

Parents' perspectives and experiences of having a child with hereditary hearing loss

By

Sinead Amber Ross

RSSSIN001

SUBMITTED TO THE UNIVERSITY OF CAPE TOWN

In partial fulfilment of the requirements for the degree

MSc (Med) Genetic Counselling



Division of Human Genetics

Department of Pathology

Faculty of Health Sciences

UNIVERSITY OF CAPE TOWN



Date of submission: 15 August 2016

Supervisor:

Prof Ambroise Wonkam

Co-supervisors:

Ms Nakita Laing

Dr Sumaya Mall

The copyright of this thesis vests in the author. No quotation from it or information derived from it is to be published without full acknowledgement of the source. The thesis is to be used for private study or non-commercial research purposes only.

Published by the University of Cape Town (UCT) in terms of the non-exclusive license granted to UCT by the author.

DECLARATION

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

I empower the university to reproduce for the purpose of research either the whole or any portion of the contents in any manner whatsoever.

Signature:

Signed by candidate

Signature removed

Date: 15 August 2016

ABSTRACT

Congenital hearing loss (HL) occurs in 1-2 per 1000 newborns globally. Of all the congenital diseases that occur worldwide, HL remains the most disabling, with the highest rate for age-standardised disability life years and is a significant public health concern particularly in the developing world. In South Africa, 6 in 1000 newborns, have disabling HL. Genetic aetiology accounts for half of the cases of prelingual HL and of these 70% are nonsyndromic. There is ongoing research into the genetic basis of HL in the South African setting as at present, genetic testing for HL is largely uninformative in most African populations. Previous studies have examined the psychosocial challenges experienced by parents related to their child's HL, but in South Africa there have been no studies investigating the hereditary aspects of HL and how they may be related to the experiences of parents. Furthermore, the understanding and comprehension of genetics concepts is unknown in this population and needs to be explored before appropriate genetic counselling may be provided to parents of children with hereditary HL. Therefore, the aim of this research was to examine the perspectives, experiences and understanding of parents, whose children are deaf, potentially due to a nonsyndromic genetic aetiology, in a selected setting in Cape Town.

Eleven participants were recruited through the clinical genetics service at Red Cross War Memorial Children's Hospital and the Dominican School for Deaf Children in Cape Town. Semi-structured interviews were conducted, and the transcripts analysed using the framework approach for data analysis. Through this approach, five themes were identified relating to the experience of parenting a deaf child, parents' understanding of the cause of their child's HL and the way that their child's difference or normalcy is perceived.

Most parents were unable to identify the cause of their child's HL unless there were previous cases of HL in the family, but almost all were interested in pursuing genetic testing to assist them in finding answers. The time from the diagnosis had been an emotional and confusing time for many of the participants, who found means to cope and adapt to their child's HL through faith and support systems. Each of the participants had a unique experience with regards to their child's HL, some needing to make sacrifices and overcome practical challenges in order to provide for their child. Participants both rejected shame and pity with regards to their child's HL and embraced that their child was different, part of the Deaf world, but that this difference does not define them. This research could influence the genetics services provided for this population. It is imperative to understand the experiences and comprehension of this population so that improvements can be made to provide appropriate services.

DEDICATION

I dedicate this minor dissertation to my sister,
who lost her hearing in one ear when she was 9 years-old.

For the endless laughter over the things you hear.

And never letting your hearing loss hold you back.

ACKNOWLEDGEMENTS

I would like to acknowledge and show thanks to the following individuals, without whom, this research would not have been possible:

- Firstly, I would like to thank the parents who made themselves available to be involved in this research. Being the parent of deaf child, I have now learnt, is no easy task. The job that you do as parents is inspiring. Thank you for your openness and willingness to share your stories with me.
- To the Dominican School for Deaf Children, for allowing me to use your school for my research. You are doing a commendable job. In particular, I would like to thank Sister Edna Smith, without whom I would not have been able to do this research. Your help in recruiting and contacting parents was invaluable and I cannot thank you enough for the time you took away from all of your other duties to assist me in my research.
- To my supervisor, Prof Wonkam, thank you for sparking my interest into the fascinating topic of hearing loss. I hope that we can work together in the future as your enthusiasm for hearing loss, as well as other genetic conditions, is the reason that I would like to (in at least some part) stay in academia once I have completed my genetic counselling training.
- To Nakita Laing, for always being across the hallway, and for being a friend and colleague throughout this experience. Your insight, patience and experience is greatly appreciated.
- Sumaya Mall, thank you for introducing me to the qualitative research world, and even through your move to Johannesburg, being a solid support and fierce mentor.
- To my family, although you are far, I never felt alone, and your endless support and motivation through all of my endeavours is so appreciated. In particular, thank you for the financial assistance. Dad, I know you said as long as I was willing, you would help, and that kind of encouragement is all I ever needed.
- A very special thank you to my fellow students, in particular Gill and Tarryn, for being my hypothetical “rescue remedy” for the last year.
- To the rest of my “other family”, the clinical team in the Division of Human Genetics. You are all fantastic clinicians and your dedication to the field of genetics is inspiring.
- And finally, to Jonathan, for your unyielding support and love.

TABLE OF CONTENTS

Declaration	i
Abstract	ii
Dedication	iii
Acknowledgements.....	iv
Table of contents	v
List of terms and abbreviations.....	ix
List of tables and figures	i
Chapter 1: Introduction	1
1.1 Chapter introduction.....	1
1.2 Epidemiology of hearing loss	1
1.3 Definition and characterisation of hearing loss	2
1.4 Genetic aetiology of hearing loss.....	3
1.5 Hearing loss in the South African context.....	5
1.6 Hearing loss as a disability versus Deafness as a culture.....	6
1.6 Chapter summary.....	8
Chapter 2: Literature review.....	9
2.1 Chapter introduction.....	9
2.2 Literature review	9
2.2.1 Psychosocial challenges related to hearing loss	9
2.2.2 Specific challenges related to inheritable hearing loss	12
2.2.3 Genetics services in South Africa and the role of genetic counselling for hereditary hearing loss	13
2.3 Aim and objectives	16

2.3.1 Aim of this study.....	16
2.3.2 Objectives of this study	16
2.4 Chapter summary.....	16
Chapter 3: Methods.....	17
3.1 Chapter introduction.....	17
3.2 Research design	17
3.2.1 Qualitative research	17
3.2.2 The framework approach to qualitative data analysis.....	18
3.3 Study population and sample	19
3.3.1 Study settings	19
3.3.2 Study population	20
3.3.3 Sampling method	20
3.3.4 Sample size	20
3.4 Data collection	21
3.5 Data analysis.....	22
3.6 Validity and reliability.....	23
3.7 Ethical considerations	24
3.7.1 Ethical approval	24
3.7.2 Consent.....	24
3.7.3 Confidentiality	24
3.7.4 Risks and benefits to participants	25
3.8 Chapter summary.....	25
Chapter 4: Results and discussion	26
4.1 Chapter introduction.....	26

4.2 Reaching the research objectives	26
4.3 Socio-demographic data of participants	27
4.4 Identified themes	31
4.5 Theme 1 - The initial journey: shock, acceptance and adaptation	31
4.5.1 The loss of a “normal” child	31
4.5.2 Support and its role in acceptance and adaptation	35
4.6 Theme 2 - Comprehension and understanding of the cause of their child’s hearing loss	38
4.6.1 “I don’t know where it comes from”	39
4.6.2 Guilt and blame	40
4.6.3 The need for answers	42
4.7 Theme 3 – Role of the mother: the experience of being a parent of a deaf child	45
4.7.1 Practicalities and sacrifice	45
4.7.2 The need to protect.....	49
4.8 Theme 4 - Being different: shame, stigma and the Deaf world.....	51
4.8.1 Stigma, shame and the rejection of pity	51
4.8.2 The Deaf world	54
4.9 Theme 5 - Being normal: “Deaf people are people”	56
4.10 Chapter summary.....	57
Chapter 5: Conclusions, strengths and limitations of this study and recommendations for future research	58
5.1 Chapter introduction.....	58
5.2 Conclusions	58
5.3 Strengths of this study.....	60
5.4 Limitations of this study.....	60

5.5 Practical implications of this research	61
5.6 Recommendations for future research.....	61
References	63
Appendices	73
Appendix A: Information sheet for participant recruitment	73
Appendix B: Participant consent form	75
Appendix C: Questionnaire	76
Appendix D: Interview guide.....	77
Appendix E: Permission from the Western Cape Education Department.....	79
Appendix F: Permission from the Dominican School for Deaf Children	80
Appendix G: Ethical approval	81
Appendix H: Study deviation form.....	83

LIST OF TERMS AND ABBREVIATIONS

CF – cystic fibrosis

DALYs – disability-adjusted life-years

dB - decibels

Deaf – membership to a culturally distinct minority group

deaf – colloquial term used to describe someone with severe-to-profound hearing loss

DSDC – Dominican School for Deaf Children

EHDI – Early hearing detection and intervention

GCs – Genetic counsellors

GJB2 – Gap junction protein beta-2

HIV/AIDS – Human immunodeficiency virus/Acquired immunodeficiency syndrome

HL – Hearing loss

MOU – midwife obstetric unit

NBS – Newborn screening

NSHL – Nonsyndromic hearing loss

RCWMCH – Red Cross War Memorial Children’s Hospital

SA – South Africa

SASL – South African Sign Language

SCD – sickle cell disease

UCT – University of Cape Town

USA – United States of America

WHO – World Health Organisation

LIST OF TABLES AND FIGURES

Figure 1: Anatomy of the human ear (Fox, 2008).....	2
Table 1: Degree/severity of hearing loss (Clark, 1981)	3
Figure 2: Causes of prelingual hearing loss (>40dB) in children (Smith et al., 2014)	4
Figure 3: Impact of hearing loss on the family	10
Table 2: Number of participants recruited by the various recruitment methods and locations	26
Table 3: Participant characteristics.....	27
Table 4: Child characteristics	28
Figure 4: Pedigree showing the relationship between P6 and P10.....	28
Table 5: Socio-demographic information of the participants and their child	30
Figure 5: Grief process model of parents of children with disabilities. Adapted from Andereg, Vergason & Smith (1992)	34

CHAPTER 1: INTRODUCTION

1.1 CHAPTER INTRODUCTION

This introductory chapter outlines, (1) the epidemiology of hearing loss, (2) the definition and characterisation of hearing loss and in particular (3) the genetic aetiology of hearing loss, as well as explores (4) hearing loss in the South African context and (5) the dichotomised view of d/Deafness.

1.2 EPIDEMIOLOGY OF HEARING LOSS

Hearing loss (HL) is a significant public health issue, particularly in the developing world. Often referred to as the “silent epidemic”, HL is the most prevalent sensorineural defect and a significant contributor to disease burden globally (Olusanya, Neumann & Saunders, 2014).

Of all the congenital disorders that occur worldwide, HL remains the most disabling, with the highest rate for age-standardised disability-adjusted life-years (DALYs) which is approximately 1.5-2 times that of congenital heart disease and other congenital anomalies (Murray *et al.*, 2015). Since 1990, despite vast improvements in hearing aid and implant technologies, HL has increased in rank as the 13th leading cause of DALYs, and is the only sensory deficit to make the top 25 list of all-disease causes of DALYs in the world (Murray *et al.*, 2015).

Regardless of the age at which HL develops, it may have far-reaching implications for the quality of life, ability to communicate and economic independence of those affected. Disabling congenital HL will be experienced over a lifetime, that is affecting all stages of life, from the development of speech and language to the ability to achieve academically and vocationally (Kotby *et al.*, 2008, Mason & Mason, 2007). Children with HL may be at increased risk for physical or emotional abuse, stigmatisation and societal integration (Jones *et al.*, 2012). Adults with HL may be further disadvantaged by depression, loneliness, restricted job opportunities and thus reduced income. Therefore, HL belongs not only to those affected but is a substantial public health concern (Olusanya, Neumann & Saunders, 2014).

The first estimate of the global prevalence of disabling HL (moderate-profound HL in one/both ears), conducted in 1985, estimated that approximately 42 million people (0.9% of the world’s population) had a disabling hearing impairment. Ten years later, in 1995, this number more than doubled to 120 million people (2.1% of the world’s population) (Smith, 2001, 2003). In 2011, the World Health Organisation (WHO) estimated that there are 360 million people (>5% of the world’s population) living with disabling HL, with 32 million of these being children under the age of 15 years (WHO, 2015).

These estimates are based on disabling HL but this incidence would be significantly higher if it included milder and unilateral HL.

Hearing loss is estimated to affect 1 in 1000, and 3 in 1000 live births in developed and developing countries, respectively. According to the WHO, the prevalence of disabling HL in children is greatest in Asia and Sub-Saharan Africa, with a prevalence rate of 6 per 1000 live births reported in South Africa (SA) (Swanepoel, Störbeck & Friedland, 2009).

1.3 DEFINITION AND CHARACTERISATION OF HEARING LOSS

Physiologically, hearing is a complex sensory process. Sound waves travel down the external auditory canal and strike the tympanic membrane (ear drum), causing it to vibrate. These vibrations are then passed to the bones of the middle ear, which amplify the sound and transfer these waves to the cochlear in the inner ear. Once in the cochlear, sensory hair cells convert these waves into electrical impulses which travel via the auditory nerve to the brain (figure 1). The brain then translates these impulses as sounds (Barrett *et al.*, 2010).

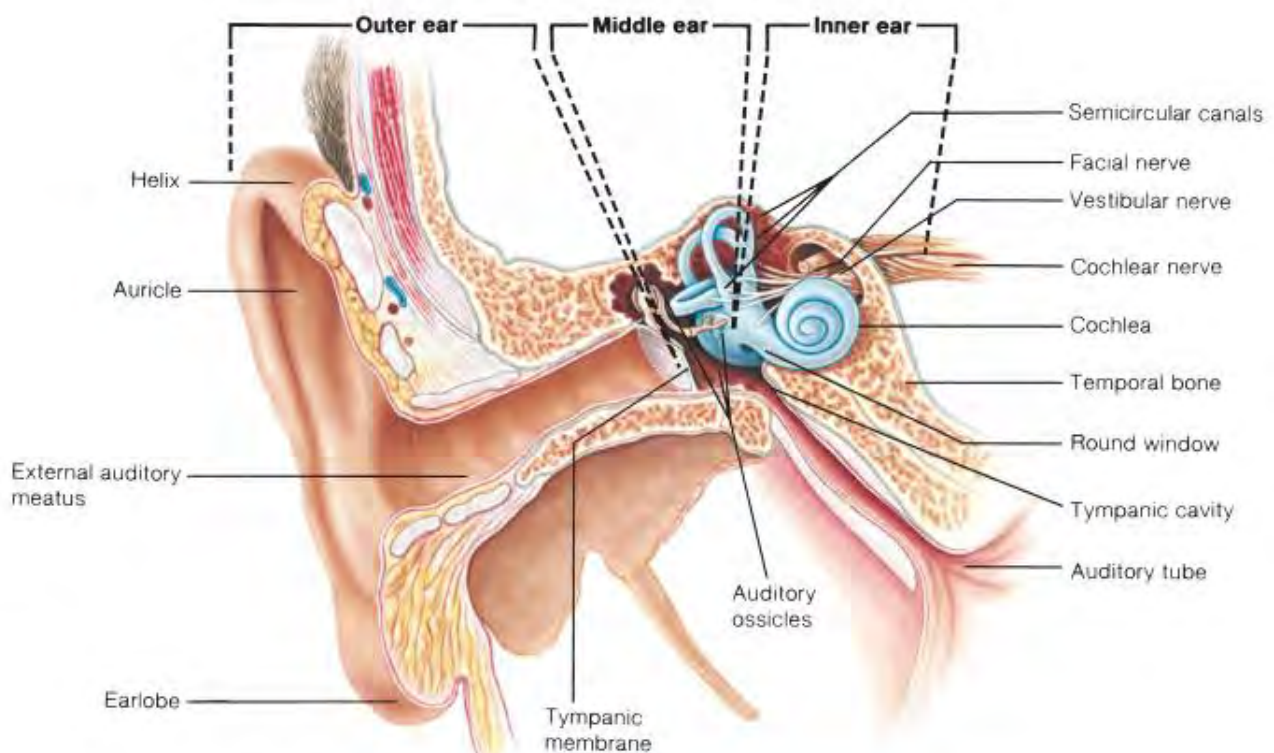


Figure 1: Anatomy of the human ear (Fox, 2008)

The loudness of a sound is correlated to the amplitude of a sound wave, while the pitch is correlated to its frequency. The amplitude of sound is represented in terms of the maximum pressure change at

the tympanic membrane, this is measured in decibels (dB). An audiometer, used to measure auditory perception, presents pure tones at differing frequencies via earphones to the subject. The threshold intensity at each frequency is then measured as an objective measurement of the degree of hearing ability (table 1) (Barrett *et al.*, 2010). Hearing loss is defined as disabling when the loss of hearing is greater than 40 decibels in the better hearing ear for adults and greater than 30dB in children (WHO, 2015).

Table 1: Degree/severity of hearing loss (Clark, 1981)

Degree of hearing loss	Hearing loss range (dB)
Normal	-10 to 15
Slight	16 to 25
Mild	26 to 40
Moderate	41 to 55
Moderately severe	56 to 70
Severe	71 to 90
Profound	91+

Hearing loss is further characterised by its age of onset (congenital or later onset; prelingual or postlingual), type of HL (conductive, sensorineural or mixed), and aetiology (environmental or genetic) (Carey & Palumbos, 2016).

According to the aetiology of HL, there are three main types; those that are the result of environmental factors such as meningitis, or maternal rubella during pregnancy, those that are the result of an interplay of genetic predisposition as well as environmental factors, such as age-related HL, and those that are based purely on genetic influences, such as Waardenberg syndrome (Parker & Bitner-Glindzicz, 2014).

1.4 GENETIC AETIOLOGY OF HEARING LOSS

Genetic aetiology accounts for 50% of prelingual HL in children (figure 2). Genetic forms of HL may be further delineated according to their inheritance pattern; either recessive, dominant, X-linked and in a small proportion mitochondrial. When HL is associated with clinical features in at least one other

body system it is classified as syndromic and accounts for approximately 30% of genetic HL. There are over 400 syndromes described that include HL (OMIM, 2016). Well-defined syndromic forms of HL include Waardenberg syndrome which is associated with pigment abnormalities, and Usher syndrome, in which HL is coupled with vision loss, retinitis pigmentosa (Parker & Bitner-Glindzicz, 2014, Smith *et al.*, 2014).

Approximately 70% of genetic HL cases are isolated and referred to as nonsyndromic. Nonsyndromic hearing loss (NSHL) is highly heterogeneous with varying modes of inheritance, age of onset and severity.

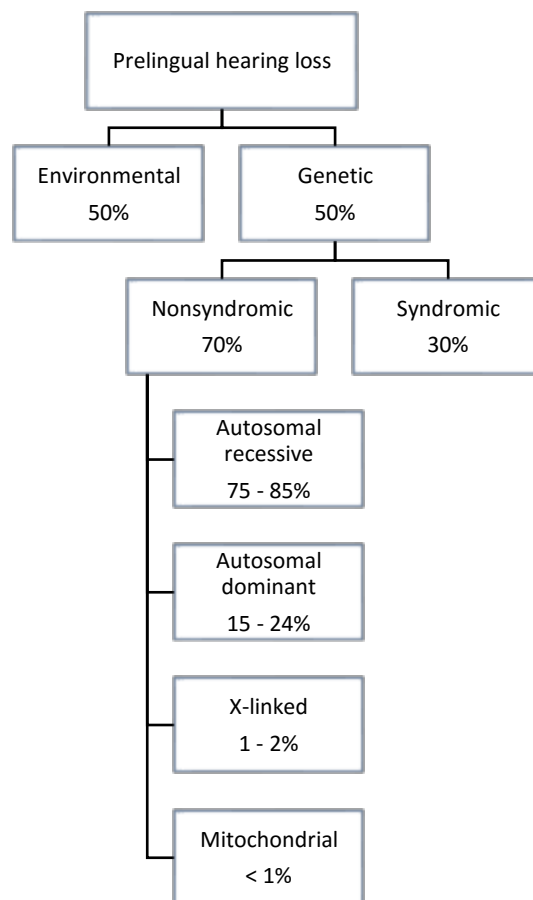


Figure 2: Causes of prelingual hearing loss (>40dB) in children (Smith *et al.*, 2014)

More than 1% of all genes (of which there are approximately 20 000) are involved in determining the structure and functioning of the ear, which alludes to the complexity of this organ. During embryological development there is a propensity for genetic “mishaps” involving the ear and hearing (Copley & Friderichs, 2010). Mutations may occur in many of the inner ear’s systems, including the control and adhesion of hair cells, neurotransmitter release, together with intracellular and intercellular transport (Egilmez & Kalcioğlu, 2016).

In 1997, mutations in the gap junction protein beta-2 (*GJB2*) gene, which codes for the connexin 26 protein, was discovered as the leading cause of autosomal recessive HL in most populations of European and Asian ancestry, accounting for approximately 50% of cases (Kelsell *et al.*, 1997, Denoyelle *et al.*, 1997). Connexin 26 plays a critical role in potassium cycling in the cochlear (Chang, 2015). Subsequent to this discovery the gap junction genes remain the major contributors to deafness globally (Chan & Chang, 2014). Other common genes involved in HL include; *GJB6*, *SLC26A4*, *MYO15A* and *OTOF*.

To date, approximately 170 NSHL loci have been mapped and 98 genes identified (Hereditary Hearing Loss Homepage; [http://hereditaryhearingloss.org./](http://hereditaryhearingloss.org/)). However, as many as 1000 NSHL genes remain to be identified based on diseases associated with HL and unique inner ear transcripts (Hertzano & Elkon, 2012).

Interestingly, unlike many of the world's populations, the prevalence of *GJB2*- or *GJB6*-related NSHL in several sub-Saharan populations is close to zero, for example Kenya and Uganda in East Africa (Gasmelseed *et al.*, 2004, Javidnia *et al.*, 2014), Cameroon in Central Africa (Bosch *et al.*, 2014b, Wonkam *et al.*, 2015), Nigeria in West Africa (Lasisi *et al.*, 2014) as well as in South Africa (Bosch *et al.*, 2014b, Kabahuma *et al.*, 2011). This prompts further investigation into the molecular genetic cause of HL using next generation sequencing which is revealing new mutations in African populations.

1.5 HEARING LOSS IN THE SOUTH AFRICAN CONTEXT

Context is essential in understanding the role that HL loss plays in the lives of those affected. In a country where unemployment and the HIV/AIDS epidemic are rife, poverty is further exasperated by SA's history of oppression and segregation. The quality of life for many South Africans is depressed and malnutrition and lack of access to health care and education is widespread. Several large cities are considerably urbanised, whereas much of the population lives in larger rural settlements and migration between the two are common. In this context, the deaf child in SA may face several barriers, including inaccessibility to basic services, transportation problems, violence and crime as well as discrimination according to race, gender, culture and disability (van der Spuy & Pottas, 2008).

In the developed world, newborn screening (NBS) has become common practice, resulting in earlier diagnosis and consequently better outcomes for children with HL as treatment and intervention strategies may be initiated earlier. Initial screening for HL should occur before the first month of life, which should be followed by further diagnostic evaluation and referral to early intervention before the age of 6 months (Joint Committee on Infant Hearing, 2007).

In a developing country like SA, where resources are restricted and NBS for HL is not widespread, more than 90% of children born do not have access to screening or early detection. Although early hearing detection and intervention (EHDI) has begun to be implemented in both the private and public health care sectors, services in the public sector are far scarcer with only 7.5% of public hospitals offering some form of newborn hearing screening. This is a worrying statistic as approximately 85% of the South African population rely on public health care (Theunissen & Swanepoel, 2008). Conversely, 53% of private health care obstetric units offer NBS (Meyer *et al.*, 2012), but the private health sector only serves a minority of the population.

In SA, the average age of diagnosis of HL is 23 months, which is markedly delayed for effective intervention implementation (Swanepoel, Störbeck & Friedland, 2009). This age may increase in more rural areas of the country, which would be more comparable to the ages of diagnosis seen in other African countries. For example, in Cameroon the mean age of HL diagnosis is 3.3 years (Wonkam *et al.*, 2013) and in Kenya the mean age of HL diagnosis is 5.5 years (Omondi *et al.*, 2007). A later diagnosis has far-reaching implications as it impedes speech, delays language development and can have effects on cognitive, emotional and psychosocial development of the child. These initial consequences can then have far-reaching effects on education, employment and societal integration (Kotby *et al.*, 2008).

After diagnosis, it is crucial that intervention takes place for appropriate management of a child's hearing impairment. Early intervention includes amplification and therapeutic intervention as well as a programme that supports the family during this experience (Swanepoel, Störbeck & Friedland, 2009). The aim of early intervention is to allow the child to access communication, meet the developmental needs of the child and provide the family with appropriate information so that decisions regarding communication modality, method of amplification and schooling may be made (Carpenter, 2007).

Unfortunately in SA, there is limited infrastructure in place for those with HL and many specialised services are not country-wide (Kiyaga & Moores, 2003). Access to assistive technologies, although considered a basic human right for persons with a hearing impairment, are not widely available in developing countries. This is due to affordability, lack of awareness and a chronic shortage of professionals and services to provide appropriate realisation of this basic human right (Störbeck, 2012).

1.6 HEARING LOSS AS A DISABILITY VERSUS DEAFNESS AS A CULTURE

Several definitions are required to understand how the terms HL and deafness are used in the literature as well as in this study. Firstly, HL and hearing impairment are two terms that are often used

interchangeably, referring to audiometric profiles below threshold levels for normal hearing. These terms are used by health care professionals to describe the pathological loss or absence of hearing. The term HL in itself is imperfect, as those born with a congenital hearing impairment have not “lost” their hearing as it was never there to begin with. These terms, although used widely in published literature, are not necessarily the most appropriate, but HL was chosen for this study because of its wide use and hearing impairment was avoided due to its negative connotations.

Deaf (with a small “d”) is a more colloquial term that refers to those who have severe-to-profound HL. Deaf (with a capital “D”) refers to the cultural identity of being deaf, forming part of a Deaf community¹. Someone who is Deaf may not consider themselves to be impaired or that they have lost their hearing, their deafness is not considered pathological (Smith *et al.*, 2014).

As described above, there are several means of classifying those with HL so that their deficit may be quantifiable. This medical model of deafness reinforces that those with HL are abnormal. Those that have a hearing impairment are seen according to their communicative disability, without regard for the implications that HL may have on the individual. Advancing medical technologies, such as hearing aids and cochlear implants, have further fuelled society’s view of deafness as a disability and science’s role in eradicating it (McKee, Schlehofer & Thew, 2013).

The term disability may be defined as a physical or cognitive limitation, but as Munoz-Baell & Ruiz (2000) state, “deaf people are more disabled by their transactions with the hearing world than by the pathology of their hearing impairment”. Without a common language, D/deaf people may feel excluded from the “hearing world”. But as Davis (2006) explains “The “problem” is not the person with the disabilities; the problem is the way that normalcy is constructed to create the “problem” of the disabled person.”

Historically, the eugenics movement was constructed with the aim of eliminating those who may be seen as defective. Alexander Graham Bell promoted the idea that human speech be prioritised over sign language and that if the deaf chose deaf partners, the result would be a race of deaf people, which needed to be avoided. The ideals of the eugenicists, although morally outrageous, may still be adopted by medical – and particularly genetic – research with deaf people (Davis, 2006).

With the advent of specialised genetics services and the boom in genetic technologies since the completion of the Human Genome project, there is the idea that genetics holds the key to “curing” or

¹ It is important to note that hearing status is not the only defining feature of those who form part of the Deaf community. The cultural grouping may include hearing family members and friends of someone who is Deaf.

“eradicating” disabilities (Miller & Levine, 2013). Although researchers may have the best intentions by wanting to prevent HL, the Deaf community’s views need to be taken into consideration, as deafness to them is not an impairment but a unique feature that is part of their identity. In this way, the Deaf may see themselves as differently abled rather than disabled and it is imperative that genetics research is sensitive to the ideals, values, views and opinions of the Deaf community. Their diversity, rather than their assumed disability, needs to be respected and preserved both through research and in the health care setting (McKee, Schlehofer & Thew, 2013).

1.6 CHAPTER SUMMARY

The first chapter of this study provides an introduction to HL, both epidemiologically and within the South African setting. The difference between d/Deafness is introduced as it relates to familial HL. The physiology of hearing and how HL is characterised are briefly presented as well as genetic HL introduced. In the next chapter, an empirical literature review is presented as it relates to the research at hand and the aims and objectives of this study are presented.

CHAPTER 2: LITERATURE REVIEW

2.1 CHAPTER INTRODUCTION

The second chapter of this study will present an empirical literature review related to certain aspects of HL. Literature was searched for using the Google Scholar and PubMed databases, employing the following search terms: *hearing loss, parents, family, genetics, hereditary, disability* and *genetic counselling* as well as permutations of these terms.

The themes extracted from previous literature are presented as they relate to this study, namely, (1) the psychosocial challenges related to HL, (2) specific challenges related to HL that is inherited and (3) the role of genetic counselling for hereditary HL.

The literature review is also systematically presented so that it may contextualise the rationale for this study before the aims and objectives of this study are introduced.

2.2 LITERATURE REVIEW

2.2.1 PSYCHOSOCIAL CHALLENGES RELATED TO HEARING LOSS

Many aspects of family life may be altered in response to the diagnosis of HL in a child and in this way, “deafness belongs not just to the child but to the entire family” (Henderson & Hendershott, 1991).

Given that 90% of deaf children are born to hearing parents, who may have limited experience with HL, the time following diagnosis may be turbulent as the family adapts to a new reality (Jackson & Turnbull, 2004). In response to the diagnosis, parents have previously expressed intense emotions coupled with fear, uncertainty and grief (Jackson, Traub & Turnbull, 2008). The grief response may be attributed to the loss of a “normal” child and may be acute or chronic in its manifestation, but requires parents to find ways to cope and adjust to their new experience (Kurtzer-White & Luterman, 2003).

From the perspective of family life, many aspects of everyday life will undoubtedly be affected by having a child with HL (outlined in figure 3). The first of these, commonly referred to in the literature, is that of communication. Many parents and family members may be faced with learning a new language in order to communicate effectively with their deaf child, which may have marked influences on their social interactions (Henderson & Hendershott, 1991). Learning a new language as an adult may be difficult, frustrating and time-consuming for parents. Without a shared language, children, adolescents and adults with HL may feel excluded from family and social interactions (Jackson, Traub

& Turnbull, 2008) which will have effects on their psychosocial wellbeing and quality of life (Olusanya, Neumann & Saunders, 2014).

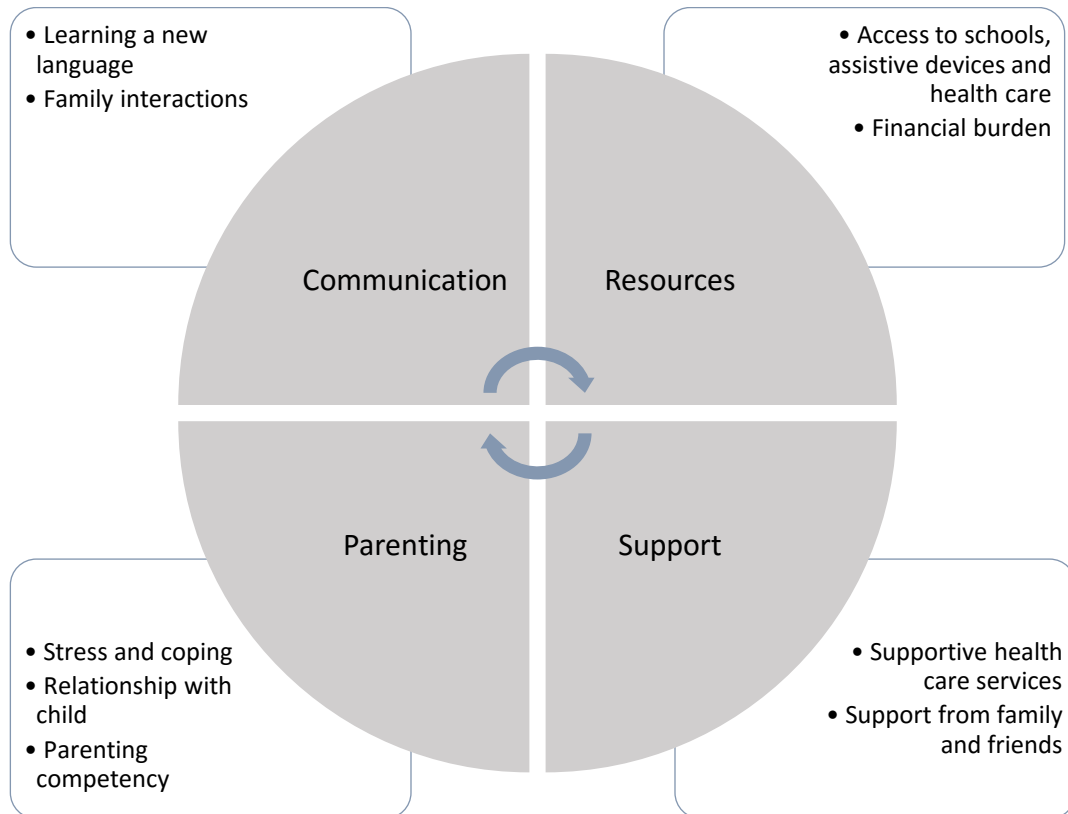


Figure 3: Impact of hearing loss on the family

Following a diagnosis of HL in a child, hearing parents may grapple with their preconceived ideas about parenting, whether they are equipped to having a child with specialised needs and whether they have access to information in order to make decisions regarding their child. This may leave parents to face feelings of incompetency at raising a deaf child, and question their role and ability to effectively parent in this unfamiliar situation (Mason & Mason, 2007).

In studies where parental stress is compared between those having a child with HL and parents whose children do not have HL, there have been conflicting views on whether having a child with HL is more stressful. In some cases, parental stress has been increased in relation to having a child with HL (Quittner *et al.*, 2010) whereas in others, the level of stress is dependent on the time at which parents were interviewed, at some stages, being no different to the stress experienced by other parents. This suggests that at differing times during a child's development, there will be stressors that directly

influence parent-child interactions, but this is not necessarily dependent on the hearing status of the child (Lederberg & Golbach, 2002).

Parents who have previously expressed high levels of stress, confusion and helplessness in relation to their child's HL, attributed this stress to communication difficulties, struggles with discipline and worries regarding schooling and education (Lederberg & Golbach, 2002). These disruptive emotions experienced by parents of a deaf child may be influenced by their ability to make use of coping resources (Zaidman-Zait *et al.*, 2016). On the other hand, it cannot be assumed that all parental and family experiences with HL will be negative, as families who are able to adapt to the fact that they cannot ameliorate the child's HL, may grow closer as a family. Reaching this state can have lasting, positive implications for acceptance of the child as well as acceptance that parenting a child with HL has its associated difficulties which may be overcome with effective parenting strategies and means of support (Blacher, 1984).

Parents of newly diagnosed children with HL need to make use of information and resources in order to make decisions regarding their child's future. The first of these is whether they want to make use of assistive devices to allow their child to access spoken language. In some cases, cochlear implantation at an early age can directly influence how a deaf child will access speech and be able to communicate. In many cases however, this opportunity may not be feasible, due to financial constraints or limited access to specialised health care (Kerr, Tuomi & Müller, 2012). The use of hearing aids for those with profound HL may be ineffective, or parents may choose to forgo hearing assistance for cultural reasons (discussed in section 2.2.2).

Previous research has examined the difficulties and disruptive consequences of a diagnosis as well as coping strategies to alleviate stressors, and positive behaviours such as resilience and routine, to lessen unfavourable outcomes (Ross & Lyon, 2007, Hintermair, 2006). Parents of deaf children have expressed their need for support, in the form of support groups with other parents. Being able to hear other's stories and advice alleviates stressors during the critical period of adaption following diagnosis (Poon & Zaidman-Zait, 2013). Effective referrals to support groups and psychology services are recommended for health care professionals (Jackson, Traub & Turnbull, 2008) and educational systems that serve children with hearing impairments are advised to provide group sessions with families, in order to continue addressing and managing the complex issues that arise when deafness is part of family life (Freeman, Dieterich & Rak, 2002).

Having a deaf child and its associated challenges may be further exasperated by the financial burdens related to the child's HL. Parents have previously mentioned that they need support with regards to the practicalities of having a child with HL, requiring financial assistance for assistive devices,

specialised services and transport (van der Spuy & Pottas, 2008). These troubles may directly influence parental stress (Meinzen-Derr *et al.*, 2008).

Access to other resources also play a role in the impact that HL may have on the family. In SA there is a concentration of schools for the deaf and specialised services in urban cities and less so in rural areas (Kiyaga & Moores, 2003). This may mean that either families need to locate themselves close to services and resources that serve deaf children or be without access to resources that may be crucial in the deaf child's care and development. The same can be said with regards to health care (as discussed in the previous section).

It can be seen that there are diverse challenges that affect family life in relation to having a child with HL. Previous research has focused on the experiences of parents as they are integral to the lives of their child and bear many of the challenges related to their child's HL.

2.2.2 SPECIFIC CHALLENGES RELATED TO INHERITABLE HEARING LOSS

Research related to the psychosocial aspects of HL for the caregiver of the deaf child has previously been investigated in this setting (de Andrade & Ross, 2005, de Andrade, 2015, Kara & Harvey, 2016, Hagedorn, 2015). However, for the purposes of this study, it is proposed that HL of a genetic aetiology may present complexities different to HL of environmental aetiology.

Firstly, the majority of genetic causes of HL are the result of recessive genes, carried by many, and generally only present clinically when both parents transmit the same gene. Therefore, many generations may pass without a case of deafness in a family (Benderly, 1990). Unaware that one may carry a gene for deafness, parents often fail to identify the cause of their child's HL as hereditary (Rodrigues *et al.*, 2013).

Secondly, there may be enhanced stigma felt by parents of children with inherited HL. When the terms genetic and hereditary are introduced, parents may feel stigmatised knowing that they have "passed" this onto their child. These issues have not been explored in the South African context.

A study conducted by Sankar *et al.* (2006) examined the relationship between genetic cause and stigma in the context of hereditary and non-hereditary aetiologies of deafness/HL and breast cancer, and compared to sickle cell disease (SCD) and cystic fibrosis (CF), which are caused by single gene mutations. The Deaf participants assigned a positive interpretation of the genetic cause of their HL, as for many it meant growing up in a family in which they could communicate. This incorporation of a genetic condition as a positive feature of identity was attributed to the fact that the participants' HL was dominantly inherited, and therefore their deafness was seen as normal and valued. However, in

the case of recessive HL, individuals are less likely to have other family members that are deaf and would consequently experience the same limited communication with family and peers as those individuals whose HL is acquired. In the case of conditions that are solely genetic in aetiology, SCD and CF, participants stated that one benefit of having a genetic condition from birth is that one can adapt to the condition. Overall, it was found that individuals interpret the cause of their condition in the context of their lived experiences (Sankar *et al.*, 2006).

Third, parents may struggle to find a reason for their child's condition experiencing guilt and shame. These can be seen as psychological defences against helplessness in receiving difficult news, and provide the illusion of control (Weil, 2000). When feelings of guilt go unresolved, serious implications may ensue, such as hopelessness and disruption of effective parenting. Accurate understanding of the cause of their child's condition, through appropriate genetic counselling can assist in relieving and normalising feelings of guilt (Kessler *et al.*, 1984).

Misunderstandings of inheritance and reproductive risks in genetic causes of disease can have further implications. Information that may be provided at the time of a genetic diagnosis of HL, such as recurrence risks and possibilities for their child's reproductive future are often poorly retained, possibly due to the fact that it seems irrelevant in a time of crisis and confusion. However, the recall of this important information is based on its perceived relevance. When a parent's guilt is involved, emotional relevance may influence how the information is retained (James *et al.*, 2006).

2.2.3 GENETICS SERVICES IN SOUTH AFRICA AND THE ROLE OF GENETIC COUNSELLING FOR HEREDITARY HEARING LOSS

It is well-known that specialised services are lacking in SA, this is true of audiology services but even more so for genetics services. At present, there are only 3 urban centres that offer a comprehensive genetics service in SA, which includes diagnosis, genetic counselling and comprehensive care for those affected by a genetic condition (Beighton *et al.*, 2012). These genetics services integrate clinical and laboratory facilities with the aim to help those affected or concerned about a disorder with a significant hereditary component. With limited resources and a shortage of trained genetics professionals – only 11 medical geneticists and 18 practicing genetic counsellors (J. Greenberg, personal communication, 2015) – most of the South African population are without access to a genetics service.

The University of Cape Town (UCT) is one of four training centres for medical geneticists and one of two for Master's degree training in genetic counselling (Beighton, 2012), the other being the University of Witwatersrand in Johannesburg. This means that most of the research conducted by

genetics academics is undertaken in the larger cities in SA, with the more rural areas going unnoticed. Furthermore, due to the vast number of genetic conditions, it also means that many conditions go under researched in our setting.

South Africa is a unique setting that requires local research undertakings in a variety of conditions that may be genetic in aetiology. Researchers should aim to provide insight into those affected by genetic conditions and propose plans for appropriate services to be provided to these groups.

There has been a paucity of genetics-related research into the hereditary aspects of HL in the South African setting, with the initial aetiological research conducted more than 30 years ago (Sellars & Beighton, 1983) and just followed recently by novel molecular research (Kabahuma *et al.*, 2011, Bosch *et al.*, 2014b, Lebeko *et al.*, 2015, Bosch *et al.*, 2014a). At present, and over the last few years, there is on-going research into the molecular basis of hereditary HL in black African populations (Lebeko *et al.*, 2015, Lebeko *et al.*, 2016, Wonkam, 2015). There have also been several qualitative inquiries into the psychosocial aspects of HL in the South African setting (Mall & Swartz, 2011, Kara & Harvey, 2016, de Andrade, 2015) but at present, there have been no studies that marry the two – the psychosocial related to the hereditary.

Genetic counsellors are in the unique position of providing information in a supportive environment, focusing on the individualised needs of the family (Kaimal *et al.*, 2007). After being informed of a genetic aetiology for a condition, genetic counselling aims to encourage a better understanding of genetic concepts, inheritance patterns and recurrence risks, as well as assisting parents in the coping process (Rodrigues *et al.*, 2013). Parents have previously expressed feelings of guilt and responsibility after hearing that their child's deafness is genetic (Bosteels, Van Hove & Vandembroeck, 2012, Rodrigues *et al.*, 2013) but at this time, the experiences of parents in the South African setting is unknown.

Genetic testing for HL has been explored in other settings and has taken into account the views of hearing parents, deaf parents and the Deaf community. Brunger *et al.* found that hearing parents have a positive attitude towards genetic testing for HL. Most indicated that they wanted to know what caused their child's HL and that this information would be helpful for both future medical management and knowing the chance for recurrence. The authors also describe the participants' inaccurate beliefs and poor understanding of genetics concepts and encourage that genetic counselling be afforded both pre- and post-testing to provide appropriate and accurate information. In this way, parents may make informed decisions and have a clearer understanding of the test result, especially for the comprehension of recurrence risk (Brunger *et al.*, 2000).

Conversely, Deaf adults in some studies have had a mostly negative attitude towards genetic testing (Middleton, Hewison & Mueller, 2001). Further research by Middleton's group in the United Kingdom with Deaf people (part of the Deaf community) and hard of hearing people (part of hearing society) found that Deaf people, who do not see their HL as a disability, were more cautious regarding genetics and that hard of hearing participants felt more positive regarding the advances in genetics. Interestingly, some of the Deaf participants were excited by the advances in technology, as it could mean that they could select for deafness. Sadly, one third of this sample thought that the aim of genetic counselling was to reduce the burden of deafness in society, which highlights the lack of awareness of how genetic counselling may be provided for hereditary HL. If Deaf people are unaware or misunderstand the aims of genetic counselling, then the worry is that they will not make use of this service (Middleton, Emery & Turner, 2010). However, in a recent study in Nigeria, that involved 150 deaf participants (61.3% males, 38.7% females) with a mean age of 26.7 years \pm 9.8. The majority of survey respondents reported believing genetics does more good than harm (79.3%) and 57% expressed interest in genetic testing (Adedokun *et al.*, 2015).

In other settings, as well as in SA, genetic counselling students undergo cultural competence training, but not specifically Deaf awareness training. As most deaf children are born to hearing parents, it is more likely that genetic counsellors (GCs) will interact with hearing parents when discussing genetic testing. Without knowing the difficulties and positive features of raising a deaf child, hearing parents may seek advice and guidance from GCs and as such, GCs should be equipped to assist these parents - be that by referral to support groups, providing important information or general psychosocial support. Interactions with Deaf people may not only be in the prenatal and paediatric setting (regarding genetic testing), but also in the other settings where GCs work, such as cancer genetics. Therefore, it is imperative that GCs be prepared and comfortable in conducting sessions both with hearing parents of deaf children as well as with Deaf people (Nagakura *et al.*, 2015).

Nagakura *et al.* recommend that genetic counselling students be trained in cultural competence and accepting diversity, especially of minority groups such as the Deaf community. However their research has found that not only do genetic counselling students in the United State of America (USA) have little or no interaction with deaf individuals or the Deaf community but most were unsure of how to interact effectively with D/deaf individuals due to a lack of Deaf awareness training i.e. understanding Deafness as a culture, sign language, cochlear implants, terminology and types of deafness (Nagakura *et al.*, 2015).

It can be seen from previous research that the role of genetic counselling for HL needs attention. Given the potential richness of parents' experiences, this minor Masters dissertation will focus on the

experiences of parents of children with hereditary HL in the aim of providing a better clinical genetics service to the deaf child and their family.

2.3 AIM AND OBJECTIVES

2.3.1 AIM OF THIS STUDY

This research aims to examine the perspectives, experiences and understanding of parents, whose children are deaf, due to a putative nonsyndromic genetic aetiology, in a selected setting in Cape Town.

2.3.2 OBJECTIVES OF THIS STUDY

- (1) To recruit a selected group of participants with children with congenital hearing impairment of potentially nonsyndromic genetic origin.
- (2) To perform a face-to-face interview of participants to explore parents' experiences of having a child who has hereditary HL, in various areas of burden (family dynamics, financial, child's quality of life and social and medical support).
- (3) To explore parents' perceptions and understanding of the cause of their child's HL to determine the role for genetic counselling services in this population.

2.4 CHAPTER SUMMARY

In the second chapter of this study, the literature was reviewed and a gap identified. The aim and objectives of this study were presented. In the next chapter, the methods employed in this research are specified and explained.

“Qualitative research humanises science”

- Inge Hutter

3.1 CHAPTER INTRODUCTION

This chapter outlines: (1) the study rationale for qualitative research, (2) why qualitative methodologies are appropriate for research in the field of genetic counselling, and (3) details of the recruitment, data collection and analysis.

3.2 RESEARCH DESIGN

3.2.1 QUALITATIVE RESEARCH

This study is designed as an in-depth qualitative study. There are several reasons for the decision to use qualitative research methods in this study.

First, genetic counselling research often aims to ensure better service delivery to those affected by genetic conditions (Parker *et al.*, 2000). The assistance provided by genetics services often extends beyond the consultation with the patient, to families and communities. The psychosocial aspect of genetic counselling requires engagement by the counsellor with the emotional experience of the consultant. During training and clinical practice, genetic counselling students as well as genetic counsellors are often faced with sensitive and multifaceted issues regarding genetic conditions. Skills that are imperative to the genetic counsellor are similar to those needed to conduct interviews. Genetic counselling students are also required to present cases to their colleagues and ensure that their cases are followed-up which are valuable skills in qualitative research for evaluating and analysing data (MacFarlane, Veach & LeRoy, 2014). Qualitative research embodies a personable and humanistic approach to research that requires the researcher to engage with the experiences of the participants (Morse, 2012). In this way, genetic counselling embodies many of the same goals and qualitative research is well-suited to answering questions related to genetic conditions and hence this approach was utilised for this study.

Second, often in clinical research, the research question cannot be answered by numbers and statistics, comparisons and correlations. Instead the research focuses on understanding the emotions, perspectives and actions of individuals affected by a medical condition (Holloway, 2005). By identifying issues from the perspectives of the study participants, qualitative research aims to

understand the meaning and interpretation that they give to their experiences (Hennink, Hutter & Bailey, 2010). Qualitative research focuses on the experiences of those being studied, which allows the greatest promise of guiding clinical practice to make a difference in people's lives (Merriam, 2014).

Qualitative research adopts a naturalistic and interpretive approach in viewing a particular phenomenon. Naturally occurring social events may be understood through exploring the experiences, beliefs and perspectives of those influenced by the phenomena. Qualitative research reflects a dynamic social world in which the actions of those influenced by a phenomenon may be studied (Whitehead, 2012). Parents of deaf children have particular experiences that deserve to be illuminated (de Andrade, 2015), however in the South African setting, the experiences of parents of children with hereditary HL is unknown.

There is value and importance in showing interest in the circumstances and experiences of people (Hesse-Biber & Leavy, 2010). Qualitative research is people-centred; and aims to explore and understand the meaning that a person gives to a particular phenomenon. Therefore, the role of the qualitative researcher is to illuminate the intricacy of the phenomena that may be poorly understood (Silverman, 2015).

3.2.2 THE FRAMEWORK APPROACH TO QUALITATIVE DATA ANALYSIS

The framework approach, developed in the 1980's for social policy research has shown to be a useful tool for qualitative data analysis for healthcare research (Smith & Firth, 2011). This analytical method facilitates rigorous and transparent management of data by conducting analysis systematically through a four-step process. The thematic framework utilized by this method results in organisation of data into themes and related sub-themes (Spencer, Richie & O'Connor, 2003).

The initial step in this approach is familiarisation. This first step is crucial in building the foundation and structure of the analysis. In order to decide the themes and concepts which will be applied to the data, the researcher must fully immerse themselves in the raw data by listening to interview recordings, reading and re-reading transcripts. In this way, a rough list of key ideas may be developed. The second step is to identify a thematic framework with the aim of producing a detailed index of the data. Categories are developed for ease of exploration of the data, but remain focused on the aims and objectives of the study. Next, the researcher must annotate the transcripts according to the thematic framework (or index) developed in the previous step. This process of indexing, should be systematically applied to the whole data set such that links between categories may be visualised and can be grouped according to theme. The final step of this approach is to summarise the data by organising it into thematic charts according to the identified themes. In this way, charts will contain

similar concepts so the findings may be interpreted and explained, in the context of the study's objectives and the themes that have emerged (Spencer, Richie & O'Connor, 2003, Smith & Firth, 2011, Pope, Ziebland & Mays, 2000).

The framework approach was chosen for this study as it enables the researcher to explore the data and interpret the participants' experiences in a transparent fashion. It is also an ideal method for novice qualitative researchers as it allows for analysis to be conducted in a systematic manner, ensuring academic rigour (Spencer, Richie & O'Connor, 2003, Smith & Firth, 2011).

3.3 STUDY POPULATION AND SAMPLE

3.3.1 STUDY SETTINGS

Settings: Dominican School for Deaf Children (DSDC) and Red Cross War Memorial Children's Hospital (RCWMCH)

Initially, parents whose children attend the Dominican School for Deaf Children (DSDC) in Cape Town were recruited for the study. The DSDC is a boarding school hosting children affected by HL from various areas of the Western Cape Province in SA. Previous genetics outreach clinics have been conducted at the school and children were selected according to the obtained clinical information, and putative nonsyndromic genetic aetiology of their HL. Parents were then contacted for involvement in this study by letters sent out through the school nurse to their children, together with the consent form (Appendix A and B).

The DSDC advised the researcher that it may be difficult to recruit parents as many of the learners travel to and from home by public transport or that provided by the school, leaving the parents with little or no contact with the school. It was also generally advised that parents of these learners may further be preoccupied with concerns involving unemployment and socio-economic constraints and therefore be unable to attend interviews at the school, as many do not attend the school's regular meetings. Following a lack of engagement of parents from the school, it was decided to expand the population by recruiting parents through different means.

A clinical genetics service offered through the RCWMCH was chosen as the second setting from which parents were recruited for the study. Children with a variety of suspected and diagnosed genetic conditions are seen through this service, including those with HL where other causes had been excluded. An exhaustive search of the patient files was conducted, and several patients were identified as fitting. Parents were contacted if their child had been seen by a clinical geneticist at

RCWMCH and the child's HL was attributed to a nonsyndromic genetic aetiology. These parents, that fit the inclusion criteria (described below), were contacted for involvement via phone calls. The inclusion of this second group of parents was warranted as these parents already had contact with our genetics service, even though their child did not necessarily attend the aforementioned school for deaf children.

3.3.2 STUDY POPULATION

Parents, of either gender, were included according to the following criteria; they are the biological parent of the child and do not have a hearing impairment. The latter criterion was included for ease of communication during interviews.

In order to ensure that the participants' children have a possible nonsyndromic genetic cause for their HL, other inclusion criteria may have included that; they have more than one child with a hearing impairment, which alludes to the fact that the HL was not acquired; their children are the result of a consanguineous marriage, which results in an increased frequency of autosomal recessive deafness; and other causes for their child's HL have been excluded by a specialist.

Parents were excluded from this study if their child's HL can be attributed to an environmental cause, if the child has syndromic HL, or if they themselves have a hearing impairment.

3.3.3 SAMPLING METHOD

The method of sampling used in this study is purposive, and is used as a way of gathering the best information by selecting participants most likely to have the experience or expertise to provide valuable insights on the research topic as well as those that are accessible to be involved in the research (Marshall, 1996). Cases were chosen because they illustrate the feature in which we were interested. For purposive sampling to be effective, it demands that we critically consider factors of the population that are being studied and that the sample is chosen on this basis (Silverman, 2015). Purposive sampling is best utilised in smaller scale research and is guided by both time and available resources (Denscombe, 2010).

3.3.4 SAMPLE SIZE

Eleven parents were recruited for this study. Qualitative, unlike quantitative research, aims to collect rich data, and is less focused on quantity (Marshall, 1996). In qualitative research the interest is in the depth of the information gathered and the variation in the experience of each participant, and therefore a large sample size is usually impractical and unfavourable (Hennink, Hutter & Bailey, 2010).

There was no predetermined sample size in this study, rather sampling was carried out with the aim of reaching saturation. Variations in the data were assessed by initial analysis of the interviews to determine when, or if, saturation would be reached i.e. when the interviews did not shed further light on the phenomenon under investigation (Glaser & Strauss, 2009). When no further participants made themselves available for the study, by not consenting or not engaging with the initial letter or contact, a decision was made that no further participants would be recruited. This decision was based on the short time-frame with which this study needed completion, as well as the difficulties faced in contacting and recruiting potential participants, and was agreed upon by the researcher and the supervisors.

3.4 DATA COLLECTION

A short questionnaire was given to participants after consent was obtained, and completed prior to the interview (Appendix C). Closed-ended questions were used to collect demographic information that is brief, straightforward and uncontroversial, which was used subsequently as data for analysis (Denscombe, 2010).

Following collection of this information, the researcher conducted face-to-face interviews using a question guide (Appendix D). The question guide was designed based on information from existing literature on the subject of HL but also included questions that were focused on hereditary aspects which has not been previously described in this setting. The open-ended questions were carefully worded to ensure neutrality, to remove bias and any suggestive wording. However, there was room for flexibility by using probes and follow up questions as a means of eliciting data. The interview guide was reviewed by two independent supervisors to ensure validity. Pilot interviews were conducted to determine if there were flaws, weaknesses or limitations within the interview design. In this way, changes were made to the interview question guide prior to implementation of the study (Kvale, 2007). Methods of extracting detail, depth, vividness, nuance and richness from the interviews were studied, as well as techniques to show empathy and effectively hone the “art of hearing data” (Rubin & Rubin, 2011).

Most of the interviews took place at the school, for convenience for both parents and the researcher, and one interview was conducted at our office on the Health Sciences Campus, UCT. Permission was obtained by both the Western Cape Education Department (Appendix E) and the Dominican School for Deaf Children (Appendix F) so that this research could be conducted at the school. Interviews lend themselves to collection of data based on emotions, opinions and experiences and deal with more sensitive issues that may be based on privileged information (Denscombe, 2010, Mack *et al.*, 2005).

Therefore, interviews took place in a quiet, private room with few distractions, for further encouragement of disclosure.

When organising an appropriate time to meet, parents were asked if they preferred to have the interview conducted in a language other than English. One of the participants indicated that she required the interview be conducted in isiXhosa and another parent required a sign language interpreter. Parents whose preferred language was one other than English, were then administered a questionnaire in their preferred language, if it was requested, or otherwise walked through the questionnaire with an interpreter. Following this, should an interpreter have been required for the interview phase (if indicated by the participant), a lay interpreter assisted the researcher for the interview. Although it is not ideal to use a lay interpreter, both for confidentiality reasons and for accuracy in the interpretations, it is common practice in the South African setting to make use of lay persons due to a lack of available trained interpreters (Kilian, Swartz & Joska, 2010).

During the interviews, participants were asked a series of open-ended questions (Appendix D). Open-endedness allows participants to contribute as much information as they want to, fully expressing their viewpoints and experiences, but also allows the interviewer to ask probing questions as means of a follow-up (Turner, 2010). All of the sessions were conducted by researcher and audiotape-recorded. To ensure that subtle nuances and nonverbal interactions were not disregarded, the researcher took brief notes during the interviews to supplement the recordings.

3.5 DATA ANALYSIS

The interview recordings were transcribed verbatim for subsequent data analysis using Transcriptions software (developed by David Haselberger). A code was assigned to each participant to ensure confidentiality and any discernible information that may identify the participants was removed during transcription. Each of the participants were assigned an alphanumeric code (e.g. P1-P11) and when participants make reference to their child, using his or her name, their child was given a letter of the alphabet corresponding to their parent's numeric code (e.g. A-L). Notes were taken after interviews which included observations and reflection, as well as information on the date, time, setting and any external factors that may have influenced the interview. Recordings, notes and other documentation valuable to this research were kept under lock and key or on a password-protected computer, which was backed up to an external hard drive regularly.

Following transcription, the data was critically examined to identify themes using the framework approach. This analysis occurred simultaneously with data collection. Qualitative data analysis necessitates the researcher immerse themselves in the data so that they may be true to their

participants by analytically telling their stories within the context of the research question (Hennink, Hutter & Bailey, 2010). For this reason, the framework approach was used to systematically analyse the data.

Transcriptions were imported into NVivo10 (QSR International), a data management program. The use of a data analysis computer program aids in facilitating the manipulation of data, allows one to run more complex searches, visually explore codes generated and data security is maintained in a time effective manner. By carefully listening to the audio recordings and reading and rereading the transcriptions, early thoughts and ideas were generated. Engaging with the data is crucial for familiarity, understanding and insight that becomes the basis of the analysis (Silverman, 2015).

The steps described in 3.2.2 were employed as the data was analysed so that themes were developed according to the aims and objectives of this study. These themes were discussed with the study supervisors. Extracts of the data were chosen according to their relevance and representation of the emerging themes for presentation of the data as results of this study.

3.6 VALIDITY AND RELIABILITY

Qualitative research has been criticised for its lack of validity, whether the ways in which the researcher interprets an account accurately represents the phenomena to which it relates (Hammersley, 1992). The concept of validity - simply another word for truth - addresses the credibility of interpretations in qualitative research. Beyond the anecdotal approach to research, inferences made by the researcher need to be supported by the data, as well as rational in the context of previous research (Peräkylä, 2011). Validity of an inference is achieved when a researcher does not merely take a subjective approach to the data but rather scrutinizes the data, confronting deviant cases and triangulates the data so that valid interpretations can be made (Silverman, 2015). Methods for the treatment of data during analysis aim to produce more valid findings. These methods may include comprehensive data treatment where deviant cases are actively sought and anomalies are addressed to strengthen the analysis (Silverman, 2011). These methods were employed in this study, together with collaborative efforts from the researcher and the supervisory team, to critically examine the data to increase its validity. In order to eliminate research bias and ensure validity, independent supervisors examined three of the interviews individually and generated their own codes, which were then discussed to ensure that the researcher had interpreted the data accurately.

Reliability refers to the degree of consistency of data interpretations within their contexts and future replicability of research. This is achieved through transparency during the research process and a detailed methodology of data analysis – one of the main advantages of utilising the framework

approach. When an interview is audio-recorded and transcribed, there is room for weakening the reliability of the interpretation if subtle nuances, pauses or body language are not included in the explanation. Further strengthening of the reliability of data may include incorporation of observations or field notes into written transcripts, including interviewer questions into the presentation of data so that context may be understood by the reader, and transcribing verbatim (Silverman, 2015). All of these methods were employed in the current study to ensure that the findings are reliable.

3.7 ETHICAL CONSIDERATIONS

3.7.1 ETHICAL APPROVAL

Ethical approval was granted by the University of Cape Town's Faculty of Health Sciences Health Research Ethics Committee (FHS HREC/REF: 699/2015, Appendix G).

A deviation from the recruitment of participants was made in one case, a mother of a child who herself had a hearing impairment. Initially, this group was excluded as the researcher did not have adequate funding or access to a sign language interpreter. However, a lay interpreter, a teaching assistant at the school was used for this interview. A deviation form was completed as per the HREC recommendation (Appendix H) and permission was granted to include this data in the study.

3.7.2 CONSENT

Informed consent is essential in ensuring autonomy during research. By providing as much information as possible regarding the research, without deception, prospective participants have the opportunity to autonomously chose to participate (Silverman, 2015). To ensure that informed consent is achieved, information regarding the research must be explained in a manner that is understandable (which may require translation and further explanation of the consent form) and not forcefully persuasive (Mack *et al.*, 2005).

Following ethics approval, individuals meeting the inclusion criteria for this study were approached through their child's school or the genetics service at RCWMCH. Through letters or phone calls, the potential participants were explained the background and purpose of the study. Assurance was given that all of the information would be kept confidential. No methods of coercion were used, and interested parties were allowed to ask questions before written consent was obtained as well as consent for the interviews to be recorded.

3.7.3 CONFIDENTIALITY

The principle of confidentiality in research is paramount and every insurance was taken to protect the privacy of the participants recruited for this study and the confidentiality of their personal information (WMA, 2008).

Completed forms, questionnaires and audiotape recordings of the interviews were kept on a password-protected computer, or under lock and key, and the researcher alone had access. Each of the participants were assigned a code to avoid personally identifiable information appearing in transcripts or data. Children's names were removed from transcripts and reporting of the data. The minor-dissertation supervisor will retain the recordings and transcripts for a period of time post-graduation for dissemination of this work in peer reviewed journals.

3.7.4 RISKS AND BENEFITS TO PARTICIPANTS

Participants were made aware, before consent was taken, that this research did not aim to provide medical benefits to either the participants or their children. Some of the questions asked were of a sensitive manner and emotions that may have been evoked were a potential risk to participants in this study. These risks were minimised using culturally sensitive terminology, empathy and allowing the participant to terminate the interview and withdraw from the study at any time. Referral to a qualified genetic counsellor or psychologist was made if deemed necessary.

There is also a well-documented cathartic effect of research that involves allowing participants to openly share their stories. Qualitative interviewing may allow participants to confess difficult emotions that they usually wouldn't disclose (Lupton, 1998, Dickson-Swift *et al.*, 2007), and may be a benefit of involvement in this study.

Reimbursement was offered to all participants in acknowledgement that they had allocated time away from other obligations and may have incurred travel expenses. Had the participants withdrawn from the study or chosen to terminate the interview at any time, they were still reimbursed.

We committed to adhere to the ethical principles laid out in the Declaration of Helsinki, to promote and ensure respect for all human subjects and protect their health and their rights (WMA, 2008).

3.8 CHAPTER SUMMARY

In this chapter, the research methodology is presented. Qualitative methodology was chosen for this study and the reasons explained. The methods employed here are fully described as well as the ethical considerations. In the next chapter, the results and discussion will be presented.

CHAPTER 4: RESULTS AND DISCUSSION

4.1 CHAPTER INTRODUCTION

The fourth chapter of this study will include (1) an overview on how this study reached its objectives, (2) describe the socio-demographics of the research participants and (3) present and discuss the themes that were generated from the research, providing excerpts from the interview data to support the findings. Existing research accompanies the interpretation of the themes where appropriate.

4.2 REACHING THE RESEARCH OBJECTIVES

The first objective of this study was to recruit a group of parents of children with a putative nonsyndromic origin of their HL. Clinical notes compiled after consultation with the children, either conducted at the school on the genetics outreach or at the clinical genetics service offered at the RCWMCH, were scrutinised to select children whose HL was not attributed to an environmental or syndromic cause. Due to the fact that genetic testing for HL is largely uninformative in the South African population (Bosch *et al.*, 2014b), even those who had undergone connexin gene testing did not have a confirmed genetic diagnosis. For this reason, cases that had been seen by a genetics registrar or a consultant medical geneticist, and their HL was concluded to likely have a nonsyndromic aetiology, were flagged for involvement in the current study.

Of a potential 41 participants that were initially contacted for involvement, due to the potential nonsyndromic genetic aetiology of their child's HL, 11 interviews were conducted, representing a response rate of 27% of the potential participants identified.

Table 2: Number of participants recruited by the various recruitment methods and locations

Recruitment method	Number of potential participants	Letter returned/ contact made (number of participants)	Number of interviews conducted
First batch of letters to DSDC	18	8	6
Second batch of letters to DSDC	9	3	0
Records from RCWMCH Genetics Clinic	14	6	5
Total	41	17	11

4.3 SOCIO-DEMOGRAPHIC DATA OF PARTICIPANTS

Tables 3 and 4 below present the socio-demographic data of the participants:

Table 3: Participant characteristics

Category of participant		
<i>n</i>	11	
Mean age (SD)	35.8 (8.6)	
	<i>n</i>	%
Gender		
Male	0	0
Female	11	100
Race ²		
Black/African	3	27.3
Mixed/coloured	8	72.7
Home language ³		
English	5	45.5
Afrikaans	4	36.4
IsiXhosa	3	27.3
SASL	2	18.2
Marital status		
Single	5	45.5
Married	5	45.5
Engaged	1	9.1
Employment status		
Employed	7	63.6
Unemployed	4	36.4
Household monthly income (Rand)		

² Historically, the South African apartheid government influenced Deaf education by segregating learners according to race. In the 1930s, the DSDC was only allowed to accept 'coloured' learners. Since the demise of the apartheid regime, Deaf education in SA remains segregated and racial discrimination is particularly apparent in SASL studies (Reagan, Penn & Ogilvy, 2006).

³ Totals do add up to 100% as many of the participants are bilingual in their home environments.

0 - 3000	5	45.5
3000 - 7000	3	27.3
7000 - 10 000	2	18.2
> 10 000	1	9.1
Family history of HL		
Yes	3	27.3
No	8	72.7

SD – standard deviation

The total number of children (with and without HL) in the participants' families ranged from one to four; only one participant had two deaf children. The children were almost equal in gender across participants with an age range of 3 to 17 years and a mean of 9 years. The child's age of diagnosis, although not asked specifically in the administered questionnaire, was mentioned in each of the interviews and is included in table 4, so that the range may be visually depicted, with a mean age of diagnosis of 17 months. This data is summarised in table 4 below.

Table 4: Child characteristics

Category of child		
	n	11
Mean age (SD)	9,3 (4,6)	
Mean age at diagnosis (months) (SD)	16,7 (12,4)	
	n	%
Gender		
Male	6	54,5
Female	5	45,5

SD – standard deviation

Two of the participants were from the same family (P6 and P10), and their relationship is depicted in the pedigree above (figure 4). These participants, together with P9 (because of her previous child with HL), were the only participants with which there was a previous history of HL in the family. The children of the other participants were all the first family member to have been diagnosed with HL.

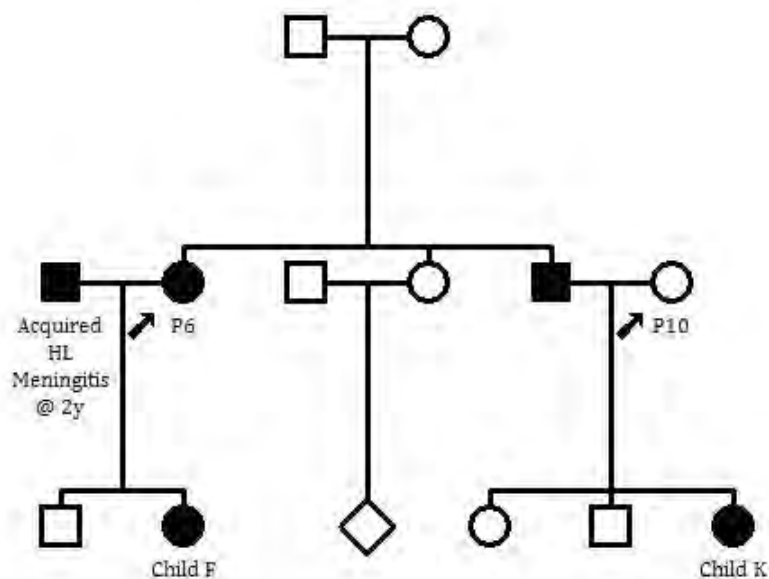


Figure 4: Pedigree showing the relationship between P6 and P10. Shaded circles (for females) / squares (for males) represent those with hearing loss (HL) in the family.

Eleven participants were interviewed in the study and their socio-demographic information, as well as some information on their child, are fully outlined in table 5 below. All of the participants were the mother of the child, and no fathers made themselves available for an interview. Almost half of the participants were single mothers (5/11), while the other half were either engaged or married (6/11) – not necessarily to the child’s father. The age of the participants ranged from 23 to 47 years, with a mean age of 35.8 years. All of the 11 participants lived in urban areas. The home language of participants included English, Afrikaans, isiXhosa and South African sign language (SASL), and some indicated that they are bilingual in their home environments. Just over half of the participants had finished high school (6/11), with only two having further tertiary education.

Table 5: Socio-demographic information of the participants and their child

Partici pant code	Sex	Age (years)	Home language	Sex of child	Age of child (years)	Child code	Child's age at diagnosis of HL (months)	Marital status	Number of children	Number of children with HL	Family history of HL
P1	F	29	Afrikaans	Male	5	A	9	Single	2	1	No
P2	F	44	isiXhosa	Male	17	B	36	Single	2	1	No
P3	F	26	English	Male	6	C	13	Engaged	2	1	No
P4	F	36	isiXhosa	Female	6	D	9	Single	2	1	No
P5	F	29	English/ Afrikaans	Female	6	E	20	Married	2	1	No
P6	F	42	SASL	Female	13	F	2	Married	2	1	Yes
P7	F	32	IsiXhosa/ SASL	Male	11	G	37	Single	2	1	No
P8	F	39	English	Male	12	H	30	Married	2	1	No
P9	F	47	Afrikaans	Male	8	J	15	Married	4	2	Yes
P10	F	47	English/ Afrikaans	Female	15	K	9	Married	3	1	Yes
P11	F	23	English	Female	3	L	4	Single	1	1	No

4.4 IDENTIFIED THEMES

The following themes were identified from thorough examination of the transcripts, as described in chapter 3. The themes and sub-themes that were identified are summarised in table 6 below.

Table 6: Themes and sub-themes generated

Theme	Sub-themes
The initial journey: shock, acceptance and adaptation	<ul style="list-style-type: none">- The loss of a “normal” child- Support and its role in acceptance and adaptation
Comprehension and understanding of the cause of their child’s hearing loss	<ul style="list-style-type: none">- “I don’t know where it comes from”- Guilt and blame- The need for answers
Role of the mother: the experience of being a parent of a deaf child	<ul style="list-style-type: none">- Practicalities and sacrifice- The need to protect
Being different: shame, stigma and the Deaf world	<ul style="list-style-type: none">- Stigma, shame and the rejection of pity- The Deaf world
Being normal: “Deaf people are people”	

4.5 THEME 1 - THE INITIAL JOURNEY: SHOCK, ACCEPTANCE AND ADAPTATION

The first theme identified focuses on the initial journey of becoming the parent of a child with HL. Participants experienced shock, denial, grief and confusion after the diagnosis of HL for their child and sought means of coping and adapting so that they make come to accept their new situation. In this first theme, the loss is not solely of the child’s hearing but can also be seen as the loss of what they had anticipated for their child.

4.5.1 THE LOSS OF A “NORMAL” CHILD

The diagnosis of HL for their child was a difficult experience for each participant in this study. Participant 11 explained, in the following quote, how unexpected a diagnosis of HL can be for a parent:

“No one really expect to have a deaf child or something wrong with their child.” (P11)

A participant described that without knowledge of previous history of HL in the family, the diagnosis can be a shock:

“It came as a shock because it was the first time in our family.” (P3)

This shock was coupled with heightened emotions and confusion for many of the participants. Becoming the parent of a deaf child has been described as a roller-coaster of experiences. It is undoubtedly a time of confusion and uncertainty as the parent begins to mourn the loss of their child’s hearing, the loss of a “normal” child (Bosteels, Van Hove & Vandebroek, 2012). This loss is of the life that they had expected, what they had hoped for and anticipated (Kurtzer-White & Luterman, 2003).

The response to this loss is related to both the meaning and context of the loss within the participants’ circumstance. Meaning may be influenced by perceived severity, implications for the future and cultural and familial beliefs and context is the environmental factors that may influence the response to the loss, such as support, financial constraints and coping resources (Lazarus & Folkman, 1984). In line with this model of how loss is perceived, one might think that a deaf parent, who has a first-hand understanding of what it means to be deaf, would respond to the diagnosis of HL in their child differently to that of a hearing parent. However, for both a hearing mother (P8) and a deaf mother (P6) in this study, the experience was emotional and confusing:

“having all these emotions and being so, so sad, asking questions about why my child” (P8)

“When she was born I didn’t know that she was deaf. When she is growing my mother told me that the child was deaf and I didn’t accept it, I was full of emotions when I hear my child was deaf.” (P6)

Denial is a common coping strategy in response to adverse events (Kübler-Ross, 1969). A few of the participants shared that they had experienced denial at some stage in their initial journey, as with participant 6’s above quote and as described by participant 10: “I was in a state ... I just couldn’t accept it”. These participants used denial as a temporary defence against the unexpected diagnosis. Denial that is temporary usually fades as one moves towards accepting the reality of the loss, or in this case the diagnosis (Kübler-Ross & Kessler, 2014). Chronic denial, however, may become pathological and maladaptive in its presentation if it persists, leading to adverse effects on the parent as well as the child (Kampfe, 1989, Kurtzer-White & Luterman, 2003).

Participant 4 experienced prolonged denial in response to her daughter's HL, denying the fact that her child's HL would persist and therefore avoided formal (medical) recognition of her child's HL:

"On that time that I did notice that my child had the problem with the deafness, but I didn't go to the doctor at that time, I was staying at home and thought that maybe it was something for the moment and was not going to continue." (P4)

Participants in this study described stages of acute grief as a reaction to the diagnosis, which formed part of their journey:

"I was trying now and thinking back of how I felt at the time and where we are now and I said to myself I don't want to be there again, never. Because of the way we felt." (P8)

Grief is a complex emotion that has prompted researchers to develop models to explore the way in which grief is experienced. Kübler-Ross's five stages of loss, originally based on death and dying, may be extrapolated to other settings in which grieving occurs. The well-known first stage is denial and isolation. As discussed above, participants in this study experienced this first stage of their grief for their child's HL. The second stage is anger, followed by bargaining, depression and acceptance (Kübler-Ross, 1969).

Bereavement models are often applied to the parental response to a child's loss of ability, in this case, the child's loss of hearing, but several differences exist between death and disability. A striking difference is that of time. As Hewson explains, "unlike the finality of death, the full impact of a loss of ability cannot be faced and resolved at any given point in time. The loss endures. New crises, and thus new loss responses, arise repeatedly in relation to the loss of ability" (Hewson, 1997). Therefore, other models have been developed that focus on the parental response to the loss of ability rather than assuming that the response will be one of grief that can be modelled within the pre-existing research on bereavement.

One such model is the three-phase cycle developed by Anderegg, Vergason and Smith which was primarily developed for special education teachers. Anderegg observed 130 parents from two support groups and developed a visual model to depict the stages and behaviours that parents of disabled children experienced. The first stage is confronting, where shock, denial, and blame/guilt are responses to the first suspicion or realisation of the child's condition. Parents may be confronted by new realities throughout their child's development and thus re-enter this phase of grief several times, for example a deaf child that develops some speech but regresses after entry into a school that focuses on sign language. The second stage revolves around adjustment, which may include depression, anger and bargaining as a parent adjusts to the diagnosis in their child. Parents may feel hopeless,

experience hostility towards others or themselves and may search for explanations and understanding. The final stage is that of adaptation, where parents need to realistically evaluate their lives for changes that can be made to better handle their child's condition. They may need to assess and adjust their role as a parent to realise the needs of their child and make plans for the future (Anderegg, Vergason & Smith, 1992).

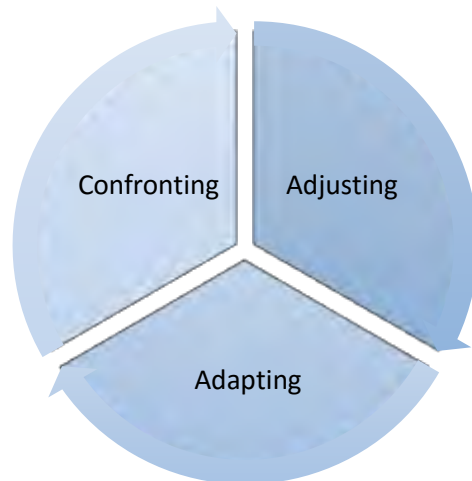


Figure 5: Grief process model of parents of children with disabilities. Adapted from Anderegg, Vergason & Smith (1992)

As shown in previous quotes in this section and subsequent quotes to follow, many of the participants experienced this cycle of confronting, adjusting and adapting. Some of the participants remained in one of the phases for extended periods and re-entry into certain phases was commonly described following the initial journey.

One case, however, that of P11 (a 23-year-old mother of a 3-year-old daughter) described how acceptance was easier for her, and that dwelling on the diagnosis was unnecessary for something that could not be changed:

“so for me when she was born it wasn't like a big deal that she was deaf or when I found out that she was deaf it wasn't a big deal because I knew there is nothing I can do about it so why fret about something that you can't change. So I just accepted it the way it is and carried on, what's the point in sulking?” (P11)

Participant 11's experience was somewhat different to that of most of the other participants, as her daughter reportedly underwent NBS soon after birth, as she explains in the following excerpt:

“So it’s standard procedure to do a hearing test at the MOU [midwife obstetric unit] so she was about, how old was she, a few days ... So when she was four months old they confirmed that she has a hearing loss and that it’s profound so we kind of expected it to be that way so we didn’t like take it very hard ... We had prepared ourselves already, from the beginning.”
(P11)

In this way, NBS mitigated the upset of the diagnosis for participant 11, which may have encouraged earlier adaptation to the diagnosis and acceptance of her daughter’s HL.

In the following sub-theme, means of support will be discussed as they relate to parents’ ability to accept their child’s HL.

4.5.2 SUPPORT AND ITS ROLE IN ACCEPTANCE AND ADAPTATION

Acceptance and adaptation to the diagnosis of deafness has been reported as one of the most challenging factors that families need to face (Ahlert & Greeff, 2012). Adaptation begins with the birth and subsequent diagnosis of a deaf child, through their development into adulthood. The process of adaptation is dynamic and is influenced by daily stressors and how they are appraised while raising a child with HL. Supportive resources may be utilised by families so that they may positively influence adaptation to these stressors (Bennett, Deluca & Allen, 1995, Singer & Irvin, 1989).

Several participants in this study described religion as means of strength and support as they came to accept and adapt to their child’s diagnosis of HL. These participants incorporated their religious beliefs into the process of acceptance; that their child is a blessing from God, that they were chosen by God because of their strength and that the support that they received from their church enabled them to come to terms with the diagnosis. This is illustrated in the following quotes:

“I feel like God gave me a child like E because he thinks I can handle it and I can handle it.” (P5)

“I have a good support system and you know they accept H even in church and they love him very much ... I am very fortunate to have that support system.” (P8)

This was also true of cases where HL was already in the family;

“I feel I had a daughter like that it’s not impossible it can maybe be another one. I said God is blessing me.” (P9)

“so but lot of people told me about she will be born deaf ... it is in their family [her husband’s family] so then I said well it’s God’s will, if it happens then I must just accept it” (P10)

Religion and faith are described as both a coping mechanism and means of support in this study. In previous research, religious beliefs and practices empowered parents of children with disabilities, providing hope and personal strength for adaptation (Bennett, Deluca & Allen, 1995, Kara & Harvey, 2016). Faith may be a form of continual, stable support that parents utilise while managing the daily challenges associated with their child's condition beyond the initial diagnosis.

In the South African setting, 8 of 10 black South Africans consult with traditional healers. People may approach traditional healers for a variety of health, social or emotional issues. Traditional healers remain an important aspect of SA's cultural diversity and many people consult these healers for ear-related concerns (de Andrade & Ross, 2005). An example of methods used to treat prelingual HL and the subsequent inability to speak by traditional healers is described in de Andrade's thesis, where caregivers explain how the traditional healer cut the tongue of the child so that they may speak (de Andrade, 2015). A similar account was described by participant 4 in this study, who consulted with alternative forms of healing:

"To me it was a shock because I didn't understand why my child isn't talking. I went to the church, that church it was the church for when you pray, you must believe to the thing that they say to you, but the person who said to me that she is going to talk, she cut my child here (indicating the web at the base of the tongue)." (P4)

Participant 4 was initially confused at the suspicion of HL in her daughter and experienced prolonged denial (as previously discussed). After failed attempts to understand the cause of the condition, she sought advice from a traditional healer through her church. Without support from the health care system following her daughter's diagnosis, she turned to a faith healer from her community in desperation. After the healer's 'treatment', and her daughter was still unable to speak, she was directed to the DSDC, which has been valuable in allowing her to better understand and come to terms with her daughter's condition. Sadly, however, her acceptance was difficult, as it meant giving up hope that her daughter would speak.

"Now I am telling myself that my child is going to stay like that, just recognising now that there is nothing that is going to happen for her." (P4)

Most of the participants described the time of diagnosis and the process of acceptance as a turbulent time, that was emotional and often requiring means of support that were not necessarily afforded to them at the time of diagnosis.

P2: *"I think that they was supposed to give counsel at that time. But I didn't have a counsel at that time because it was difficult, difficult. I think was supposed to give support, the counsellor but I didn't at that time."*

Interviewer: *"So you didn't get that counselling and you thought that you needed it?"*

P2: *"No, no. I was needed the counselling."*

The stress that parents experience following a diagnosis of HL has been previously studied, and is influenced by a number of factors including support, coping resources and resilience (Poon & Zaidman-Zait, 2013, Zaidman-Zait *et al.*, 2016, Ahlert & Greeff, 2012, Hintermair, 2006).

Social support systems may be formal or informal. Formal support encompasses support networks from professionals or institutions, such as audiologists, social services, schools or intervention centres (Rodrigo *et al.*, 2007). Literature suggests that audiologists may not necessarily be well positioned to provide newly diagnosed patients and their families with supportive counselling (Kurtzer-White & Luterman, 2003). Referral to other formal support services, such as psychotherapy or support groups specifically aimed at helping families in their positions may therefore be beneficial to families of newly diagnosed deaf children.

In addition to participant 2's quote previously, other participants in this study expressed that they would have wanted more support and information from formal support structures or experienced parents. This is demonstrated in the following quotes by participant 8 below:

Interviewer: *"What do you think would have been helpful?"*

P8: *"Like a support group with parents with deaf children that have gone through the process of finding out about your child because you know it was our first child it was difficult you feel silly, you don't even know how to change a nappy and now you must find out that your child is deaf, it's terrible and as a parent, as a mother ... Would have been nice, would have been very helpful because believe me, at a point we got very very lonely because here you have little baby, second baby and you have this deaf little child and ja⁴ the friends were there but they didn't really have that background of um working or dealing with deaf children and um it would have been nice to have that support, have a phone call from parents of deaf children, to just say you know how are you doing? Are you okay today? Don't worry, it will get better."*

Participant 8, in the above quote, describes not only how lonely she felt and overwhelmed by the diagnosis, but that she would have benefitted from other, experienced, parents' guidance and

⁴ "Ja" means yes in Afrikaans

support. Informal social support refers to relationships with family, friends and others in one's community. These supportive assets have the ability to influence how parents cope and adjust to their child's diagnosis, by alleviating some of the stressors related to having a deaf child (Lederberg & Golbach, 2002, Poon & Zaidman-Zait, 2013, Rodrigo *et al.*, 2007). The ability to overcome the initial crisis – the diagnosis – relies on parents' ability to integrate their experience into their home and family life, so that they may return to a level of functioning, ultimately acceptance. The role that both formal and informal support play in this process is crucial (Walsh, 2002).

Over time, and at the time of the interviews, most of the participants thought back on their experience of the diagnosis, reflecting on how difficult it may have been but also how after time, how they reached acceptance and have adapted to their child's condition, again relating to the diagnostic journey that these parents faced. The quotes below illustrate some of these experiences:

"It's very difficult. You know, because but now I have started getting used to it. In the beginning I was like... how do I handle it? How will I communicate with him? But now, it is not so difficult."
(P1)

"So you feel sad, but sometimes when you learn and then you go into it and you learn more and you think, this is all ok, this is like not really a big thing actually." (P1)

"We made it. And when I think back oh my goodness how did we do this. He is growing up so fast and how did we do this, how did we get through this. Yes, it's been quite a journey." (P8)

This first theme encompasses the initial journey that participants faced after diagnosis of HL in their child. Each experience is distinct and depends on a parent's ability to confront, adjust and adapt to their new reality. Accepting their child's diagnosis is shaped and assisted by supportive entities, such as their religious beliefs, formal supportive services and informal support. As discussed in this theme, parents may go through stages of grief that will be influenced by external stressors, context and understanding. In the following theme, the hereditary cause of the participants' children's HL will be explored.

4.6 THEME 2 - COMPREHENSION AND UNDERSTANDING OF THE CAUSE OF THEIR CHILD'S HEARING LOSS

The time from diagnosis to acceptance was a journey unique to each participant. Many of the questions that they faced regarding the cause of their child's deafness remain unanswered. The second theme identified concentrates on the third objective of this study; parents' perceptions and understanding of the cause of their child's HL. By exploring the causality that parents attributed to

their child's HL, discussion in this theme involves parents' understanding and misconceptions regarding genetics and recurrence risk, opinions concerning genetic testing and feelings of guilt and blame. By understanding how parents perceived the cause of the child's HL, the need for services, such as genetic testing and genetic counselling, that provide parents with an explanation of the mechanism behind hereditary HL, was determined. This is the first study in the South African setting where the hereditary mechanism behind prelingual HL has been explored.

4.6.1 "I DON'T KNOW WHERE IT COMES FROM"

The first sub-theme encompasses parents' knowledge of the aetiology of their child's HL. The title of this theme, although not true of all parents, shows that most of the parents who were interviewed did not know or were unsure of what caused their child's HL. Often this confusion left parents feeling helpless and looking for answers.

"I don't know where my child's deafness is coming from. So that's why I don't actually like to talk about it very much because I don't know where it comes from." (P1)

When participants were asked whether they thought that their child's HL could potentially be explained by a genetic cause, several parents questioned this as there was no one else in the family:

"I don't think, because in my family there is no one. We don't have no one. He is the first one actually who is deaf." (P3)

Families where there is no previous history of deafness are usually unsuspecting of the diagnosis. As with most recessively inherited HL, there is often no history of HL in the family or extended family. Thus, parents may present with information seeking behaviour and struggle without a definitive cause to attribute to their child's HL (Steinberg *et al.*, 2007, Parker *et al.*, 2000).

Two of the participants attributed stress during pregnancy as the cause of their child's HL:

"The time I was pregnant I was have a problem because my boyfriend (pause) we quarrelled all the time ... So I think maybe the time I was pregnant I had a problem ... I was alone at the time I was pregnant ... So maybe was stress too much." (P2)

"If maybe stress can cause the deafness to the child then maybe it can be, because the time I was pregnant ... I was very stress." (P7)

As with previous research (Steinberg *et al.*, 2007), when there was a family history of HL, as with participants 6 and 10, these parents are able to correctly identify their child's HL as hereditary.

“They explained that she was deaf but I knew it was in the family.” (P6)

Interviewer: *“What do you think caused J’s deafness?”*

P10: *“For me it’s because of the genetics.”*

Interestingly, participant 9 who has two deaf children, was unable to identify the potential hereditary component of her children’s HL. She attributed her eldest daughter’s HL to an accident that happened in the home involving cement poured over her face at an early age, and was unsure of what had caused her second child to also have HL.

Although public awareness of genetics is increasing, these parents may have different understandings, preconceived ideas and misconceptions about hereditary traits. Words and concepts such as genetics, genes and DNA have become popularised through media and television but much of it remains poorly understood (Greenberg *et al.*, 2012). Given that most basic genetics concepts are only taught in Grade 12 Life Sciences (Department of Basic Education, 2011), it is no wonder that many of these concepts are difficult to understand and retain for the majority of the public. As described in chapter 1, marked social differences and access to higher education in SA means that there is less dissemination and comprehension of genetics concepts resulting in a large proportion of families affected by a genetic disorder who are unable to explain why this is happening in their family (Greenberg *et al.*, 2012). As only 6 of the 11 participants finished high school, and the fact that Life Sciences as a subject is not mandatory, many of the participants in this study may not have had access to basic genetic concepts and were therefore unable to identify this as a potential cause of their child’s HL.

4.6.2 GUILT AND BLAME

When the terms genetic and hereditary are introduced, parents may ascribe personal responsibility to their child’s condition, even if it is out of their control. Feeling personally responsible often provides parents with the illusion of control (Kessler, Kessler & Ward, 1984). An expected event, such as the diagnosis of HL in a child, has the ability to shatter a parent’s world of normalcy and understanding. Parents may grapple with the existential question of “why did this happen?”, leading to a search for the agent responsible, which can often mean assuming personal blame or feelings of guilt (Weil, 2000).

Guilt is defined as “an unpleasant emotion accompanied by beliefs that one should have thought, felt, or acted differently” (Benetti-McQuoid & Bursik, 2005). Self-blame is further defined as holding oneself responsible for “causing, contributing to, or exasperating” a particular situation (Moses, 2010).

Several participants, as described in the above sub-theme, attributed their child's HL to something that they may have done during pregnancy, such as stress, or when unable to find a cause for their child's HL, one of the participants questioned whether it was her fault, experiencing self-blame:

"I did blame myself because during when I was giving birth to E I did like give up, because I couldn't push anymore." (P5)

Interestingly, one of the participants explained that knowing there is genetic cause for her child's HL, would ease guilt, as no one would be to blame.

Interviewer: *"You said before that you felt guilty."*

P5: *"Ja because I did think it was my fault because I was like giving birth and I was giving up because my body couldn't take it anymore, that's why I was feeling guilty."*

Interviewer: *"But you wouldn't feel guilty if it was a genetic cause?"*

P5: *"No, no."*

Interviewer: *"Why not?"*

P5: *"I don't know (pause) probably because I will feel it is not my fault and I don't want to blame anybody at all."*

In this way, P5 is saying that she has no control over what genetic traits she passes on to her child, and that with a definitive genetic aetiology, she would no longer blame herself for her child's HL. Relatedly, P8 explains that blame is counterproductive to acceptance and adaption to having a deaf child:

"From my point of view, you can't help for your genetics I think, you didn't form yourself and if there are two people who are put together with some genetic default and you have a deaf child and you are the cause, I don't think that you can blame the other because you didn't know but um you know if there is if there was deafness in the family maybe you could have had yourself tested, things like that you know, precautions." (P8)

In the last section of the above quote, participant 8 highlighted that if there is previous family history of HL and there was a test available, testing would be helpful to determine the risk of passing on the HL and that precautions could possibly be taken. Genetic testing was an important aspect discussed by many participants and will be considered in the following sub-theme 3.6.3.

A study by James *et al.* (2006) looked at the mode of inheritance of certain genetic conditions and how it influenced guilt and blame, as well as stigma and understanding of recurrence risk. One hundred and ten individuals from 49 families (37 X-linked and 12 autosomal recessive) completed

surveys with regards to how mode of inheritance has influenced aspects of their family life. It was found that X-linked mothers, in particular, experienced feelings of guilt as well as self-blame in response to their child's condition and X-linked fathers were more likely to blame the other parent. Mothers of children with autosomal recessive conditions experienced significantly less guilt than their X-linked counterparts. These attributions of blame and parental feelings of guilt are important factors to take into consideration when dealing with genetic conditions. Unresolved feelings of guilt and blame may be harmful to parents' relationships with each other as well as parent-child relationships (James *et al.*, 2006).

The role of genetic counselling to address and manage blame and guilt in hereditary HL has been previously examined. Parker *et al.* conducted a survey, administered to parents of children with HL, to determine what parents and families wanted from a clinical genetics service. Of the respondents (n=130), 12.3% expressed interest in their genetics provider addressing issues of self-blame (Parker *et al.*, 2000).

Interestingly, prior to genetic testing, parents of children with HL had conflicting views regarding the value of genetic testing in reducing feelings of personal responsibility and blame. Some saying that it would lessen these feelings of guilt, while others felt that they would blame themselves more if a genetic cause of their child's HL was identified (Steinberg *et al.*, 2007).

Genetic counsellors are equipped to deal with parental feelings of guilt and blame by allowing them to openly express these emotions, normalising their feelings of guilt and are able to help parents to come to terms with the fact that they are not responsible for their child's genetic condition (Weil, 2000).

4.6.3 THE NEED FOR ANSWERS

Genetic testing for HL in the South African setting is under scrutiny, as previous research has shown that testing for the common genetic mutations in most other populations, has been largely uninformative in the South African population (Bosch *et al.*, 2014b, Kabahuma *et al.*, 2011). Rich diversity and genetic heterogeneity means that further research into the genetic basis of HL in African populations is crucial before testing will be useful and widely available. At present, the National Health Laboratory Services in SA have testing, via full coding region sequencing, available for three genes responsible for NSHL, namely connexin 26, 30 and 43 (J. Grobbelaar, personal communication, 2016).

Four of the participants' children reportedly underwent previous genetic testing, either through Tygerberg Hospital or RCWMCH. The perceived usefulness of the test and the results that were returned, is described by participant 11 below:

P11: *"She did a genetic test at Tygerberg Hospital, and they did one here [RCWMCH]."*

Interviewer: *"And did you get the results back?"*

P11: *"Yes, but they say that can't test, I don't know what you call it, all the genes. They said the most common deaf genes they tested."*

Interviewer: *"Did you get a result back?"*

P11: *"No that's the thing, they can't figure out where her deafness comes from. Because it might be in one of her other genes they can't test for, or might just be out of the blue. Or maybe it's like I have 2% of deaf genes and her father have 3% and those mix and then becomes 5%."*

The above example further illustrates the lack of understanding regarding genetics concepts and how the results of the genetic test were fed back to the family. This highlights the importance of genetic counselling to manage misconceptions, provide families with an appropriate explanation of genetic test results and manage the uncertainty that is associated with highly heterogeneous conditions, like HL. Participant 2 said that the test came back normal, which further fuelled her confusion, as she may not have been properly informed about the limitations of testing for hereditary HL, especially in the South African setting.

Without a definitive diagnosis, the need for answers prevailed, and all but one of the participants expressed interest in pursuing genetic testing. When probed about why they thought that the testing would be useful, participants explained that they wanted to "learn more" (P1), that it would provide "peace of mind" (P3), and that to know where their child's HL comes from, would advise how it may be handled in the future, for their child's future offspring, and for the risk of recurrence.

Genetic counselling for HL is particularly valuable by providing aetiological information and dispelling misconceptions. It may be helpful for parents to relieve guilt (as previously described) and enhance psychosocial wellbeing as well as providing appropriate information regarding recurrence (Alford *et al.*, 2014).

In this study, the participants were asked what they thought the chance was for themselves to have another deaf child, or for their child one day to have a child who is also deaf. Some of the participants had interesting insights into recurrence:

“I was worried that time when I was pregnant, I was worried about that also. But the doctor said if there was anything then there was like a 50/50 chance.” (P5)

“I spoke to my eldest [children], said to them, you must understand now it is running through the family so I don’t think you must have more children if you are not ready for deaf child. But the doctor said at Red Cross that it is mostly the girls. Yes. Running in the girls.” (P10)

Many aspects influence how recurrence risks are perceived. Participant 10’s misconception about girls being the ones to have HL is interesting as her husband is deaf. The proposed inheritance pattern of HL for P10’s husband and daughter is autosomal dominant, which would mean that for every pregnancy of hers the chance to have a child with HL would be 50%. If her older children are hearing, and no other factors are taken into account (such as the genetic status of their partners), then they would not have a child with HL. This again illustrates the importance of appropriate genetic counselling for this family, in order to dispel their misconceptions regarding recurrence, so that it does not influence reproductive choices.

One participant (P8) stated that her and her husband had decided not to have more children due to a lack of understanding regarding the risk for recurrence. In this way, misunderstanding affected her reproductive choices, which may have been resolved if the family had undergone genetic counselling to discuss their risks and avenues that could have been taken should they wish to have another child but not want to take the chance of having another child with HL.

The value of genetic counselling in demystifying parents’ beliefs about the cause of their child’s HL was investigated in Portugal by Rodrigues *et al.* (2013). Forty-four parents of children with prelingual NSHL caused by mutations in *GJB2* took surveys before and after a genetic counselling session. Prior to the counselling session, only 13.6% of the parents knew what had caused their child’s HL, compared to 84.1% post-counselling. Additionally, 95.5% of the parents felt that their expectations had been met in the session and 93.2% were able to correctly remember the recurrence risk (Rodrigues *et al.*, 2013). This study, although performed on a relatively small sample size, does highlight the important role that genetic counselling may play in assisting parents to understand the mechanism behind their child’s HL as well as the risk for recurrence.

In contrast to P8’s previous quote about concerns for recurrence, P6 (a deaf mother), hoped for a deaf grandchild one day:

P6: “Sometimes I think about F and she also asks me, maybe one day she will also have a deaf child.”

Interviewer: *“Do you think it is better in the family to have a deaf child or to have a hearing child?”*

P6: *“Deaf. Because it is expanding the family.”*

When deafness is familial, it is usually assumed that the child’s HL is similar and therefore parents do not pursue genetic testing (Kaimal *et al.*, 2007), this was not the case for the deaf mother in this study. She was interested in testing for its potential to identify a child with HL, which would be favourable. This is similar to a popularised story about a lesbian couple in the USA that chose to have a deaf child by selecting a sperm donor based on his family history of congenital HL. This choice, made infamous by the media, was the couple’s way of including their child in the Deaf culture that made up a substantial part of their identities (Spriggs, 2002).

Some may see the deaf trait as something unfavourable, but a large part of the Deaf community wishes to make their population larger and will seek genetic counselling and advice on how to use genetic testing, such as prenatal diagnosis or preimplantation genetic diagnosis, to ensure that they pass on the deaf trait to their offspring (Emery, Middleton & Turner, 2010, Middleton, Hewison & Mueller, 2001). The concept of the Deaf world will be further discussed in theme 4.

4.7 THEME 3 – ROLE OF THE MOTHER: THE EXPERIENCE OF BEING A PARENT OF A DEAF CHILD

The third theme addresses the second objective of this study, where the research aimed to explore the experience of parents who have a child with hereditary HL in various areas of burden. Although an assumption, that their experiences were burdensome, the participants described moments of both difficulties in having a child with HL, but also how their lives had been altered in a positive way. Becoming the parent of a deaf child influenced the participants’ lives in many aspects, from finding new methods of communication, dealing with the practicalities of having a child with different needs, to the additional worries and need to protect their deaf child. The sub-themes below speak to the role of the mother, as a parent, and how it has been influenced by the diagnosis of HL in their child.

4.7.1 PRACTICALITIES AND SACRIFICE

As reviewed by Jackson and Turnbull (2004), a diagnosis of HL can have far-reaching implications on family life. The impact that HL has on family resources and the practical implications, were evident in this study. These implications included communication difficulties, the need to stay home to care for

the child, absenteeism at work or resignation so that they could attend multiple clinic appointments, the struggle to access health care as well as financial insecurities.

Difficulties with communication came up in all of the interviews except for P6, who is also deaf and shares a common language with her child. Although the DSDC offers sign language classes for parents and families to attend on Saturday mornings, many children were weekly boarders, as previously described, and parents explained difficulties in attending the weekend classes due to transport or financial difficulties. This consequently influenced how well a parent could communicate with their child, and thus the relationship that they have with the child.

“It is difficult for me to come (to the sign language classes at the school) because I am struggling with the money. I am supposed to come on Friday to fetch her and then I can’t come on Saturday again. The money is the problem. And also I am staying far.” (P4)

For most of the participants interviewed, SASL is their second or third language that has needed to be acquired later in life, which means it will never be as natural as their first spoken language. Learning a new language as an adult is both time-consuming and can challenge a parent’s sense of competence (McKee, 2006). This may lead some parents to feel insecure about their lack of skill in communicating effectively with their child which will have reaching ramifications on effective parenting (Mason & Mason, 2007). Participant 7 showed some of her insecurity in using sign language with her 11-year-old son who is proficient:

“I come for lessons here. He is trying to teach me but if I don’t understand quickly he tells me that I am dumb.” (P7)

Communication is understandably a difficulty that many parents of children with HL face. Parents are usually the ones to first introduce speech to their children. Without being able to hear, many deaf children will go without a means of communicating with their families until they have a hearing device to aid in any residual hearing or are able to attend a school where they may learn how to communicate through sign language (McKee, 2006). Several of the participants in this study described how the roles of language acquisition are reversed. Their child now becomes the teacher. This is illustrated in the following quotes:

“He don’t understand me, I don’t understand him at that time but now we get together, it has been our second year now that we started sign language. But we not there yet on a professional level but we still (pause) I am learning from him, he is teaching us.” (P3)

“At home sometimes when she wants something and I don’t understand it then she will show me what it means in sign and I will remember it.” (P5)

Different burdens exist for each family when they are faced with a child with HL. Transport issues and financial barriers were extenuating factors that influenced how many of the participants dealt with the role of being a parent of a deaf child. Eight of the eleven participants have a gross family monthly income of less than R7000. Resources need to be divided according, as per Maslow’s hierarchy of needs (1970), which means that often providing food and heat for the family is of greater importance than expensive therapeutic interventions (such as cochlear implants) or burdensome travel costs to and from clinic appointments and school events (Greenberg *et al.*, 2012). The financial status of some of the participants was remarked upon to be a cause of stress, and is shown in the following quote:

“Our financial status isn’t so good ... we have been struggling for a long time. It’s the first time in a very long time that we are struggling actually struggling for such a long period of time and it’s not nice knowing I have a child I have to take care of and financial status is like going down.” (P11)

This financial burden may be further exasperated by parents needing to forgo working so that they may care for their child with HL. This was described by two of the participants below:

“...because I have to go to hospitals in and out with E... and she has to see the speech therapist like every Friday, and then that was becoming an issue at work so then I go and resign to sort her out before I go to work again.” (P5)

“I am not working because I have to look after her” (P11)

The issue of transport and financial barriers that make access to schooling difficult may be rectified if schools for deaf children were more widely available. One mother, P5, explains that even though there is a need for schools for the deaf throughout the greater Cape Town area, all of the schools are found close to the city:

“If you ask the principal how many kids are living inside here (boarders), most of the kids are living on that side of the world, Strand side, there is no school for the deaf child that side. Everything is here Cape Town side.” (P5)

From the participants’ accounts, it can be seen that the practicalities of having a child with HL are sizeable. Given that just under half of the participants were single mothers, the practical implications of caring for a child with additional needs fell solely on them, to provide for their children, make time to learn a new language and ultimately play the role of sole provider and carer for their deaf child.

Although men are more actively involved in parenting today, there are still gender stereotypes and gender-related practices globally that see women as continuing to be the primary caregivers for their children (Harvey, 2015). This was evident in the current study, as only mothers made themselves available for interviews.

The role of the mother, as the primary caregiver, means that many of the mothers in this study are laden with the additional care and resources that they need to provide for their child with HL. As seen by previous quotes, these mothers needed to make sacrifices in their lives in terms of finances and employment in order to care for their deaf child but further sacrifices had also been made by participants in terms of their relationships and social lives. This is illustrated by participant 11 below:

“For me it’s very time consuming and all the responsibilities, having a child on its own like, just a child without special needs is already hard, it takes up all your time and then having a child with special needs you have to go here, there, you have appointments all the time and you don’t really have time for anything else.” (P11)

Two participants described feeling alone, that they had to face the difficulties of having a deaf child without means of support:

“I do everything on my own mostly, we have everyone at the school, the teachers, but I’m alone.” (P1)

“There are times when I just feel why me? You get so depressed it feels like you are alone. Especially during the week and then I will just go into my room, lock myself up from everyone and I just started crying silent.” (P3)

One mother, when asked what would have been helpful, explained that a support group with parents going through similar experiences, would have aided her in not feeling so alone and helpless:

Interviewer: *“What do you think would have been helpful?”*

P8: *“Like a support group with parents with deaf children that have gone through the process of finding out about your child because you know it was our first child it was difficult you feel silly, you don’t even know how to change a nappy and now you must find out that your child is deaf, it’s terrible and as a parent, as a mother.”*

Participant 8’s insecurities about whether she could effectively deal with her child’s diagnosis was aided by support that she subsequently received from both friends, family and the church (as described in chapter 4.5.2). Through fundraising and a generous donation, her son had bilateral cochlear implants placed, the only child in the sample who was afforded this chance for spoken

language. This had an extremely positive influence on the family and for P8. She describes her ability and joy to hear her son's voice and not miss out on milestones as he becomes a teenager:

"We had the choice to raise the money for the cochlears or sending him to um the deaf school and we chose to raise the money for the cochlears. And I am so happy that we did that! I um I couldn't imagine not hearing his voice ... Do you hear that H's voice is starting to break? Oh my goodness. And if he was signing, you know we wouldn't have heard that, so I am just so happy that he is speaking, that he talks." (P8)

Given that cochlear implants are not covered by the public health system in SA, many children who would benefit from the technology are not afforded this opportunity. Not only is the initial assessment and implantation costly, but patients need access to a specialist facility and the long-term costs of maintenance and rehabilitation mean that most children with HL in the South African setting cannot access this specialised intervention (Kerr, Tuomi & Müller, 2012).

The practical demands of caring for a child with additional needs are sizeable. Increased time demands, worries about security and limited access to supportive services have been shown previously to influence how families deal with a deaf child (Jackson, Traub & Turnbull, 2008) and are apparent in this sample as well. Additionally, there are added pressures associated with financial security, access to schooling and health care, which further intensify the difficulties that a parent of a deaf child faces in the South African setting.

4.7.2 THE NEED TO PROTECT

Parenting a deaf child is unique, in the sense that it is associated with challenges that may be different to that of a hearing child. A striking factor that surfaced in several of the interviews was a need to protect. A mother's need to protect her child is undoubtedly hardwired, but extenuating factors, such as a child's inability to hear, may influence the extent to which a mother worries and therefore feels she needs to protect her child.

A few of the participants resided in informal settlements on the outskirts of urban areas in Cape Town. These shack-based settlements are generally overpopulated, with more than 16.4% of South Africans residing in informal settlements around the country (Albertyn, 2006). Two of the participants in this study, regaled stories about how their living situations added stress and worry about their child's safety, as illustrated by participant 7 below:

"And sometimes I am scared to leave him alone, maybe I am going to work, then no one to stay with him, that is the other challenge because I think if they you know we stay there in the

informal settlement and I think what if what if it burns and then he can't hear maybe people they are shouting to stay away from the houses because there is a fire.” (P7)

Another participant is reminded of a traumatic experience when her child fell victim to an accident due to his HL:

P2: *“And the time he was involved in the car accident, B he he wants friends, they were play together. So it's more special the way they play maybe on the road, I say you mustn't go to the road because you deaf, if the cars coming you didn't hear. Your friends they hear the sound but they can't say B, must go! Only is the problem, the time the truck was coming B he didn't hear, he just (claps hands together) and B was ride a bicycle and I don't want, that time I didn't bought a bicycle because of deafness but he borrowed from his friend a bicycle and just (claps) the car accident. So it was a problem ... And I hear that sound the time he (claps) hit my son. Always even if I, even now, if I hear the sound, like the car maybe is just running too much, I saw in my mind that day, the day that B was involved in the accident.”*

Interviewer: *“You remember that time...”*

P2: *“Every day. When I hear that sound.”*

Participant 5 expressed her fear of being unable to protect her child, especially given that her child is a boarder at the school. Although her child is only 6-years-old, she understands that her daughter belongs to a vulnerable population, and she needs to be protected against sexual advances, with the fear that she may be taken advantage of, as seen previously in a study regarding parents' anxieties related to HIV/AIDS for their deaf children (Mall & Swartz, 2011). This is further illustrated by participant 10's following quote:

“I want her to have a happy life and that people won't take advantage.” (P10)

Given participant 10's experience with deafness, from her husband's family, she was aware that a child or adult with HL may be taken advantage of. She insists that her other children, who are older than her deaf daughter, look after her.

“I know my friends, my husband's friends, they are both deaf, the dad and the mom, but their children is normal. But those children are taking advantage of that two people (gasp) so I thank God that I am the one who can hear.” (P10)

In the above quote, participant 10 is both acutely aware that hearing children of her husband's deaf friends are guilty of exploitation and taking advantage of their deaf parents. Mistreatment of deaf persons is a longstanding problem. Deaf women in particular are exposed to greater acts of violence,

abuse and exploitation, both in the workplace and at home (Kiyaga & Moores, 2003). Sexual abuse of deaf children and adults has been reported on for decades (Kvam, 2004, Sullivan, Vernon & Scanlan, 1987) and the dangers of HIV/AIDS transmission, particularly in the South African setting, is a worry for parents and families of deaf individuals (Mall & Swartz, 2011). Due to language barriers, cultural differences and the paucity of information that is available to deaf youths, indulging in high risk sexual behaviour is a valid concern of parents both in this study and in previous research in South Africa (Mall, 2012).

Parenting a child is no small feat but the additional challenges that parents of children with HL face, have been discussed in this theme. These challenges include increased practical demands and needing to make sacrifices in various areas of life in order to care for a deaf child. Furthermore, the need to protect the deaf child is paramount for these parents as they are aware of the dangers that may arise in response to their HL.

4.8 THEME 4 - BEING DIFFERENT: SHAME, STIGMA AND THE DEAF WORLD

The fourth theme identified revolved around the idea that being deaf meant being different. Participants were confronted by their child's "difference" through stigma and shame. The concept of a Deaf world is explored as it related to the participants' views of how their child experiences their HL.

4.8.1 STIGMA, SHAME AND THE REJECTION OF PITY

Hearing loss is not the only condition with which stigma is associated. Previous studies have examined stigma in relation to other disabilities (Green, 2003) and conditions that have a genetic aetiology and those that may be caused by non-genetic factors (Sankar *et al.*, 2006). Stigmatisation, as a social phenomenon, begins when a person is labelled by a feature that sets them apart from others, in this case being deaf. Felt stigma refers to the experience of the person who has been labelled and subsequently ostracised (Scambler, 1998).

Stigma, however, does not only affect those who possess the stigmatising feature but tends to spread to others who have a relationship with the stigmatised person. This is referred to as courtesy stigma, and can be felt by parents, friends and family members of those with disabilities and differences that fall victim to stigmatisation (Goffman, 1963).

Most of the participants made mention to times that they felt their child or their family member (in the case of those with familial HL) had experienced stigmatisation due to their HL. This may have been

in the form of ridicule, as participant 7 explains below, or as participant 11 says “it is like deaf people are excluded because nobody else can communicate with them.”

“Sometimes when like he like to visit to my sister then G doesn’t want to go alone, so every time I have to go with him because he says yoh I can’t go alone because I can’t talk, they going to laugh at me.” (P7)

Children as well as adults are not only the victims of stigma but may also be the perpetrators as participant 10 describes:

Interviewer: *“Do you ever feel anyone treats you differently in the community or in society in general because you have a deaf family?”*

P10: *“Yes, the first time we moved to our home the people I mean coloured people really, they were teasing. They will call him like a dommie⁵ but I am so strict, I am very um um how do I say, I won’t allow it then they will call him like dommie.”*

P10: *“Oh I hate it. Then I will always tell them, um what if you have a disability and someone called you that it’s not nice, I mean he’s got a name.”*

“[The children in the neighbourhood] are shouting in J’s ears, they are mocking her, she’s deaf and then she would come and cry, she had that feeling.” (P10)

In response to the courtesy stigma that the participants felt, there was an overwhelming sense of rejection of any pity that may be associated with their child’s stigma. As with Kara and Harvey’s study of black South African mothers of children with HL (2016), parents took a definite disliking to forms of external pity. This is further illustrated in this study by the following quotes by participant 5:

“It’s just... sometimes you feel like you want to do more. Especially for a child who is deaf or a person who is deaf. But now nowadays people is ... feeling sorry but they don’t want to hear I’m sorry because you are deaf, like there is a reason for everything.” (P5)

“When we going out people is like looking or they come to you to say oh I’m sorry your child can’t talk then I want to snap and say don’t say sorry.” (P5)

As discussed by Green (2003), parents’ perceived stigma may lead to emotional distress. This may be dealt with in a number of ways. Firstly, parents may lash out, getting angry at the pity that others may

⁵ “Dommie” means the dumb/stupid one in Afrikaans

feel for their situation (as shown in the above quote). Secondly, they may find it necessary to deal with the burden of stigma by educating people about their child's condition, as participant 8 explains:

"Sometimes the grown-ups ask questions like is there something wrong with his brain and I will say no it's a cochlear device." (P8)

And thirdly, parents and the affected child may react with secrecy, which may be directly related to shame (Green, 2003). Deafness can be viewed as an invisible disability. When a deaf person does not use a hearing aid or conceals it from onlookers, one may not look twice or think that there is something "wrong" or different about them. In this way, deaf children and adults may avoid stigmatisation through secrecy but it also emphasises the shame that they experience in relation to their condition.

"You will see he wears a cap [to cover his cochlear implants]. He um he doesn't like it when people stare at him in the malls, he hates it... He is growing up and he is noticing, he is becoming more mature, noticing people staring at him and he will put his hoodie on, he will put his cap on because he is becoming more aware of people noticing him." (P8)

"[My husband] is also very shy where it comes to wearing the hearing aid also ... in the shop I asked him what he want but in sign language and it seems like he don't want to respond like I mustn't talk in front of people" (P10)

In the above quotes, shame is experienced by the deaf individual, both a child and an adult. O'Connell's autoethnography provides a narrative of what it is like to be the recipient of stigma and the subsequent shame that he felt in response to his deafness. As a child in school and into his adult life, he reports on the embarrassment he felt and how this played on the formation of his identity (O'Connell, 2016). Ideally, accounts from the deaf individual themselves holds the promise of truly understanding how a child or adult with HL will experience shame, but interestingly in this study, mothers of children with HL were acutely aware of their child's shame. Participant 8 in particular, illustrates in the above quote that although her child has been afforded the opportunity to hear through cochlear implantation, he is embarrassed as a teenager to have his hearing devices be seen by onlookers that may stare or judge him as being different. Furthermore, participant 10's deaf husband refuses to use sign language in public as it will draw attention to his HL.

For these reasons, and more that will be discussed in the next sub-theme, deaf individuals may gravitate towards one another, finding similarity in their difference. When stigma, labelling and judgement befalls deaf individuals, exclusion from the hearing world may be a social decision in favour of inclusion and acceptance in a Deaf world (McKee, 2006).

4.8.2 THE DEAF WORLD

To understand what is meant by a Deaf world, one must first recognise the cultural differences that exist between those who are deaf and those who are Deaf. As described in chapter 1, deaf refers to the colloquial term for those with HL whereas those who identify with the Deaf community are considered Deaf and their HL is part of their identity rather than a pathology with which they are afflicted.

There are two opposing models of deafness; (1) that it can be seen from the pathological standpoint, i.e. that HL is a medical condition and therefore intervention strategies and remediation are required to correct or compensate for a hearing impairment, and (2) that deafness is a cultural identity, that it cannot be defined as an audiological term but rather as a sociocultural model. The Deaf are a distinct community of individuals shaped by the experience of being deaf (Carey & Palumbos, 2016). Just because someone has a hearing impairment, does not automatically mean that they will form part of a Deaf community (as cited by Munoz-Baell & Ruiz, 2000). The use of assistive devices means that many deaf individuals are able to make use of spoken language and therefore assimilate to the hearing society. Conversely, hearing family members, children and spouses of those who are Deaf may identify with the Deaf community, because having a HL is not the only defining feature of this cultural group (Reagan, Penn & Ogilvy, 2006).

Deaf mentors may play an important role in introducing deaf children and their families to the Deaf world. Hearing parents of deaf children have previously identified the importance of their child's integration and experience of the Deaf world, that personally and culturally identifying with a Deaf community, although it is different from the hearing world that they inhabit, is an important aspect in their child's life. Children who find themselves in a hearing world may inevitably feel different, whereas integration into a Deaf world can be vital for the child's cognition, socialisation and identity (McKee, 2006).

Participant 2 describes in the following quotes, how her child attempts to be part of both worlds, but that she encourages her child to fit into the Deaf world, the world where "he belongs":

"He is a soccer player, he likes to speak all the time. But he have a two two teams. Like he have a deaf team and also at school. But even at location he have another team to soccer ... That is why I say he has lost the normal things now. He know only the deaf. He belong of deaf. Even in the location at the time he plays soccer, I say B don't play the normal person because when you injured you have a problem but when you injured at deaf deaf, they know what is happening ... So go to where you belong, at deafs." (P2)

Other participants viewed it as important that their child finds commonality with other deaf people:

“When she is old, if she can find a place, with other people like her, it will be better for her. She will feel right when she is in the community that is the same to her.” (P4)

“She must have to connect with people who are in her world, of the deaf world” (P5)

Interviewer: *“You said this deaf world, um do you think that she is now almost part of a different community?”*

P5: *“No, no I don’t mean it like different, I mean it like she has to get, there is people who is deaf. She has to get used to the people who is the same like her. That’s what I mean by two worlds, she have our world and she have the deaf world also.”*

As described above, one can see that the participants identified the importance that their child assimilates into the Deaf world. This is echoed by McKee (2006), who went further to say that children who find themselves in a hearing world may inevitably feel different.

When deafness is familial, there is the assumption that integration into the Deaf community is commonplace; that children are introduced immediately into the Deaf world because their parents are already a part of it. This was the case for P6’s teenage daughter, who attends weekly meetings for the Deaf community for women in Cape Town.

When it is introduced at home from an early age, for those with familial HL, there is smoother integration into the Deaf culture. Hearing parents of deaf children may find it more difficult to see their child adopt the ways of another culture, which may include complete rejection of speech in favour of sign language as a first language, and integration into a world where they, as hearing individuals, do not necessarily belong. Some parents believe that connecting these two worlds, so that they may coexist means that there need not be such a distinction between the two ‘worlds’ (McKee, 2006). Historically, social exclusion and oppression of deaf people meant that instead of conforming to the label of disabled, the Deaf separated themselves from the hearing world. This separation of the two worlds is further widened as communication barriers mean that differing social practices are not understood by the other (Munoz-Baell & Ruiz, 2000). Participant 10 provides a solution to this, that by more people learning sign language, the more the two worlds can engage:

“But I really think they must get um the people that can hear involved in this Deaf [world], to understand Deaf people because I think if they understand them then it won’t be like this.” (P10)

In summary, this theme encompasses the internal and external features of stigma, shame and being part of a different 'world'. Parents react differently to the courtesy stigma that they experience in relation to their child's HL with several parents outright rejecting any shame or pity that they receive from external parties, often referring to their child as "normal". This will be further discussed in the next theme, but introduced below by participant 11:

P11: *"The only thing I don't like is when people say shame, she is deaf. I say why shame. I don't like the word shame."*

Interviewer: *"Why don't you like it?"*

P11: *"It's just... because she is not dying, she is just like normal. She can't hear, she can't speak but she does everything else like a normal child and she is not dependent on me, so there is no reason to say shame."*

4.9 THEME 5 - BEING NORMAL: "DEAF PEOPLE ARE PEOPLE"

As introduced in the previous theme, within the medical model of disability, a child with HL is seen as having a deficit. It focuses on the child's non-normativity. What is considered normal is subjective. In this model, a child with HL will be classified as 'different' or 'flawed', which may lead parents of deaf children to feel they need to normalise their child as they respond to the projections outlined in this model of disability (Harvey, 2015).

The term "normal" was mentioned in almost all of the interviews, however used in different contexts. Some of the participants described how they consider their child to be normal, that their HL does not define them, and they shouldn't be labelled as disabled and different. Others were acutely aware that their child is "not normal", in the sense that normal means hearing.

"I didn't look at E and say my child is disabled or she is like something like that. No. I treat her and [her sister] the same." (P5)

This normalisation can be seen as a defence mechanism by parents in this study. By insisting that their deaf child is no different to their other children, or to other hearing children, allows these parents to defend their child against the pathological model of deafness, whereby they would be seen as defective due to their HL. On the other hand, parents may adapt seamlessly to the diagnosis of a deaf child due to a family history of deafness or previous experience with deaf children and adults. In this way, their child is normal, from their frame of reference.

For most of the mothers interviewed (8/11), their child was the first experience that they had with deafness. As previously discussed, the shock and subsequent acceptance and adaptation to the

diagnosis meant that many of the participants have become advocates for deaf ability, as shown in the subsequent quotes. Instead of seeing their child as disabled, they promote the abilities of the child, are proud of what they are able to achieve and encourage awareness regarding their child's ability to overcome their differences. Their child, like any other, has dreams and aspirations that they encourage, and support that their deafness does not define them. This is illustrated in the following quotes:

"... yes you have a deaf child but let me show you that even though you have your deaf child he can still achieve you know he can still achieve what other children can ... And you as a parent must instil that in your child that you can, don't let your deafness hold you back." (P8)

"Deaf people are people, they have their own minds, they can do whatever they want." (P11)

As seen in a previous study with mothers of deaf children in the South African setting, there is a paucity of South African literature that discusses deaf ability. There is limited knowledge in the public regarding the abilities that deaf people may possess which "perpetuates the misconception that the absence of hearing is the absence of mind" (Kara & Harvey, 2016). Participant 5 sums this up as "[She] is deaf yes but she is not stupid".

With further research, awareness and acceptance of the Deaf culture, normalising deafness will lead to a greater acceptance of deaf people and dispelling of misconceptions and stigmatisation.

4.10 CHAPTER SUMMARY

In this chapter, the results of the study were laid out and discussed as they related to the objectives of the study. Five themes were generated from the interview data and they were explored through quotes from the participants and corroborated by existing literature. In the following chapter, conclusions will be drawn and the limitations and strengths of this study reviewed as well as recommendations made for future research in the field of hereditary HL.

CHAPTER 5: CONCLUSIONS, STRENGTHS AND LIMITATIONS OF THIS STUDY AND RECOMMENDATIONS FOR FUTURE RESEARCH

5.1 CHAPTER INTRODUCTION

The final chapter of this study will (1) summarise the pertinent findings of this research, (2) discuss the strengths and limitations of the study and (3) provide suggestions for future research and practical implications of this research.

5.2 CONCLUSIONS

This study aimed to explore the experiences of parents who have a child with hereditary HL in a selected population in Cape Town and their perceived burdens of caring for a child with HL. By investigating these parents' perceptions and understanding regarding the hereditary basis of their child's HL, this research aimed to assess the need and suitability of genetic services for this population. Eleven biological parents of children with HL of a putative nonsyndromic aetiology were interviewed. These interviews were then transcribed and data was analysed using the framework approach so that five themes were developed from the participants' accounts according to the objectives of this study.

The first theme discussed the initial journey following diagnosis of HL. It was found that participants experienced shock, denial and grief in response to the diagnosis of HL in their child. Acceptance and adaptation to the diagnosis was explored as it related to parents' ability to overcome the loss of their child's hearing, ultimately the loss of a "normal" child. Participants described means of support that played a crucial role in accepting their child with HL and adapting to their new experience.

The second theme spoke directly to the hereditary aspects of the participants' child's HL. Several participants experienced misunderstanding regarding the cause of their child's HL. As with previous research, those participants who had previous family history of HL were able to accurately identify the cause of their child's HL as hereditary, whereas most of the participants whose child was the first with HL in the family were unable to identify the genetic aetiology of their child's HL. These misunderstandings meant that several participants experienced guilt and blame in response to the diagnosis of HL in their child. Without knowing the cause of their child's HL, most of the participants in this study had positive attitudes towards genetic testing, in the hope that it would provide them with answers and that the information gained from testing would be of use in the future.

One of the participants in this study had undergone genetic testing and genetic counselling and she was able to correctly identify her child's HL was hereditary but was still unable to explain genetics and how it related to her child's HL and the risk for recurrence. This highlights the need for appropriate genetic counselling and the provision of resources, both pre- and post-testing so that genetic information is well-understood and retained.

It was evident from the research that the practicalities of raising a deaf child were immense. Language and communication barriers as the most apparent, but also financial difficulties and access to schooling and health care. Parents often needed to make sacrifices for their child which included forgoing work so that they could attend multiple clinic visits, as well as the emotional sacrifices and burdens of caring for a child with specialised needs without means of support. Parents in this study found that they had the added worry of needing to provide their deaf child with protection, that their HL made them vulnerable. Overall, the role of the parent, and in this case, the mother was influenced by having a child with HL.

It was found that parents of deaf children experience stigma related to their child's HL. Interestingly, several of the participants strongly rejected any pity that they may have received regarding their child's HL but were also aware that their child may experience shame in response to their felt stigma, that they were aware that their HL made them different. The concept of the "Deaf world" and being part of the Deaf community was mentioned by some of the participants. These parents encouraged their child to find people who are like them, so that they may be accepted and that their deafness need not be a disability but rather form part of their identity.

As a defence mechanism or acceptance of their child, many of the participants used the term "normal" when referring to their child and their abilities. Rather than focusing on the child's disability, these participants advocate that their child's HL does not define them and that they do not need to be held back or be disadvantaged due to their HL.

Although some of these findings fundamentally replicate those of previous studies, such as loss, coping strategies and stigma, the unique context of this study and the immense challenges that these parents face, enhance the implications of these findings.

The findings of this study have provided valuable insight into the perceptions and experiences of parents of children with HL, and in particular, hereditary HL, which has not been explored previously in this setting. This research has several strengths, that are listed below, as well as limitations, but ultimately has the potential to impact the services that are provided to families of children with hereditary HL.

5.3 STENGTHS OF THIS STUDY

- This study is the first undertaken in the South African setting to employ qualitative methodology to examine the parental experience of hereditary HL in the child.
- Several of the participants felt the interview process to be therapeutic, and that discussing their child and the impact that their HL has had on their lives was both helpful and healing.
- This small convenience sample illuminated novel issues related to the context of the selected setting in Cape Town and is potentially relevant to other developing countries.
- The interview guide was designed to include open-ended questions which encouraged participants to answer freely and openly. There were no predetermined themes. However, as data analysis occurred in parallel to data collection, the ideas and themes emerging from the initial interviews were explored in subsequent interviews.
- As there are only a handful of genetic counselling students undertaking qualitative research regarding genetic conditions, this research is valuable for the growing field of genetic counselling in SA.

5.4 LIMITATIONS OF THIS STUDY

- This was the researcher's first introduction to qualitative research and interviewing. Although the purpose of undertaking a new methodology for research was for practical growth, the consequence was that the first few interviews were not as well conducted as the later ones. Ideally an experienced qualitative interviewer would have used, but would mean that the researcher would not have been able to engage with the participants.
- Most of the interviews were conducted in English. Although not all of the participants' first language, the offer was made to include an interpreter, but only two of the mothers agreed to this. As the interpreters were lay persons working at the school, the quality of their interpretations may have been affected.
- Purposive sampling does lend itself to ascertainment bias. The views of those unwilling to participate, unable to attend the interview or less communicative are not included.
- Only a small proportion of those contacted for involvement made themselves available for an interview. This lack of engagement by many of the parents may be explained by a number of factors. Firstly, many of the children who attend the DSDC are there as boarders – either weekly or for the term – and their homes far from the research site, making it difficult for parents to travel to the school for the interviews. Although an offer was made by the researcher to travel to the parents for the interview, none of the potential participants took

up this offer. Secondly, the nature of this research may cause parents to not be interested in being interviewed, as their experiences with their child's HL may be unfavourable and they may not wish to discuss these matters. Furthermore, as previously reported, many South Africans are faced with burdens (van der Spuy & Pottas, 2008) and therefore cultural, economic and linguistic barriers prevail between researchers and their proposed subjects (Benatar, 2002).

- Some of the participants brought children and family members to the interviews which may have influenced their candour.
- Similarly, the interviews that were conducted at the school may have had the same effect as parents may have felt that the researcher had an association with the school and that perhaps their answers would influence the care of their child.

5.5 PRACTICAL IMPLICATIONS OF THIS RESEARCH

Aspects of this research have practical applications and have been recommended below:

- Better communication and referrals from audiology to genetics services at RCWMCH and from other clinics and hospitals.
- Information sheets and supportive services offered to parents of newly diagnosed children with HL and translated into several languages, using appropriate terminology.
- The formation of a support group, ideally through the school, so that parents may openly discuss issues that they are having with parents who are in a similar situation.
- It is important, for both audiology services and genetics health care professionals, to understand the experiences and perspectives of parents of children who have hereditary HL, with the aim of better service delivery and meeting the specialised needs of these families.

5.6 RECOMMENDATIONS FOR FUTURE RESEARCH

The following recommendations have been made based on this research, to guide future research on the topic:

- Examining the child and adolescent's view of what has caused their HL and what they have experienced in relation to their deafness.
- Exploring the experiences of hearing children of D/deaf adults. As they live between two worlds, their insight into the lives of their family and the experiences that they have with regards to being hearing in the Deaf world.

- Research into the father's role as a parent of a deaf child will be valuable. Although not a prerequisite for this study, all of the participants were mothers. Fathers may present new insights into parenting a deaf child, and may have differing views on the concepts of hereditary and the emotions they assign to it.
- As research is ongoing to determine the molecular basis of HL in black South African populations, testing may become available and guide the clinical diagnosis of many children whose families do not have a definitive cause for the child's HL. Results will need to be fed-back to the families and may present interesting findings with regards to their understanding of the cause of their child's HL. A qualitative approach would be most suited to this.

6.7 CHAPTER SUMMARY

In the final chapter of this study, the conclusion drawn from the research are presented, together with the strengths and limitations of the research. Both practical implications of the research as well as recommendations for future research are discussed.

REFERENCES

- Adedokun, B. O., Yusuf, B. O., Lasisi, J. T., Jinadu, A. A., Sunmonu, M. T., Ashanke, A. F. & Lasisi, A. O. 2015. Perception of Genetic Testing for Deafness and Factors Associated with Interest in Genetic Testing Among Deaf People in a Selected Population in Sub-Saharan Africa. *Journal of Genetic Counseling*, 24(6):1037-1043.
- Ahlert, I. A. & Greeff, A. P. 2012. Resilience Factors Associated With Adaptation in Families With Deaf and Hard of Hearing Children. *American Annals of the Deaf*, 157(4):391-404.
- Albertyn, C. 2006. *Informal settlements: A perpetual challenge?*, Cape Town, University of Cape Town Press.
- Alford, R. L., Arnos, K. S., Fox, M., Lin, J. W., Palmer, C. G., Pandya, A., Rehm, H. L., Robin, N. H., *et al.* 2014. American college of medical genetics and Genomics guideline for the clinical evaluation and etiologic diagnosis of hearing loss. *Genetics in Medicine*, 16(4):347-355.
- Anderegg, M. L., Vergason, G. A. & Smith, M. C. 1992. A visual representation of the grief cycle for use by teachers with families of children with disabilities. *Remedial and Special Education*, 13(2):17-23.
- Association, W. M. 2008. WMA declaration of Helsinki - ethical principles for medical research involving human subjects.
- Barrett, K. E., Barman, S. M., Boitano, S. & Brooks, H. L. 2010. Hearing & Equilibrium. *Ganong's Review of Medical Physiology*. 23 ed. New York: McGraw-Hill Companies.
- Beighton, P., Fieggen, K., Wonkam, A., Ramesar, R. & Greenberg, J. 2012. The University of Cape Town's contribution to medical genetics in Africa - from the past into the future. *SAMJ: South African Medical Journal*, 102(6):446-448.
- Beighton, P., Fieggen, K., Wonkam, A., Ramesar, R. & Greenberg, J. 2012. The University of Cape Town's contribution to medical genetics in Africa: from the past into the future. *SAMJ: South African Medical Journal*, 102(6):446-448.
- Benatar, S. R. 2002. Reflections and recommendations on research ethics in developing countries. *Social Science & Medicine*, 54(7):1131-1141.
- Benetti-McQuoid, J. & Bursik, K. 2005. Individual differences in experiences of and responses to guilt and shame: Examining the lenses of gender and gender role. *Sex Roles*, 53(1-2):133-142.
- Bennett, T., Deluca, D. A. & Allen, R. W. 1995. Religion and children with disabilities. *Journal of Religion and Health*, 34(4):301-312.

- Blacher, J. 1984. Sequential stages of parental adjustment to the birth of a child with handicaps: fact or artifact? *Mental Retardation*, 22(2):55-68.
- Bosch, J., Lebeko, K., Nziale, J. J. N., Dandara, C., Makubalo, N. & Wonkam, A. 2014a. In search of genetic markers for nonsyndromic deafness in Africa: A study in Cameroonians and black South Africans with the GJB6 and GJA1 candidate genes. *OMICS: A Journal of Integrative Biology*, 18(7):481-485.
- Bosch, J., Noubiap, J. J. N., Dandara, C., Makubalo, N., Wright, G., Entfellner, J.-B. D., Tiffin, N. & Wonkam, A. 2014b. Sequencing of GJB2 in Cameroonians and black south Africans and comparison to 1000 Genomes project data support need to revise strategy for discovery of Nonsyndromic deafness genes in Africans. *OMICS: A Journal of Integrative Biology*, 18(11):705-710.
- Bosteels, S., Van Hove, G. & Vandenbroeck, M. 2012. The roller-coaster of experiences: becoming the parent of a deaf child. *Disability & Society*, 27(7):983-996.
- Brunger, J., Murray, G., M., O. R., Matthews, A. L., Smith, R. J. H. & Robin, N. H. 2000. Parental attitudes toward genetic testing for pediatric deafness. *American Journal of Medical Genetics*, 671621-1625.
- Carey, J. C. & Palumbos, J. C. 2016. Advances in the understanding of the genetic causes of hearing loss in children inform a rational approach to evaluation. *The Indian Journal of Pediatrics*, 83(1):1-7.
- Carpenter, B. 2007. The impetus for family-centred early childhood intervention. *Child: Care, Health and Development*, 33(6):664-669.
- Chan, D. K. & Chang, K. W. 2014. GJB2-associated hearing loss: Systematic review of worldwide prevalence, genotype, and auditory phenotype. *The Laryngoscope*, 124(2):E34-E53.
- Chang, K. W. 2015. Genetics of hearing loss—Nonsyndromic. *Otolaryngologic Clinics of North America*, 48(6):1063-1072.
- Clark, J. G. 1981. Uses and abuses of hearing loss classification. *Asha*, 23(7):493-500.
- Copley, G. J. & Friderichs, N. B. 2010. An approach to hearing loss in children. *South African Family Practice*, 52(1):34-39.
- Davis, L. J. 2006. Constructing normalcy. *The disability studies reader*. 2 ed. New York, NY: Routledge.
- de Andrade, V. & Ross, E. 2005. Beliefs and practices of black south African traditional healers regarding hearing impairment. *International Journal of Audiology*, 44(9):489-499.
- de Andrade, V. M. 2015. "We Do Not Understand Each Other:" *The Experience of Caregivers of Deaf Children in a Rural South African Setting*. PhD, Univeristy of Witswatersrand.

- Denoyelle, F., Weil, D., Maw, M. A., Wilcox, S. A., Lench, N. J., Allen-Powell, D. R., Osborn, A. H., Dahl, H. H. M., *et al.* 1997. Prelingual deafness: High prevalence of a 30delG mutation in the Connexin 26 gene. *Human Molecular Genetics*, 6(12):2173-2177.
- Denscombe, M. 2010. *The Good Research Guide: For Small-Scale Social Research Projects*, Maidenhead, England, McGraw-Hill Professional Publishing.
- Dickson-Swift, V., James, E. L., Kippen, S. & Liamputtong, P. 2007. Doing sensitive research: What challenges do qualitative researchers face? *Qualitative Research*, 7(3):327-353.
- Education, S. A. D. o. B. 2011. *National Curriculum Statement: Life Sciences*, Pretoria, Department of Basic Education.
- Egilmez, O. K. & Kalcioğlu, M. T. 2016. Genetics of nonsyndromic congenital hearing loss. *Scientifica*, 20161-9.
- Emery, S. D., Middleton, A. & Turner, G. H. 2010. Whose deaf genes are they anyway? The deaf community's challenge to legislation on embryo selection. *Sign Language Studies*, 10(2):155-169.
- Fox, S. I. 2008. Sensory Physiology. *Human Physiology*. 10 ed. New York, NY: McGraw Hill Higher Education.
- Gasmelseed, N. M., Schmidt, M., Magzoub, M. M., Macharia, M., Elmustafa, O. M., Ototo, B., Winkler, E., Ruge, G., *et al.* 2004. Low frequency of deafness-associated GJB2 variants in Kenya and Sudan and novel GJB2 variants. *Human Mutation*, 23(2):206-207.
- Glaser, B. G. & Strauss, A. L. 2009. *The discovery of grounded theory: Strategies for qualitative research*, Transaction publishers.
- Goffman, E. 1963. *Stigma notes on the management of spoiled identity*, Englewood Cliffs, NJ, Prentice Hall.
- Green, S. E. 2003. "What do you mean 'what's wrong with her?'": Stigma and the lives of families of children with disabilities. *Social Science & Medicine*, 57(8):1361-1374.
- Greenberg, J., Kromberg, J., Loggenberg, K. & Wessels, T.-M. 2012. Genetic counseling in South Africa. In: KUMAR, D. (ed.) *Genomics and health in the developing world*. Oxford: Oxford University Press.
- Hagedorn, E. 2015. *Caregivers' experiences of the diagnosis of hearing loss*. MSc Audiology, University of Cape Town.
- Hammersley, M. 1992. Some reflections on ethnography and validity 1. *International Journal of Qualitative Studies in Education*, 5(3):195-203.
- Harvey, C. 2015. Maternal subjectivity in mothering a child with a disability: A psychoanalytical perspective. *Agenda: Empowering women for gender equity*, 29(2):89-100.

- Hearing, J. C. o. I. 2007. Year 2007 position statement: Principles and guidelines for early hearing detection and intervention programs. *Pediatrics*, 120(4):898-921.
- Henderson, D. & Hendershott, A. 1991. ASL and the Family System. *American Annals of the Deaf*, 136(4):325-329.
- Hennink, M., Hutter, I. & Bailey, A. 2010. The nature of qualitative research. *Qualitative research methods*. Los Angeles, CA: SAGE Publications.
- Hertzano, R. & Elkon, R. 2012. High throughput gene expression analysis of the inner ear. *Hearing Research*, 288(1-2):77-88.
- Hesse-Biber, S. N. & Leavy, P. L. 2010. Designing qualitative approaches to research. *The practice of qualitative research*. 2 ed. Los Angeles: SAGE Publications.
- Hewson, D. 1997. Coping with loss of ability: "Good grief" or episodic stress responses? *Social Science & Medicine*, 44(8):1129-1139.
- Hintermair, M. 2006. Parental Resources, Parental Stress, and Socioemotional Development of Deaf and Hard of Hearing Children. *Journal of Deaf Studies and Deaf Education*, 11(4):493-513.
- Holloway, I. 2005. *Qualitative Research in Health Care*, Maidenhead, Open University Press.
- Jackson, C. W., Traub, R. J. & Turnbull, A. P. 2008. Parents' Experiences With Childhood Deafness: Implications for Family-Centered Services. *Communication Disorders Quarterly*, 29(2):82-98.
- Jackson, C. W. & Turnbull, A. 2004. Impact of Deafness on Family Life: A Review of the Literature. *Topics in Early Childhood Special Education*, 24(1):15-29.
- James, C. A., Hadley, D. W., Holtzman, N. A. & Winkelstein, J. A. 2006. How does the mode of inheritance of a genetic condition influence families? A study of guilt, blame, stigma, and understanding of inheritance and reproductive risks in families with X-linked and autosomal recessive diseases. *Genetics in Medicine*, 8(4):234.
- Javidnia, H., Carson, M., Awubwa, R., Byaruchanga, R., Mack, D. & Vaccani, J. P. 2014. Connexin gene mutations among Ugandan patients with nonsyndromic sensorineural hearing loss. *Laryngoscope*, 124(9):E373-376.
- Jones, L., Bellis, M. A., Wood, S., Hughes, K., McCoy, E., Eckley, L., Bates, G., Mikton, C., *et al.* 2012. Prevalence and risk of violence against children with disabilities: A systematic review and meta-analysis of observational studies. *The Lancet*, 380(9845):899-907.
- Kabahuma, R. I., Ouyang, X., Du, L. L., Yan, D., Hutchin, T., Ramsay, M., Penn, C. & Liu, X.-Z. 2011. Absence of GJB2 gene mutations, the GJB6 deletion (GJB6-D13S1830) and four common mitochondrial mutations

- in nonsyndromic genetic hearing loss in a south African population. *International Journal of Pediatric Otorhinolaryngology*, 75(5):611-617.
- Kaimal, G., Steinberg, A. G., Ennis, S., Harasink, S. M., Ewing, R. & Li, Y. 2007. Parental Narratives About Genetic Testing for Hearing Loss: A One Year Follow Up Study. *Journal of Genetic Counseling*, 16(6):775-787.
- Kampfe, C. M. 1989. Parental reaction to a child's hearing impairment. *American Annals of the Deaf*, 134(4):255-259.
- Kara, N. & Harvey, C. 2016. The social construction of 'deafness': explored through the experiences of Black South African mothers raising a deaf child. *South African Journal of Psychology*, 1-12.
- Kelsell, D. P., Dunlop, J., Stevens, H. P., Lench, N. J., Liang, J. N., Parry, G., Mueller, R. F. & Leigh, I. M. 1997. Connexin 26 mutations in hereditary non-syndromic sensorineural deafness. *Nature*, 387(6628):80-83.
- Kerr, G., Tuomi, S. & Müller, A. 2012. Costs involved in using a cochlear implant in South Africa. *South African Journal of Communication Disorders*, 59(1):16-26.
- Kessler, S., Kessler, H. & Ward, P. 1984. Psychological aspects of genetic counseling. III. Management of guilt and shame. *American Journal of Medical Genetics*, 17(3):673-697.
- Kilian, S., Swartz, L. & Joska, J. 2010. Competence of interpreters in a South African psychiatric hospital in translating key psychiatric terms. *Psychiatric Services*, 61(3):183-200.
- Kiyaga, N. B. & Moores, D. F. 2003. Project MUSE - deafness in Sub-Saharan Africa. *American Annals of the Deaf*, 148(1):18-24.
- Kotby, M. N., Tawfik, S., Aziz, A. & Taha, H. 2008. Public Health Impact of Hearing Impairment and Disability. *Folia Phoniatrica et Logopaedica*, 60(2):58-63.
- Kübler-Ross, E. 1969. *On death and dying: What the dying have to teach doctors, nurses, clergy, and their own family*, New York, Macmillan Publishing Company.
- Kübler-Ross, E. & Kessler, D. 2014. *On grief and grieving: Finding the meaning of grief through the five stages of loss*, United States of America, Simon & Schuster.
- Kurtzer-White, E. & Luterman, D. 2003. Families and children with hearing loss: Grief and coping. *Mental Retardation and Developmental Disabilities Research Reviews*, 9(4):232-235.
- Kvale, S. 2007. *Doing Interviews*, London, Sage Publications Ltd.
- Kvam, M. H. 2004. Sexual abuse of deaf children. A retrospective analysis of the prevalence and characteristics of childhood sexual abuse among deaf adults in Norway. *Child Abuse & Neglect*, 28(3):241-251.

- Lasisi, A. O., Bademci, G., Foster, J., Blanton, S. & Tekin, M. 2014. Common genes for non-syndromic deafness are uncommon in sub-Saharan Africa: a report from Nigeria. *International Journal of Pediatric Otorhinolaryngology*, 78(11):1870-1873.
- Lazarus, R. S. & Folkman, S. 1984. *Stress, appraisal, and coping*, New York, Springer Publishing Company.
- Lebeko, K., Bosch, J., Nzeale, J. J., Dandara, C. & Wonkam, A. 2015. Genetics of hearing loss in Africans: Use of next generation sequencing is the best way forward. *Pan African Medical Journal*, 20.
- Lebeko, K., Sloan-Heggen, C. M., Noubiap, J. J., Dandara, C., Kolbe, D. L., Ephraim, S. S., Booth, K. T., Azaiez, H., *et al.* 2016. Targeted genomic enrichment and massively parallel sequencing identifies novel nonsyndromic hearing impairment pathogenic variants in Cameroonian families. *Clinical Genetics*.
- Lederberg, A. R. & Golbach, T. 2002. Parenting stress and social support in hearing mothers of deaf and hearing children: A longitudinal study. *Journal of Deaf Studies and Deaf Education*, 7(4):330-345.
- Lupton, D. 1998. *The emotional self: A sociocultural exploration*, London, SAGE Pub.
- MacFarlane, I., Veach, P. M. & LeRoy, B. S. 2014. Conducting Qualitative Genetic Counselling Research. *Genetic Counselling Research: A Practical Guide*. 1 ed. United States of America: Oxford University Press.
- Mack, N., Woodsong, C., MacQueen, K. M., Guest, G. & Namey, E. 2005. *Qualitative Research Methods: A Data Collector's Field Guide*, USA, Family Health International.
- Mall, S. 2012. *HIV prevention issues for Deaf and hard of hearing adolescents: Views of parents, teachers, adolescents and organizations serving the Deaf community*. PhD, Univeristy of Stellenbosch.
- Mall, S. & Swartz, L. 2011. Parents' anxieties about the risk of HIV/AIDS for their deaf and hard of hearing adolescents in South Africa: A qualitative study. *Journal of Health Psychology*, 17(5):764-773.
- Marshall, M. 1996. Sampling for qualitative research. *Family Practice*, 13(6):522-525.
- Maslow, A. H. 1970. *Motivation and personality*, New York, Addison-Wesley Educational Publishers.
- Mason, A. & Mason, M. 2007. Psychologic impact of deafness on the child and adolescent. *Primary Care: Clinics in Office Practice*, 34(2):407-426.
- McKee, M., Schlehofer, D. & Thew, D. 2013. Ethical issues in conducting research with deaf populations. *American Journal of Public Health*, 103(12):2174-2178.
- McKee, R. 2006. Connecting hearing parents with the Deaf world. *Sites: a journal of social anthropology and cultural studies*, 3(1):143-167.

- Meinzen-Derr, J., Lim, L. H. Y., Choo, D. I., Buyniski, S. & Wiley, S. 2008. Pediatric hearing impairment caregiver experience: Impact of duration of hearing loss on parental stress. *International Journal of Pediatric Otorhinolaryngology*, 72(11):1693-1703.
- Merriam, S. B. 2014. *Qualitative Research A Guide to Design and Implementation*, United States, Jossey-Bass.
- Meyer, M. E., Swanepoel, D. W., le Roux, T. & van der Linde, M. 2012. Early detection of infant hearing loss in the private health care sector of South Africa. *International Journal of Pediatric Otorhinolaryngology*, 76(5):698-703.
- Middleton, A., Emery, S. D. & Turner, G. H. 2010. Views, knowledge, and beliefs about genetics and genetic counseling among Deaf people. *Sign Language Studies*, 10(2):170-196.
- Middleton, A., Hewison, J. & Mueller, R. 2001. Prenatal Diagnosis for Inherited Deafness—What is the Potential Demand? *Journal of Genetic Counseling*, 10(2):121-131.
- Miller, P. S. & Levine, R. L. 2013. Avoiding genetic genocide: understanding good intentions and eugenics in the complex dialogue between the medical and disability communities. *Genetics in Medicine*, 15(2):95-102.
- Morse, J. M. 2012. *Qualitative health research: Creating a new discipline*, United States, Left Coast Press.
- Moses, T. 2010. Exploring parents' self-blame in relation to adolescents' mental disorders. *Family Relations*, 59(2):103-120.
- Munoz-Baell, I. M. & Ruiz, M. T. 2000. Empowering the deaf. Let the deaf be deaf. *Journal of Epidemiology & Community Health*, 54(1):40-44.
- Murray, C. J. L., Barber, R. M., Foreman, K. J., Ozgoren, A. A., Abd-Allah, F., Abera, S. F., Aboyans, V., Abraham, J. P., *et al.* 2015. Global, regional, and national disability-adjusted life years (DALYs) for 306 diseases and injuries and healthy life expectancy (HALE) for 188 countries, 1990–2013: Quantifying the epidemiological transition. *The Lancet*, 386(10009):2145-2191.
- Nagakura, H., Schneider, G., Morris, J., Lafferty, K. A. & Palmer, C. G. S. 2015. Assessing Deaf Awareness Training: Knowledge and Attitudes of Recent Genetic Counseling Graduates. *Journal of Genetic Counseling*, 24104-116.
- O'Connell, N. P. 2016. "Passing as normal": Living and coping with the stigma of deafness. *Qualitative Inquiry*, 1-11.
- Olusanya, B. O., Neumann, K. J. & Saunders, J. E. 2014. The global burden of disabling hearing impairment: a call to action. *Bulletin of the World Health Organization*, 92(5):367-373.
- OMIM. 2016. *Online Mendelian Inheritance in Man* [Online]. [Accessed 2016].

- Omondi, D., Ogol, C., Otieno, S. & Macharia, I. 2007. Parental awareness of hearing impairment in their school-going children and healthcare seeking behaviour in Kisumu district, Kenya. *International Journal of Pediatric Otorhinolaryngology*, 71(3):415-423.
- Parker, M. & Bitner-Glindzicz, M. 2014. Genetic investigations in childhood deafness. *Archives of Disease in Childhood*, 100(3):271-278.
- Parker, M. J., Fortnum, H. M., Young, I. D. & Davis, A. C. 2000. Genetics and deafness: what do families want? *Journal of Medical Genetics*, 37(10):e26-e26.
- Peräkylä, A. 2011. Validity in research on naturally occurring social interaction. *Qualitative Research*, 365-382.
- Poon, B. T. & Zaidman-Zait, A. 2013. Social Support for Parents of Deaf Children: Moving Toward Contextualized Understanding. *Journal of Deaf Studies and Deaf Education*, 19(2):176-188.
- Pope, C., Ziebland, S. & Mays, N. 2000. Qualitative research in health care: Analysing qualitative data. *BMJ*, 320(7227):114-116.
- Quittner, A. L., Barker, D. H., Cruz, I., Snell, C., Grimley, M. E., Botteri, M. & Team, C. I. 2010. Parenting stress among parents of deaf and hearing children: Associations with language delays and behavior problems. *Parenting*, 10(2):136-155.
- Reagan, T., Penn, C. & Ogilvy, D. 2006. From policy to practice: Sign language developments in post-apartheid South Africa. *Language Policy*, 5(2):187-208.
- Rodrigo, M. J., Martín, J. C., Máiquez, M. L. & Rodríguez, G. 2007. Informal and formal supports and maternal child-rearing practices in at-risk and non at-risk psychosocial contexts. *Children and Youth Services Review*, 29(3):329-347.
- Rodrigues, F., Paneque, M., Reis, C., Venâncio, M., Sequeiros, J. & Saraiva, J. 2013. Non-syndromic Sensorineural Prelingual Deafness: The Importance of Genetic Counseling in Demystifying Parents' Beliefs About the Cause of Their Children's Deafness. *Journal of Genetic Counseling*, 22(4):448-454.
- Rubin, H. J. & Rubin, I. S. 2011. *Qualitative interviewing: The art of hearing data*, Thousand Oaks, CA, SAGE Publications.
- Sankar, P., Cho, M. K., Wolpe, P. R. & Schairer, C. 2006. What is in a cause? Exploring the relationship between genetic cause and felt stigma. *Genetics in Medicine*, 8(1):33-42.
- Scambler, G. 1998. Stigma and disease: Changing paradigms. *The Lancet*, 352(9133):1054-1055.
- Sellars, S. & Beighton, P. 1983. Childhood deafness in Southern Africa: An aetiological survey of 3,064 deaf children. *The Journal of Laryngology & Otology*, 97(10):885-889.

- Silverman, D. 2011. *Interpreting qualitative data: A guide to the principles of qualitative research*, Los Angeles, SAGE Publications.
- Silverman, D. 2015. *Interpreting qualitative data*, United Kingdom, Sage Publications.
- Singer, G. H. S. & Irvin, L. K. 1989. Family care- giving, stress, and support. *Support for caregiving families: Enabling positive adaptations to disability* Baltimore: Paul H. Brookes Publishing.
- Smith, A. W. 2001. WHO activities for prevention of deafness and hearing impairment in children. *Scandinavian Audiology*, 30(2):93-100.
- Smith, A. W. 2003. Preventing deafness—an achievable challenge. The WHO perspective. *International Congress Series*, 1240183-191.
- Smith, J. & Firth, J. 2011. Qualitative data analysis: The framework approach. *Nurse Researcher*, 18(2):52-62.
- Smith, R. J. H., Shearer, E. A., Hildebrand, M. S. & Van Camp, G. 2014. *Deafness and Hereditary Hearing Loss Overview*, University of Washington, Seattle.
- Spencer, L., Richie, J. & O'Connor, W. 2003. Analysis: practice, principles and processes. In: RITCHIE, J. & LEWIS, J. (eds.) *Qualitative research practice: A guide for social science students and researchers*; Ed. By Jane Ritchie. London: Sage Publications.
- Spriggs, M. 2002. Lesbian couple create a child who is deaf like them. *Journal of Medical Ethics*, 28(5):283-283.
- Steinberg, A., Kaimal, G., Ewing, R., Soslow, L. P., Lewis, K. M., Krantz, I. & Li, Y. 2007. Parental Narratives of Genetic Testing for Hearing Loss: Audiologic Implications for Clinical Work With Children and Families. *American Journal of Audiology*, 16(1).
- Störbeck, C. 2012. Childhood Hearing Loss in the Developing World. *International Journal of Child Health and Nutrition*, 1(1):59-65.
- Sullivan, P. M., Vernon, M. & Scanlan, J. M. 1987. Sexual abuse of deaf youth. *American Annals of the Deaf*, 132(4):256-262.
- Swanepoel, D., Störbeck, C. & Friedland, P. 2009. Early hearing detection and intervention in South Africa. *International Journal of Pediatric Otorhinolaryngology*, 73(6):783-786.
- Theunissen, M. & Swanepoel, D. 2008. Early hearing detection and intervention services in the public health sector in South Africa. *International Journal of Audiology*, 47(sup1):S23-S29.
- Turner, D. W. 2010. Qualitative Interview Design: A Practical Guide for Novice Investigators. *The Qualitative Report*, 15(3):754-760.

- van der Spuy, T. & Pottas, L. 2008. Infant hearing loss in South Africa: Age of intervention and parental needs for support. *International Journal of Audiology*, 47(s1):30-35.
- Walsh, F. 2002. A family resilience framework: Innovative practice applications. *Family Relations*, 51(2):130-137.
- Weil, J. 2000. *Psychosocial genetic counseling*, New York, Oxford University Press, USA.
- Whitehead, D. 2012. Common qualitative methods. *Nursing and midwifery research: Methods and appraisal for evidence-based practice*. 4 ed. Australia: Elsevier Australia.
- WHO. 2015. *Deafness and hearing loss* [Online]. World Health Organization. Available: <http://www.who.int/mediacentre/factsheets/fs300/en/> [Accessed 10 May 2016].
- Wonkam, A. 2015. Letter to the editor regarding "GJB2, GJB6 or GJA1 genes should not be investigated in routine in non syndromic deafness in people of sub-Saharan African descent". *International Journal of Pediatric Otorhinolaryngology*, 79(4):632-633.
- Wonkam, A., Bosch, J., Noubiap, J. J., Lebeko, K., Makubalo, N. & Dandara, C. 2015. No evidence for clinical utility in investigating the connexin genes GJB2, GJB6 and GJA1 in non-syndromic hearing loss in black Africans. *South African Medical Journal*, 105(1):23-26.
- Wonkam, A., Noubiap, J. J., Djomou, F., Fieggen, K., Njock, R. & Toure, G. B. 2013. Aetiology of childhood hearing loss in Cameroon (sub-Saharan Africa). *European Journal of Medical Genetics*, 56(1):20-25.
- Zaidman-Zait, A., Most, T., Tarrasch, R., Haddad-eid, E. & Brand, D. 2016. The impact of childhood hearing loss on the family: mothers' and fathers' stress and coping resources. *Journal of Deaf Studies and Deaf Education*, 21(1):23-33.

APPENDIX A: INFORMATION SHEET FOR PARTICIPANT RECRUITMENT

Dear Parent,

My name is Sinead Amber Ross and I am a student at the University of Cape Town, doing a Master's degree in Genetic Counselling. I am doing a small research project and would like to invite you to participate in my study.

The study aims to examine the perspectives, experiences and understanding of parents, whose children are deaf/hard of hearing due to a genetic cause, something that may run in the family or happen for the first time in your child.

This study has been initiated through the Division of Human Genetics at the University of Cape Town and is for a minor dissertation for the completion of a Master's degree in Genetic Counselling.

You have been invited for involvement in this research because:

- With your previous permission, a member of our clinical team has seen your child at their school during the Genetic Outreach Service, and confirmed that your child's hearing loss is possibly due to a genetic cause.
- OR you have two or more children that are deaf.
- OR you and your partner are in some way related.

Should you wish to participate, I would like to talk to you about your experiences of having a child who is deaf and explore your understanding of what caused your child's deafness. The discussion will take about an hour, at your child's school in a private location, where refreshments and R50 for transport will be provided. The interview will be audio recorded and any identifying information, such as your name, will be kept confidential and only be known by the researcher. Some parts of the audio-recorded interview may be used in reporting of this study.

Your participation will be entirely voluntary and you can withdraw from the study at any point, with no consequences for either you or your child. This being a research study, there will be no medical benefits to you or your child. Some of the questions that will be asked may be of a sensitive manner. If you need further assistance in this regard, referral to appropriate health care professionals will be arranged.

The Human Research Ethics Committee at the Faculty of Health Sciences, University of Cape Town, has approved this study. If you have any questions about your rights as a participant, please contact Prof Marc Blockman, Chair of the Human Research Ethics Committee on 021 406 6496.

Should you have any questions about the research project, please contact me at rssin001@myuct.ac.za or by phone at 021 406 6304 or the project supervisor Prof Ambroise Wonkam at ambroise.wonkam@uct.ac.za or by phone at 021 406 6307.

Should you wish to be involved in the study, please read the Consent Form attached.

APPENDIX B: PARTICIPANT CONSENT FORM

Study Title: Parents' perspectives and experiences of having a child with hereditary hearing loss.

1. I have been invited to participate in the above research project because I have a child who is deaf/hard of hearing.
2. I understand that both the questionnaire and the interview will be handled confidentially. The data will be used for this research project as well as publications/dissemination of information, but I, as a participant, will not be identified.
3. I understand that some of the questions may cause an emotional reaction, but the risk of harm is small and should I require further counselling, the researcher will make arrangements.
4. I understand that the interview will be audiotape-recorded. These recordings will only be available to the researcher, supervisors and examiners and will be destroyed after the research is completed. My information will not be identifiable in any dissemination of the study.
5. I understand that I am free to decide not to participate or to withdraw at any time without consequences.
6. I have been offered R50 for transport costs when attending the interview.
7. I understand that there will be no medical benefits to me or my child from this study.
8. I understand that the registered Human Research Ethics Committee at the Faculty of Health Sciences, University of Cape Town, has approved this study. I have been given contact details should I need to contact the committee about my treatment as a research participant.
9. _____ has explained the information of this study to me and my questions have been answered satisfactorily.

I hereby declare that I have voluntarily agreed to participate in the above research study and that I agree to have my interview audiotape-recorded.

Signed at _____ on _____ 2015

(Participant's signature)

(Witness's signature)

APPENDIX C: QUESTIONNAIRE

Please answer the following questions before the interview. Tick the correct circle or write your answer on the line.

1. Are you male or female?
 - Male
 - Female

2. How old are you?

3. What is your preferred home language?
 - English
 - Afrikaans
 - isiXhosa
 - isiZulu
 - Sign language
 - Other (please specify)

4. Is your child male or female?
 - Male
 - Female

5. How old is your child?

6. What is your marital status?
 - Single
 - Married
 - Informal relationship
 - Divorced
 - Widowed

7. What is the highest level of education you have completed?
 - Grade 7
 - Matric
 - University
 - Post-graduate studies
 - Other (please specify)

8. Do you live in a rural or urban area?
 - Rural (village, informal settlement, small town)
 - Urban (city)

9. How much money do all members of your household earn monthly?
 - R0 – R3000
 - R3000 – R7000
 - R7000 – R10 000
 - R10 000 – R25 000
 - R25 000 – R50 000
 - More than R50 000

10. How many children do you have?

11. How many of your children are deaf/hard of hearing?

12. Do you have other family members who are deaf/hard of hearing?
 - No
 - Yes

If yes, how many?

APPENDIX D: INTERVIEW GUIDE

- Perhaps we can start with you telling me a bit about your family?
 - o Probes:
 - Children
 - Partner
 - Living situation
 - Employment
- Can we talk about your child with hearing loss?
- How has your child's hearing loss impacted your life?
 - o Probes:
 - Home
 - Other children
 - Partner
 - Work
 - Relationships – with friends and within the community
- Could you describe your experience when you found out that he/she was deaf?
 - o Prompts:
 - What support did you receive at this time?
 - What did you need most? E.g. information, counselling
- What have been some of the challenges/significant moments managing your child's deafness?
- How do you think your child might be judged differently in society/the community because of their deafness?
- What is your understanding of what has caused your child's deafness?
 - o If "I don't know" prompt:
 - Environmental, something in the family, sickness as a baby
- Do you know what genetics means/is?
 - o Explain, genetics is...
- Do you think this may be the cause of your child's deafness?
 - o If yes: how does that make you feel?
 - o If no: How do you think parents of a child who is deaf due to genetic cause may feel differently?
- Would you be interested in testing your child to determine if their hearing loss is genetic?
 - o How do you think it would help you?
 - o Why/why not?

- Are you worried about your child?
 - o Why/why not?
- What are your main concerns for your child for the future?
 - o Prompts:
 - Finding a partner
 - Career
 - When they have children
- What do you think the chance may be, when they have children, that their child will be deaf?



Audrey.wyngaard@westerncape.gov.za

tel: +27 021 467 9272

Fax: 0865902282

Private Bag x9114, Cape Town, 8000

wced.wcape.gov.za

REFERENCE: 20151029-4627

ENQUIRIES: Dr A T Wyngaard

Miss Sinead Ross
223B High Level Road
Sea Point
8005

Dear Miss Sinead Ross

RESEARCH PROPOSAL: PARENTS' PERSPECTIVES AND EXPERIENCES OF HAVING A CHILD WITH HEREDITARY HEARING LOSS

Your application to conduct the above-mentioned research in schools in the Western Cape has been approved subject to the following conditions:

1. Principals, educators and learners are under no obligation to assist you in your investigation.
2. Principals, educators, learners and schools should not be identifiable in any way from the results of the investigation.
3. You make all the arrangements concerning your investigation.
4. Educators' programmes are not to be interrupted.
5. The Study is to be conducted from **18 January 2016 till 30 May 2016**
6. No research can be conducted during the fourth term as schools are preparing and finalizing syllabi for examinations (October to December).
7. Should you wish to extend the period of your survey, please contact Dr A.T Wyngaard at the contact numbers above quoting the reference number?
8. A photocopy of this letter is submitted to the principal where the intended research is to be conducted.
9. Your research will be limited to the list of schools as forwarded to the Western Cape Education Department.
10. A brief summary of the content, findings and recommendations is provided to the Director: Research Services.
11. The Department receives a copy of the completed report/dissertation/thesis addressed to:

**The Director: Research Services
Western Cape Education Department
Private Bag X9114
CAPE TOWN
8000**

We wish you success in your research.

Kind regards.

Signed: Dr Audrey T Wyngaard

Directorate: Research

DATE: 30 October 2015

Appendix Removed Due to visible signature

Appendix Removed due to visible signatures

2006), based on the Association of the British Pharmaceutical Industry Guidelines (ABPI), and Declaration of Helsinki 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.

HREC 699/2015

Appendix Removed due to visible signatures