person just based on their symptoms assuming that something else like being drunk or disorderly or whatever the case may be.” – P11, counsellor (Interview 4)

The different ways in which a genetic counsellor can help advocate for the HD community have been described below.

Creating public awareness

One of the ways in which counsellors can help advocate for patients is by helping to create public awareness of HD. Participant 11 explained that during her three-year involvement with HASA she experienced ample opportunity for the counsellors involved with HASA to help create awareness by doing public talks and a radio interview.

“A lot of the time when there were events they would ask me to do a talk to basically just discuss what the condition was; the medical symptoms and the genetics behind it. So I think like a role, my role was [to be] an advocate between the support group and the [public].” – P11, counsellor (Interview 4)

Participant 11 also explained that their awareness creation stretched further and included organising different events such as charity events to raise funds for, and bring attention to, the group:

“We organised like different events, charity events to raise money.” – P11, counsellor (Interview 4)

Educating the public

Another way that counsellors can advocate for their patients is by educating the public and ties in with the concept of creating public awareness. Participant 11 explained that those close to people with HD knows about the condition, but that the general public does not know about this rare disease and educating the public can be greatly beneficial.

“As I got more involved, they asked a lot about, in terms of education for the community; so they discussed a lot of the time about how they understood and their family members understood, but that the community just was not aware, they didn't understand what the condition was.” – P11, counsellor (Interview 4)

Participant 10 and 11 both explained that looking for opportunities to educate the public is a good idea to help people understand the condition.

“If someone brought up doing a presentation for [the community] and perhaps doing, going about trying to make sure that people understand what Huntington's disease is. How was the inherited, what it means for their family, common misconceptions about the condition?” – P10, counsellor (Interview 4)

Motivating for legislation

Participant 11 also explained how HASA had been wanting to create a public facility where people in the end-stage of the disease can receive palliative care. When the opportunity arises, counsellors are in the ideal position to motivate for legislation that acknowledges the needs of people and families with HD.

“To be a patient advocate, so for example if there were any policies or things that they were trying to get past, eh, through the government or ministry of health, uhm obviously to be an advocate for them to try, from a medical perspective to try and motivate for like funding or care facilities, that type of thing.” – P11, counsellor (Interview 4)

As can be seen from the discussion above, the genetic counsellor was found to not only be seen as a medical professional, but a member of the HASA community. By fulfilling the role of community members counsellors are seen as being part of an adopted the HASA family that comes with shared burden, friendship and often lasting relationships. By being part of this community genetic counsellors have the opportunity to advocate on behalf of HASA members by raising awareness of the condition, addressing stigma and misconceptions, and speaking at public and community events. In fact, the role of genetic counsellor as community member is seen as a pivotal/significant asset to the prosperity and longevity of the group.

Chapter Four

Conclusions and recommendations

4.1 INTRODUCTION

In this chapter conclusions are drawn based on the findings of the study and previous literature regarding the involvement of genetic counsellors with support groups (SGs). In addition, strengths and limitations of the study are discussed, practical implications are suggested, and recommendations for future research are made.

4.2 CONCLUSIONS

The study aimed to determine what role(s) a genetic counsellor can play in a SG for HD by exploring the thoughts and opinions of members of a HASA SG. A limited number of existing local and international studies have examined the role of a counsellor in a genetic SG. These studies report broadly on the matter and point out basic roles and responsibilities that can be associated with such involvement. The literature, however, lacks individualised perspective that qualitative data typically provides.

The study found that not only are there numerous roles for a genetic counsellor to play in a HASA SG, but that the relationship that results from such a collaboration can be mutually beneficial to both the counsellor and the group. It was found that HASA members not only disregard ethical concerns about professionals impinging upon their privacy, but that they feel a counsellor's involvement "is like a breath of fresh air" to the group (P11 quoting a HASA member).

The various ways in which genetic counsellors can be of value to a HASA SG were found to be information provider, provider of emotional support, practical helper and community member. The data also stresses the counsellor's potential gain in learning first-hand about patients' needs and wants, increased insight into their "real lives" and getting exposure in the HD community for possible future private counselling referrals.

The data provided ample proof that there is more than one way in which a counsellor can be involved with HASA. The manner in which this involvement is "packaged" was, however, seen

through multiple lenses. Some participants favoured long-term involvement with one or two specific counsellors while others felt such commitment could not be expected from a volunteer and expressed that they would be content with counsellors taking turns in visiting. Additional vantage points regarded informal involvement with a counsellor “merging into the group” versus set structured involvement.

The data revealed that preference for counsellors’ type of involvement is dependent on multiple factors including the outlook of the key role players, the perspectives of group members, and the time and place the group finds itself in. It is also dependent on the group’s needs at a certain point in time and the logistics of how the group functions.

The analysis of P15’s transcript provided insight into how different-functioning SGs have different needs even if the underlying genetic condition is the same. P15 is the only participant associated with HASA GP and the data revealed through her transcript was remarkably different to that of the counsellors involved in HASA CT. A decision was made to exclude P15’s data from the main discussion and to explore its contribution in light of the differences observed in the two groups.

The most striking difference found between HASA CT and HASA GP was the way in which the groups function. HASA CT functions in a social manner where relationships are built through conversation and fellowship while HASA GP functions more in a guest-lecturing capacity where “you literally are just part of the audience of whoever’s speaking” (P15, counsellor, Interview 5). HASA GP’s functioning was also described to mimic a “multidisciplinary meeting” with the involvement of several professionals and the outlook of growing the professional community involved with the SG.

“[The meetings] would include the uhm, neurologists, psychiatrists, genetic, the geneticists, the genetic counsellors and then psychologists, and it actually worked quite well. So if anything back then kind of like grew a little bit more.” – P15, counsellor (Interview 5)

Another difference between the groups is the interaction of the genetic counsellor with the members. In HASA CT the genetic counsellor is actively involved in SG meetings through observation, answering questions and offering emotional support. In contrast to this “intimate” involvement with the members of the group, Participant 15 described that her role was to coordinate between the different professionals taking part in the meetings.

“So what’s really ended up being my role was not so much with the support group was, but around the medical professionals around the condition. So there used to be a

professional, like a multidisciplinary meeting and I kind of then started making sure that that took place.” – P15, counsellor (Interview 5)

Despite the differences, there also were similarities noted between the two HASA groups. The first was the underlying urge to provide some type of support and relief to the HASA community and its key role players even though the groups go about the practical execution of this “relief” in different ways. A second similarity was noted as the role of “practical helper” where counsellors often refer patients to appropriate services.

“It’s the simple things like where do they go for a C-PAP machine or where do they go for help in terms of incontinence etcetera.” – P15, counsellor (Interview 5)

A third similarity was found to be a combination of “information provider” and education through structured talks.

“I have done talks, more again from a support perspective, so I used to be an insurance, so obviously there’s a lot of questioning around the disease and insurance and when to claim etc.” – P15, counsellor (Interview 5)

The differences and similarities observed between the two groups reminds of the diversity found between human beings. Humans have similar emotional, physical and social needs, but these needs differ from where in place and time a person finds him- or herself in. Support groups were analysed to have ever-changing context-dependent wants and needs – similar to that of a living organism. When acknowledging such differences in SGs at their core, it is understandable that the role of a genetic counsellor will be dependent on the specific SG and the challenges it faces at a specific time of involvement.

The versatile and adaptable role of a genetic counsellor is ideal to morph into the mould that a specific SG requires at a specific point in time. It is thus deducible that the role of a counsellor in a SG is not determined by the counsellor, but rather by the needs of the living organism that is the SG. As a SG grows and evolves over time the way in which a counsellor can contribute to the group will change. Instead of trying to establish specific roles and a set nature of involvement, a better approach may be for a counsellor to take a step back, observe and see how her multiple skillset can assist a specific group. This final thought is echoed by Participant 7 (HASA member, FG1) when he spoke about the nature of involvement of a counsellor. He alluded to the fact that involvement should not be forced, but should rather be revealed through a specific place and time... “Mmm, maybe time will tell.”

4.3 STRENGTHS OF THE STUDY

- The study is the first to be undertaken in the South African setting to qualitatively explore the role that a genetic counsellor can play in a SG for a genetic condition.
- As far as the researcher is aware it is also the first qualitative study to be conducted internationally regarding a genetic counsellor's role in a SG specifically for HD.
- The interview- and focus group guides were designed to make use of open-ended questions which motivated participants to answer openly and no predetermined themes were imposed upon the members. However, since data analysis took place in parallel to data collection, the researcher explored the ideas and themes that emerged during earlier data gathering events (DGEs) with subsequent DGEs.
- The researcher drew on three different "groups" of perspectives by making use of HASA members, genetic counsellors and a psychologist as participants thus allowing for a more wholistic view on the topic.
- Most participants were known to the researcher and establishing trust and rapport during the DGEs were easy.
- Focus group 1 (the larger of the two FGs) were conducted in the same space that SG meetings usually take place resulting in increased comfort and familiarity for participants. FG 2 took place at a member's house and all three participants were familiar with the specific setting. In addition to the comfort provided by the settings, participants were familiar with each other due to attending the same SG meeting(s) and did not have difficulty communicating together in a group.
- There is a significant void in the literature regarding the role of genetic counsellors in SGs for genetic conditions. This void has caused uncertainty about the appropriateness of a counsellor's involvement and has led to a debate on whether or not Genetic Counselling Masters students should be inspired to become involved with SGs or not. Prior to the study minimal students were getting involved with SGs without having empirical data on whether their lack of involvement was beneficial or detrimental to SGs. The study sheds much-needed light on the topic and provides insight into how genetic counsellors can become involved with SGs (taking into consideration the unique needs of different SGs). It also sheds light on the mutually beneficial relationship between counsellors and SGs and may serve as motivator for such collaborations to take part in the future for the benefit of both the genetic counselling profession and the SG society.

4.4 LIMITATIONS OF THE STUDY

- This was the researcher's first experience conducting qualitative research and as can be expected from a novel researcher, she learnt the methodology as the study unfolded. As data collection progressed, the researcher improved upon previously conducted interviews and focus groups with the result that not all DGEs were of the same quality.
- Most participants were familiar with the researcher and her involvement with HASA. Although prior familiarity could be a strength in establishing rapport during DGEs, participants may have avoided answering in a certain manner as to not offend the researcher's own involvement with HASA.
- The researcher's personal investment in HASA posed a potential threat to the objectivity and trustworthiness of the study. The researcher was aware of her biases (as discussed under "Self-reflection and -position in research study") and measures were taken to minimise the threat (discussed under "Trustworthiness").
- Despite making use of the viewpoints of different people involved with HASA, the HASA members who engaged most actively in the focus groups were those unaffected by HD. The data consequently has limited input from those affected physically by HD (discussed under "Unique contribution of members").
- No objective measure was used to determine the cognitive functioning of symptomatic participants and the researcher's discretion served to determine the extent to which participants were able to provide informed consent.
- The study did not take into consideration the potential recall bias due to the amount of time that had lapsed since participants' involvement with HASA. It also did not take into consideration that the group's functioning may have changed over the past 7 years as it developed from newly-started group to a well-established with a distinct identity.
- The research did not account for proportionality due to the purposive sampling that were done. The participant sample consisted of unequal numbers of HASA members and genetic counsellors and only one psychologist represented her category. However, the proportion HASA members to genetic counsellors to psychologists were representative of real-life involvement where significantly more HASA members are involved with the group than genetic counsellors and SGs in general seldomly have psychologists involved.
- Not all participants contributed equally during the focus groups (discussed under "Unique contribution of participants") and the number of actively-involved focus group members are less than the actual number of participants.

- The results of the study cannot be extrapolated and applied to other SGs as it was determined that even with the same condition SGs have unique identities and functions. It can merely function as starting-point for future studies to be conducted on the role of a genetic counsellor in a SG.
- The study was conducted as a mini dissertation on a Master's degree level. However, as is often found with qualitative data, the deeper the researcher immersed herself in the data, the more layers of analysis and potential discussion were identified. If this was a study on a PhD level the researcher could have explored additional layers of analysis such as the ever-changing role of a genetic counsellor in relation to the evolution of a SG. The researcher could have added a layer of analysis comparing the roles of the counsellors when the HASA CT was still being established and finding its identity versus the roles of the counsellor in a now well-established group.
- The study also did not take into consideration where in the "life cycle" of the support group the members were involved and viewpoints could have differed due to members being involved at the start of the group while identity was still undefined versus a year or two down the line where identity and function had been established.
- As mentioned under "Research background" HASA CT consists of a small group of people. The limited numbers do not allow for a true reflection of the varied population of the CT HD community. HASA members generally have a level of resources available to them that allows for private transport and some assistance with caring for affected loved-ones as opposed to socioeconomically disadvantaged HD community members. The study therefor reflects a certain level of sampling bias and the findings cannot be generalised to apply to the whole CT HD community.

4.5 IMPLICATION OF THE STUDY AND RECOMMENDATIONS FOR FUTURE RESEARCH

The practical implication of the study is that there is a role for a counsellor in SGs and that the collaboration of counsellors and SGs should be encouraged for the benefit of members and counsellors alike. Although the study revealed that the data from one group's involvement cannot be generalised to fit another, the foundation laid through this study can be used as starting point to examine the role of a counsellor in a variety of SGs. This however, does require further investigation and a following research study on a PhD level in order to capture

the magnitude of different layers of analysis. It is recommended that SGs for other conditions than HD are studied as well as SGs functioning on different platforms such as on social media and Whatsapp groups. The possibility for longitudinal studies exploring the evolving nature of a counsellor's involvement could add a significant dimension to the body of literature on the role of a genetic counsellor in a support group.

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Appendices

APPENDIX A: Information sheet and consent to be contacted

University of Cape Town, Department of Pathology, Division of Human Genetics

MSc(Med) Genetic Counselling Research Project

Title: “The role of a genetic counsellor in a support group for Huntington’s disease.”

Investigators:

Maxine du Toit (genetic counselling student) - Tel: 021 404 6235 / 082 546 8551, email: dtmax002@myuct.ac.za

Dr. Tina-Marié Wessels (genetic counsellor) - Tel: 021 406 6373/6995, email: tina.wessels@uct.ac.za

Prof Jacque Greenberg (genetic counsellor) – Tel: 021 406 6299/6695, email: jacque.greenberg@uct.ac.za

What is the aim of the study?

This study aims to explore the role that a genetic counsellor can play within the Huntington’s Association of South Africa (HASA) support group. The research would like to grasp the needs of support group members such as information and counselling they would like to receive from a genetic counsellor within a support group.

What is a genetic counsellor?

A genetic counsellor is a specialist in the medical field who helps people understand the impact of genetic conditions on their lives. They have knowledge about genetics and specialise in counselling. They provide information on living with genetic conditions, the different ways conditions are inherited, genetic testing options and they calculate risk factors of passing conditions on to the next generation. They provide emotional support, help people come to

terms with their conditions and assist them in making decisions that are best for the individual/family.

What does the research involve?

The research involves two separate processes:

Firstly, focus group interviews with members of the HASA support group. A focus group will take around an hour to complete and will consist of maximum 6 support group members. The researcher will ask questions about how the members see the involvement of a genetic counsellor in the group and the members of the group will discuss each question among themselves in the group. The focus groups will take place at Abbots College Claremont where the HASA group meetings are held.

Secondly, individual interviews with genetic counsellors who have been involved with the HASA support group in the past 5 years. During the individual interview the researcher will ask the genetic counsellor his/her view on the involvement of a genetic counsellor in the HASA support group. Individual interviews will be approximately 45 minutes long and will take place at a neutral venue to be agreed upon.

The discussion groups will make use of English as communication medium and the individual interviews will make use of either Afrikaans or English as communication medium.

Both the focus groups and individual interviews will be audio recorded by the researcher to improve accuracy of the information analysed.

What about my privacy?

To protect the privacy of the participants, all personal details will remain separate from responses given in the focus group/interview. Instead of using names, each participant will be given a study code number or pseudonym. No personal information will be shared with anyone and will only be accessible by the researcher herself.

What if I do not want to answer a question?

If you feel that you cannot or do not want to answer any specific question, for any reason, you may inform the interviewer who will then skip that question. Alternatively, in the focus group, you can just abstain from taking part in the conversation regarding the question that you do not want to answer. Participation in this study is completely voluntary and will not affect your role in the HASA group in any way. Should you no longer want to participate in the study, you may choose to withdraw at any point.

What now?

With this form you are being invited to give consent for the researcher, Maxine du Toit (genetic counselling student) to contact you at some time in the future to invite you to participate in the research study. HASA support group members will be invited to attend a focus group session while genetic counsellors will be invited to attend an individual interview.

Are you willing to be contacted for involvement in the study? YES NO

Have you attended at least one HASA support group meeting in the past 5 years? YES NO

If your answer is YES to both the above questions, please fill out your contact information below.

Name: _____

Cell phone: _____

Telephone: _____

Email: _____

Signature: _____

Date: _____

This completed form can be emailed to the researcher at: dtmax002@myuct.ac.za or alternatively can be handed back to the researcher at a HASA support group meeting.

APPENDIX B: Participant consent form

University of Cape Town, Department of Pathology, Division of Human Genetics

MSc(Med) Genetic Counselling Research Project

Title: “The role of a genetic counsellor in a support group for Huntington’s disease.”

This study aims to explore the role that a genetic counsellor can play within the Huntington’s Association of South Africa (HASA) support group. The research would like to grasp the needs of support group members such as information and counselling they would like to receive from a genetic counsellor within a support group.

STATEMENT BY PARTICIPANT

I, _____ confirm that:

I have been invited to be involved in the above-mentioned research project which has been initiated through the division of Human Genetics at the University of Cape Town. I understand that 15-25 other adult participants will be involved in the study and that my name and other personal information will not be discussed with the other participants or with anyone else not involved in the study.

I understand that the objective of the study is to determine what role a genetic counsellor can potentially play within the HASA support group.

I understand that the interview/focus group will take place in a private setting yet to be determined (possibly at the same venue as the HASA support group) at a time and date yet to be determined (possibly before support group meetings). I understand the interview/focus group will take approximately 60 minutes.

I understand that I voluntarily choose to participate in this study and if I choose to no longer continue that my decision will not in any way affect my role in the support group negatively.

I understand that the questions may cause emotional reactions and that I may choose not to answer any questions if I do not wish to do so. I understand that I may decide to stop with the interview/focus group process at any point if I feel uncomfortable or too emotional.

I understand that my involvement in the study may contribute to health care professionals having a better understanding of the role that a genetic counsellor could play in the HASA support group. This information will assist genetic counsellors in determining how they could have a positive impact in support groups in the future.

I understand that all information collected will remain confidential and will be used for research purposes only.

I understand that the interview/focus group session will be recorded for research purposes. All audio recordings will be safely stored away in locked cupboards and information stored on a computer will only be accessible via a password. I understand that only the researcher will have access to the data. All recordings will be destroyed upon completion of this study and all identities will remain anonymous.

I understand that the interview will take place in English and/or Afrikaans and that the researcher will be administering the interviews herself.

I understand that this study has been approved by the registered Human Research Ethics Committee at the Faculty of Health Sciences at the University of Cape Town. I have been given contact details should I wish to contact the committee about how I was treated as a research participant.

I have the researcher's and her supervisor's contact details if I would like to contact her regarding further questions about this study.

_____ has explained the information of this study in English/Afrikaans and I understand this information.

I HEREBY DECLARE THAT I HAVE VOLUNTARILY AGREED TO PARTICIPATE IN THE ABOVE RESEARCH STUDY AND THAT THE INTERVIEW MAY BE AUDIO RECORDED.

Signed at _____ (address of venue) on _____ (date)

Participant's name and signature

Witness' name and signature

If you have any questions regarding your rights as a research participant, please contact the Human Research Ethics Committee at the Faculty of Health Sciences of the University of Cape Town.

Professor Marc Blockman (Chairperson of the Human Research Ethics Committee): Tel: 021 406 6496

If you have any questions regarding the research or the research procedure, please contact the researcher or her supervisor:

Maxine du Toit (Researcher) Tel : 082 546 8551, email : dtmax002@myuct.ac.za

Dr Tina-Marié Wessels (Supervisor) Tel : 021 406 6373/6995, email : tina.wessels@uct.ac.za

Prof Jacquie Greenberg (Supervisor) Tel : 021 406 6299/6995, email : jacquie.greenberg@uct.ac.za

APPENDIX C: Participant socio-demographic information sheet

Age range:

18-25 years	
26-35 years	
36-45 years	
46-55 years	
56-65 years	
66+ years	

Education level:

No formal education	
Primary education	
Secondary education - without grade 12	
Grade 12	
Diploma	
University degree	

Marital status:

Single	
Stable relationship	
Married	
Divorced/ separated	

Relation to HASA support group:

Affected with HD (gene positive) - symptomatic	
Affected with HD (gene positive) - asymptomatic	
Possibly affected with HD (at risk but not yet tested)	
Family member affected with HD	
Friend affected with HD	
Genetic counsellor	
Psychologist	

APPENDIX D: Interview guide for genetic counsellors

Tell me about your involvement with HASA?

How long have you been involved with the group?

How often do you attend the group?

What is your relationship to the group?

What are your reasons for attending the group?

Tell me about your role within the HASA group?

What has your function been in the group?

How have group members responded to your presence in the group?

What questions have group members asked you?

What therapeutic role have you played in the group?

How do you see the possible need for a genetic counsellor in the HASA support group?

What are the needs that group members have expressed that could be met by the attendance of a genetic counsellor?

How do you see those needs being met by a genetic counsellor?

What positive contribution could a genetic counsellor make to the group?

How could the presence of a genetic counsellor possibly play a negative role within the support group?

What therapeutic role do you think a genetic counsellor could play in the group?

What would you as genetic counsellor like to get from the group in the future?

Would you like to share anything else about the possible involvement of a genetic counsellor in the HASA support group?

APPENDIX E: Interview guide for HASA psychologists

Tell me about your involvement with HASA?

How long have you been involved with the group?

How often do you attend the group?

What is your relationship to the group?

Tell me about your role within the HASA group?

What has your function been in the group?

How have group members responded to your presence as professional in the group?

Tell me about your understanding of genetic counselling?

What is your understanding of what a genetic counsellor does?

What experience have you had with genetic counselling?

How do you see the possible need for a genetic counsellor in the HASA support group?

How do you think HASA members experience the role of a genetic counsellor in the group?

What are the needs that group members have expressed that could be met by the attendance of a genetic counsellor?

How do you see those needs being met by a genetic counsellor?

What positive contribution could a genetic counsellor make to the group?

How could the presence of a genetic counsellor possibly play a negative role within the support group? Please elaborate.

What therapeutic role do you think a genetic counsellor could play in the group?

Would you like to share anything else about the possible involvement of a genetic counsellor in the HASA support group?

APPENDIX F: Focus group guide

Let's start off by going around in the group saying who each person is and what your involvement with HASA is

Tell me about your involvement with HASA?

How long have you been involved with the group?

How often do you attend the group?

What do you get from the group?

Tell me about your understanding of genetic counselling?

What is your understanding of what a genetic counsellor does?

Who of you have experience with a genetic counsellor? Please elaborate.

Do you think genetic counselling could be beneficial to you? Please elaborate.

How do you see the possible involvement of a genetic counsellor in HASA support group?

Do you think a genetic counsellor could be beneficial to the HASA support group?
Please elaborate.

What role do you think a genetic counsellor could play in the group?

What information would you like to receive from a genetic counsellor in the group?

What therapeutic role do you think a genetic counsellor could play in the group?

Have you had any negative experiences with a genetic counsellor in the support group?

How do you think the presence of a genetic counsellor can be detrimental to the support group?

What would you like a genetic counsellor to know about HD and your personal experience with it?

Would you like to share anything else about the possible involvement of a genetic counsellor in the HASA support group?

APPENDIX G: Ethics approval



UNIVERSITY OF CAPE TOWN
Faculty of Health Sciences
Human Research Ethics Committee



Room E53-46 Old Main Building
Groote Schuur Hospital
Observatory 7925
Telephone [021] 406 6626
Email: shuretta.thomas@uct.ac.za
Website: www.health.uct.ac.za/fhs/research/humanethics/forms

23 August 2018

HREC REF: 420/2018

Dr Tina-Marie Wessels
Human Genetics
Pathology

Dear Dr Wessels

PROJECT TITLE: THE ROLE OF A GENETIC COUNSELLOR IN A SUPPORT GROUP FOR HUNTINGTON'S DISEASE (MSc Candidate - Mrs M du Toit)

Thank you for submitting your response to the Faculty of Health Sciences Human Research Ethics Committee.

It is a pleasure to inform you that the HREC has **formally approved** the above-mentioned study.

Approval is granted for one year until the 30 August 2019.

Please submit a progress form, using the standardised Annual Report Form if the study continues beyond the approval period. Please submit a Standard Closure form if the study is completed within the approval period.

(Forms can be found on our website: www.health.uct.ac.za/fhs/research/humanethics/forms)

Please quote the HREC REF in all your correspondence.

Please note that the ongoing ethical conduct of the study remains the responsibility of the principal investigator.

Please note that for all studies approved by the HREC, the principal investigator **must** obtain appropriate Institutional approval, where necessary, before the research may occur.

The HREC acknowledges that the student, Maxine du Toit will also be involved in this study.

Yours sincerely

Signature Removed

PROFESSOR M BLOCKMAN
CHAIRPERSON, FHS HUMAN RESEARCH ETHICS COMMITTEE

Federal Wide Assurance Number: FWA00001637.

Institutional Review Board (IRB) number: IRB00001938

This serves to confirm that the University of Cape Town Human Research Ethics Committee complies to the Ethics Standards for Clinical Research with a new drug in patients, based on the Medical

HREC 420/2018

Research Council (MRC-SA), Food and Drug Administration (FDA-USA), International Convention on Harmonisation Good Clinical Practice (ICH GCP), South African Good Clinical Practice Guidelines (DoH 2006), based on the Association of the British Pharmaceutical Industry Guidelines (ABPI), and Declaration of Helsinki (2013) guidelines.

The Human Research Ethics Committee granting this approval is in compliance with the ICH Harmonised Tripartite Guidelines E6: Note for Guidance on Good Clinical Practice (CPMP/ICH/135/95) and FDA Code Federal Regulation Part 50, 56 and 312.