

Young adults' perceptions of the implications of their hereditary visual impairment: A Cape Town based study

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ATHLONE SCHOOL FOR THE BLIND



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Signature...

Signed by candidate

Date.....03 November 2017

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Abstract

In South Africa, approximately 600 000 individuals are visually impaired. Approximately one-third of genetic disorders and syndromes involves the eye, including conditions such as congenital cataracts, glaucoma, albinism, and retinal degenerative disorders. The transition into adulthood of visually disabled individuals is a crucial time, as it lays the foundation for their future. The aim of this research was to explore the level of understanding, perceptions and lived experiences of young adults aged eighteen to twenty-three who are visually impaired due to a genetic cause.

A qualitative design, utilizing a phenomenological approach was used for this study. Fifteen participants were recruited through Athlone School for the Blind, the League of Friends of the Blind and Retina SA. In-depth interviews were conducted and data obtained was analysed using thematic analysis. Five themes were identified through this approach indicating the implications of having a genetic visual condition as perceived and experienced by these young adults.

Most of the young adults experienced difficulty in understanding their condition and the genetic bases thereof and they had a strong desire to obtain clarity and knowledge via genetic counselling. The community was thought not to understand their situation either. They were unsure of the inheritance risks to future offspring and some indicated that they felt that this was a gamble they were unwilling to take, whilst others would have children. In some instances, their own visual impairment might create obstacles to raising children. Social interactions were greatly impacted and they felt isolated and tried to avoid unpleasant treatment, stigmatization and pity from the community. Intimate relationships were also noted as a challenge. Mobility is a major obstacle due to the incapacity to drive, as well as the lack of disability user-friendly public transportation and a daunting environment. They want to and feel that they can be independent and achieve the same things as sighted individuals, but society and life circumstances often create barriers to this.

This research could assist in providing information to create more efficient, patient-centred genetic services and might be informative to various organizations about targeted support to provide these individuals and methods to assist their transition to adulthood.

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List of Terms and Abbreviations

AD	Autosomal Dominant
AR	Autosomal Recessive
CC	Congenital Cataracts
DNA	Deoxyribonucleic Acid
GC	Genetic Counselling
LOFOB	League of Friends of the Blind
OCA	Oculocutaneous Albinism
P	Participant
PGD	Preimplantation Genetic Diagnosis
RE	Refractive Errors
RDD	Retinal Degenerative Disorders
RP	Retinitis Pigmentosa
SA	South Africa
UCT	University of Cape Town
Visually impaired	Moderate VI, severe VI or blind
VI	Visual Impairment
WHO	World Health Organisation
XLD	X-Linked Dominant
XLR	X-Linked Recessive

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Chapter 1: Introduction

Visual impairment has a big impact on both the health sector and the individuals living with it. At the critical period of transition from an adolescent environment to an adult environment, individuals with hereditary visual impairments transfer from being dependent on their families to becoming full adult participants within the community (Clark & Hirst, 2007). Successful navigation into this new life stage is not only crucial for young adults, but also for society in general. Transitioning into the adult domain includes moving from school to work, from home to the community, from paediatric to adult healthcare, all of which add to the perception of young adults that a satisfactory life comprises of education, a career, friendships, marriage, parenthood and the ability to make their own decisions and execute them independently (Clark & Hirst, 2007).

This stage of life is crucial in setting the foundation for adulthood in people with visual impairments. It is important for genetic counsellors to learn more about these young adults, as they will need to provide them with information regarding the genetic aspects of their condition, risks for future offspring, and to provide emotional support. By gaining insight into the perceptions of young adults living with hereditary visual impairment, genetic counsellors will have a better understanding of pressing matters that need to be addressed, as well as the level at which genetic counselling should be provided.

Genetic research is continuously evolving, and many methods have been/are being developed to identify the causes of hereditary visual impairment. As our ability to understand the genetic origins of visual disorders increases, it is necessary to acquire more insight about how young adults perceive their disorder and the way it will affect their lives. Research within this field is limited, especially in the South African context. It is therefore anticipated that by not only exploring the origin of a visual disorder, but also the perspectives and experiences of young adults living with a genetic-based visual impairment, efficient health services, management and possible future treatments that will enhance the quality of life of these individuals can be developed.

1.1 Aim

The research aimed to explore the level of understanding, perceptions and lived experiences of young adults aged eighteen to twenty-three who are visually impaired due to a genetic cause.

1.2 Objectives

- To recruit young adult participants who have a genetic-based visual impairment.
- To investigate these young adults' knowledge and understanding of their genetic condition. Such as the way it is inherited, the risk to future generations, the symptoms of the condition, how the individual expects the condition to progress and what healthcare is available.
- To ascertain visually impaired young adults' views regarding finding partners and being in relationships.
- To reveal visually impaired young adults' decisions about having offspring.
- To explore visually impaired young adults' views regarding independence.
- To gain insight into young adults' perceptions for the use of genetic counselling, healthcare, education, and organisations for the blind.

Chapter 2: Literature Review

2.1 Introduction

This chapter will review previous literature on factors relating to Visual Impairment (VI) and disability. The epidemiology of vision loss, the impact within South Africa (SA), the structure of the eye, genetics of VI, models of disability and psychosocial aspects such as procreation, social interactions, independence, mobility, employment and psychosocial wellbeing will be discussed. Furthermore, the importance of healthcare and genetic counselling for individuals with VI will be described.

2.2 Epidemiology of Vision Loss

“Blindness is defined as a corrected visual acuity of less than 3/60 or corresponding field loss to less than 10 degrees in the better eye with the best possible correction, whereas low vision is a visual acuity of less than 6/18 but better than or equal to 6/60 or corresponding field loss to less than 20 degrees in the better eye with the best possible correction by the World Health Organisation (WHO)” (Sacharowitz, 2006:140). According to the International Statistical Classification of Disease and Related Health Problems (ICD-10) and WHO, four categories of visual function exist:

- Normal vision.
- Moderate VI.
- Severe VI.
- Blindness.

Moderate and severe VI have been merged to create the term ‘low vision’. The appellation VI is comprised of low vision and blindness (Maake & Odunan, 2015).

Worldwide, approximately 37 million individuals are blind, a 0.7 percent global prevalence. Another 217 to 246 million have low vision, of which 110 million are faced with severely reduced vision which elevates their risk of becoming blind. It has been noted that VI is more prevalent in developing countries, where roughly 90 percent of individuals with vision loss are

found (WHO, 2017; Oduntan, 2005). Many individuals with low vision require sight restorative measures such as surgery and spectacles to restore their vision (Gilbert & Ellwein, 2008).

To abolish the occurrence of avoidable blindness, a global initiative called “Vision 2020: the Right to Sight”, was launched in 1999 by the WHO and the International Agency for the Prevention of Blindness. The current emphasis is on five conditions, namely cataracts, Refractive Errors (RE), vitamin A deficiency, trachoma and onchocerciasis (Sacharowitz, 2006).

Children and youngsters who are born blind, or who acquire visual loss, have many years to experience the psychosocial and economic implications related to their condition (Gilbert & Foster, 2001). VI has a variety of implications such as decreased quality of life as well as lower employment and educational opportunities (Maake & Oduntan, 2015).

2.3 Vision Loss in South Africa

In 2002 South Africa (SA) was included as a sub-region in a study along with other African countries, namely, Congo, Kenya, Ethiopia, Central African Republic and Tanzania. In this region, approximately 10 million individuals had low vision and 4 million were blind. In 2001 SA had a population count of 44 819 778 million, of which an estimated 2.3 million (5 percent) had a disability. The term disability was defined as *“a physical or mental handicap which has lasted for 6 months, or expected to last at least 6 months, which prevents the person from carrying out daily activities independently, or from participating fully in educational, economic or social activities”* (Sacharowitz, 2006: 141). Approximately 600 000 (1.3 percent) of the 2.3 million (5 percent) were found to be visually impaired, forming the primary disability group (Sacharowitz, 2006). The Department of Health issued a national guideline to prevent vision loss in SA, which noted that the country had a 0.75 percent prevalence of vision loss. In addition, 80 percent of the instances were preventable. Uncorrected RE are one of the reasons that many individuals remain visually impaired. This might be due to an incapacity to access facilities, the deprivation of eye-care services and staff, or not being able to afford them (Maake & Oduntan, 2015).

2.4 Structure of the Eye

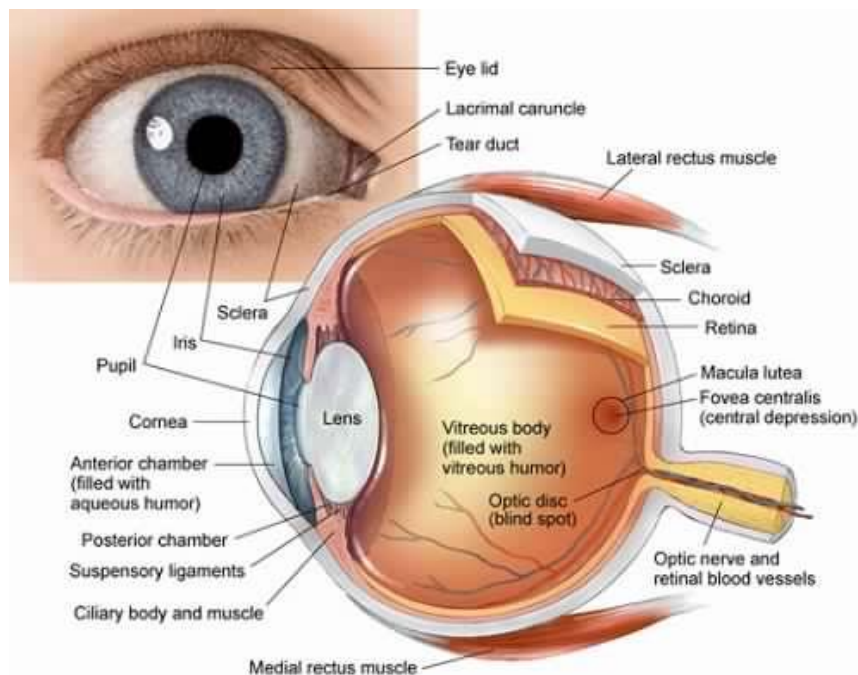


Figure 1: Structure of the eye. (<http://www.biographixmedia.com/human/eye-anatomy.jpg>)

The eye consists of three layers (fig 1). The outer layer is the fibrous coat, comprising of the sclera, which is the white segment of the eye that assists in maintaining its shape and the cornea, which is the transparent surface that refracts light into the eye (Webster & Roe, 2003).

The middle layer, known as the vascular pigmented coat, absorbs light and prevents light signals from scattering. The ability to focus on objects is conducted by the ciliary muscle, by means of adjusting the shape of the lens. The anterior chamber of the eye is filled by a watery substance produced by blood vessels in the ciliary, known as aqueous humour. It provides nutrients to the lens and cornea. Vitreous humour is a clear gel substance that assists in maintaining the spherical shape of the eye and the position of the retina. The coloured part of the eye, known as the iris, is located behind the cornea and controls the amount of light entering the pupil, by means of contraction and dilation (Webster & Roe, 2003).

The nervous coat is the inner layer of the eye. The retina, which is at the back of the eye, consists of photoreceptors and photosensitive components. Rods and cones are the two types of photoreceptors. Rods sense shape, movement and weak visual stimuli, and low light

sensitivity. Cones require more light and identify colour, brightness and finer detail. The section where the optic nerve exits the eyeball is the optic disc, otherwise known as the blindspot. This section has no photoreceptors, thus there is no image detected. In contrast, the macula produces the clearest vision. The fovea only consists of cones and is situated in the centre of the macula. The periphery is important for night vision and consists of rods (Webster & Roe, 2003).

2.5 Genetics of Visual Impairment

According to Mathebula (2012) approximately one-third of genetic disorders and syndromes involve the eye. Some of the visual conditions which the participants from this study have been diagnosed with are discussed below, as well as the various modes by which these conditions are inherited.

2.5.1 Mode of Inheritance

Autosomal Dominant (AD)

AD conditions occur are heterozygous, only one gene in a pair requires a mutation for an individual to be affected with a disorder. The inheritance risk is 1 in 2 (50%) to offspring of an affected parent and can be traced down from generation to generation. However, occasionally, a mutation can be *de novo*, occurring for the first time in the family, and no family history is present. Males and females are affected equally. This form of inheritance can display variable expressivity, which means that a range of signs and symptoms occur in individuals with the same condition. Furthermore, some people have a mutation but do not exhibit any manifestations, which is known as reduced penetrance (Harper, 2010; Turnpenny & Ellard, 2012).

Autosomal Recessive (AR)

In AR inheritance both parents of an affected individual are heterozygotes, in other words they are carriers of the mutation that causes condition. The risk to the offspring of two carriers is 1 in 4 (25%) to be affected or not a carrier and 1 in 2 to carry a single mutation from either parent.

All children of an affected individual will be obligate carriers, but will only be at-risk of being affected if the other parent is a carrier. If an affected individual has a carrier partner, there is a 1 in 2 (50%) chance that their offspring will be affected or be a carrier. Males and females are affected equally. Generally, affected individuals are siblings and observed in one generation (Harper, 2010; Turnpenny & Ellard, 2012).

X-Linked

This form of inheritance is found when mutations are found on the X chromosome. Most X-Linked conditions are X-Linked Recessive (XLR). In this form of inheritance males are found to be affected and females are carriers. There is no male to male transmission, however, all daughters of affected individuals will be obligate carriers. Sons of female carriers have a 1 in 2 (50%) chance of being affected and daughters have a 1 in 2 (50%) chance of being carriers. In rare cases females can be affected. X-Linked Dominant (XLD) conditions are predominantly observed in females, as conditions with such inheritance patterns are lethal in males (Turnpenny & Ellard, 2012; Harper, 2010).

Digenic

Digenic inheritance occurs due to the co-inheritance of mutations in two unlinked genes. The effect of one mutation only is not sufficient to lead to the disease, thus a phenotype will only manifest when heterozygous mutations in two different genes interact (Harper, 2010; Turnpenny & Ellard, 2012).

Mitochondrial

Mitochondria are organelles in the cytoplasm, not the nucleus, and contain DNA which is independent of genomic DNA. Mitochondrial DNA is exclusively inherited from the maternal parent; therefore, mitochondrial disorders are inherited from a female, however, both males and females are affected. Generally, a male does not transmit a mitochondrial disorder, even if he is affected (Harper, 2010; Mathebula, 2012).

2.5.2 Genetic Conditions

Cataracts

Cataracts are the leading cause of blindness worldwide and are classified as congenital, infantile, juvenile, pre-senile and senile. Lens transparency is dependent on structured arrangements of lens fibres and proteins (Figure 2). The variety of symptoms found in this condition include blurred vision, light sensitivity, fading of colours and halos around lights, and eventually leads to blindness. Cataracts can be isolated, associated with other ocular developmental abnormalities or other genetic syndromes. More than 39 disease associated loci have been identified. In hereditary congenital and infantile cataracts AR and X-linked inheritance have been noted, however, in most cases they are due to AD transmission. Whereas age-related cataracts result from a combination of genes and environmental factors (Mathebula, 2012).

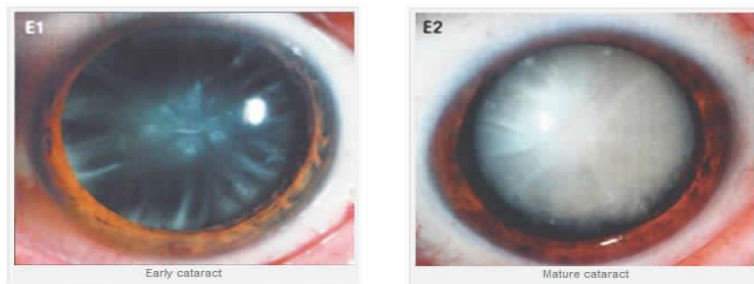


Figure 2: E1 = early cataract & E2 = mature cataract (<http://www.eyedr.co.za/what-is-a-cataract/>)

Glaucoma

Glaucoma is a disease of the optic nerve. The retina sends light generated nerve impulses to the optic nerve, which is then transmitted to the brain. The electrical signals are recognised as vision. Eyes contain a substance called aqueous humour, which normally flows through the eye and is then drained through the trabecular meshwork. A build-up of pressure occurs in the eyeball if any blockages are present in the filtration system. If left untreated, the optic nerve can be harmed or destroyed, which results in blindness (Figure 3). However, up to 25 percent of individuals with glaucoma are found to have normal eye pressure (Bhowmik, *et al.*, 2012). Primary congenital glaucoma occurs before age five, without other abnormalities. When the onset is during childhood or early adulthood it is known as juvenile open-angle glaucoma (Genetics Home Reference, 2017). Individuals with glaucoma experience visual haziness or blurring, severe eye pain, headaches, visual field loss, and see haloes around lights (Bhowmik, *et al.*, 2012). The MYOC and CYP1B1 gene mutations are the contributors to this condition.

Primary congenital glaucoma is typically inherited in an AR manner, whereas juvenile open-angle glaucoma is AD (Genetics Home Reference, 2017).

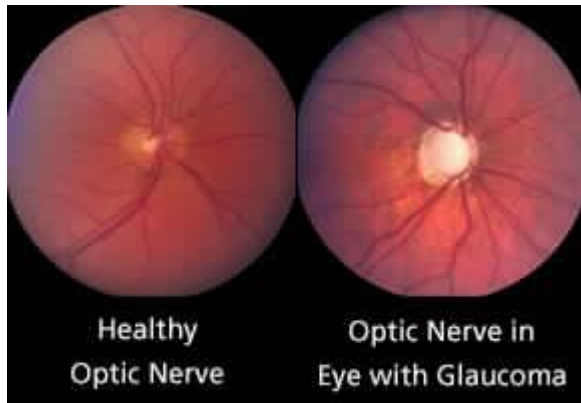


Figure 3: Impact of Glaucoma on eye

(<http://www.langfamilyeyecare.com/wp-content/uploads/Glaucoma-300x212.png>)

Oculocutaneous Albinism (OCA)

OCA is a genetic condition which is characterized by a reduction or absence of melanin in the skin, hair and eyes (Figure 4). Melanin is pigmentation which protects an individual from ultraviolet radiation. The lack of pigmentation in the eye results in uncontrolled pendular eye movements known as nystagmus which causes poor visual acuity. Strabismus and photophobia might also be present. Furthermore, reduction in pigmentation leads to abnormal projection of the visual pathway to the optic cortex and underdevelopment of the fovea (Turnpenny & Ellard, 2012; Manga *et al.*, 2013).

Manga, *et al.* (2013) states that OCA is divided into three subgroups (Figure 4). The condition is inherited in an AR manner. OCA1 is due to mutations in the TYR gene, which makes an enzyme called tyrosinase. It is found in 1/40 000 individuals worldwide and is more commonly found amongst people of Caucasian origin. OCA2 is the most common type of albinism in SA due to the strong prevalence of 1/3900 in affected black individuals. The OCA2 gene, which is located on chromosome 15, produces the P protein. When mutated, it has been found to cause albinism. Some form of pigmentation is present in these individuals. OCA3 is the second most frequent form amongst South Africans, accounting for 1/8580 individuals and is called rufous albinism. It is characterised by reddish-brown skin, deep red hair, and reddish-brown eyes. Ninety percent of cases occur due to two nonsense mutations in the TYRP1 gene, which creates

the tyrosinase-related protein 1 enzyme. All three of the above-mentioned genes are involved in the process to produce melanin (Manga, *et al.*, 2013).



Figure 4: Children with OCA1, OCA2, OCA3

<https://www.bing.com/images/search?q=albinism&FORM=HDRSC2>

Retinal Degenerative Disorders (RDD)

RDD are a group of conditions which can be confined to the macula or the entire retina is affected. Retinitis Pigmentosa (RP) affects the retina and includes disorders that are characterised by degeneration in cone and rod photoreceptors (Sahel, Marazova & Audo, 2015). Up to 80 percent of cases are non-syndromic, however, RP may also be found in syndromes such as Usher syndrome.

In rod-cone dystrophies, loss of rod functioning precedes cone degeneration. Peripheral vision is progressively lost, while central vision remains intact till a later stage (Figure 5). Loss of night vision is amongst one of the first signs and with time many individuals eventually become legally blind. Photopia and photophobia may be experienced. It is inherited in an AD, AR or X-linked manner (Sahel, Marazova & Audo, 2015). According to Sahel, Marazova and Audo (2015) the gene most commonly found in AD cases of RDD is RHO, in AR the USH2 gene is prominent, whereas in X-linked patterns RPGR and RP2 genes are the most frequently associated.

Cone-rod dystrophies are a group of visual conditions in which cone degeneration occurs prior to rod deterioration. Photophobia, loss of colour vision and acuity are the first signs. Thereafter individuals experience scotomas in the centre of the visual field as well as loss of peripheral vision along with night blindness. Approximately thirty gene mutations are associated with cone-rod dystrophy. In AD inheritance CRX and GUCY2D gene mutations are most common,

whereas RPRG mutations are associated with X-linked inheritance (Sahel, Marazova & Audo, 2015).

Stargardt Disease is the most common form of cone-rod dystrophy and causes degeneration of the macula with a deposition of lipofuscin. This leads to central vision loss and decreased night vision (Figure 6). Furthermore, some individuals experience a loss of colour vision and eventually become blind. Generally, mutations in the ABCA4 gene cause this disorder and is inherited in an AR pattern. However, ELOVL4 alterations, which is AD, are also associated with Stargardt Disease (Sahel, Marazova & Audo, 2015).



Figure 5: Retinitis pigmentosa (<http://li129-107.members.linode.com/images/content/about-vision/sim-ret.jpg>)

Figure 6: Stargardt Disease (<http://stargardtdiseaseayurveda.blogspot.co.za/2015/05/stargardts-diseases-possible-ayurvedic.html>)

Myopia

Myopia, otherwise known as near-sightedness is the commonest RE worldwide. It comprises of two categories classified by the degree of refraction error, namely, common myopia with moderate or low RE or high myopia, which can be associated with cataracts, retinal detachment, retinal degeneration and glaucoma. This visual condition causes blurry vision (Figure 7) The condition is multifactorial and has an environmental and a genetic component. It can have an AD, AR, or X-linked mode of inheritance. Myopia has been linked to approximately 70 genetic loci. Amongst others, the COL1A1, COL2A1, PAX6 and a variety of MYP gene alterations lead to myopia (Young, 2009).



Figure 7: Vision with myopia

(https://www.eyes.org/web/Eye_Health/Vision_Conditions/Myopia/web/Health_Eye/Conditions_Folder/Myopia.aspx?hkey=12f285db-5a5d-4bdc-a18b-2a5cb75a5c91).

Peters Anomaly

Peters anomaly is characterised by eye complications of the anterior segment which is comprised of the cornea, iris and lens. The different elements of the anterior segment form independently during the development of the eye. However, in Peters anomaly the cornea, iris and lens fail to separate completely from one another. This causes the cornea to be opaque, ranging from a small spot to a vast area that covers the surface of the eye, resulting in blurred vision (Figure 8). Approximately 50 percent of individuals have low vision at a young age and 25 percent are legally blind. Cloudiness which is centrally placed in the cornea is found to cause poorer vision than opacities which are off-centre. Typically, the condition is bilateral. Individuals might also experience amblyopia, cataracts, glaucoma, and strabismus (Genetics Home Reference, 2017).

The development of the anterior segment of the eye is assigned to the genes FOXC1, PITX2, PAX6 and CYP1B1. When mutations occur in these genes, development is disrupted. Most cases are sporadic and, although the genetic origin is unidentified in many instances, the condition is AD when a mutation in PITX2, PAX6 and FOXC1 is present, whereas a mutation in CYP1B1 is associated with an AR form of inheritance (Genetics Home Reference, 2017).



Figure 8: Peters anomaly

(<https://www.bing.com/images/search?q=peters+anomaly&FORM=HDRSC2>)

Coloboma

Congenital ocular coloboma is associated with the failure of the ectodermal optic vesicle fissure to close creating clefts which can affect various areas of the eye, such as the cornea, retina, iris, optic nerve, ciliary body, lens and choroid (figure 9). The location and size is an indicator of the degree of vision loss. For instance, iris colobomas generally do not lead to severe vision loss, whereas retinal colobomas result in vision loss, typically within the upper part of the visual field. Colobomas are subdivided into categories consisting of coloboma exclusively or those with cysts, microcornea or microphthalmos. Colobomas can form part of a syndrome, such as renal-coloboma syndrome and CHARGE syndrome. Approximately 20 syndromes have been found to be associated with coloboma phenotypes. However, the condition may also be isolated (Kelberman, *et al.*, 2014). This condition can be bilateral or unilateral (Gregory-Evans, *et al.*, 2004). Individuals may also experience cataracts, glaucoma or retinal detachment (Kelberman, *et al.*, 2014).

Coloboma can be caused by various chromosomal abnormalities or alterations to different genes. It is inherited in an AD, AR, or X-linked manner. Over 27 gene loci have been mapped and more than 21 genes are known to cause the condition (Gregory-Evans, *et al.*, 2004). PAX6 and SHH are key genes involved in the formation of colobomas. Other known genes include CHD7 and PAX2, STR46, SMOC1, GDF3, GDF6, RAX and SALL2 (Kelberman, *et al.*, 2014).



Figure 9: Coloboma (http://hybridtechcar.com/wp-content/uploads/2013/09/1379968437_184-2.jpg).

Uveitis

Inflammation of the uvea is known as uveitis. There are four types, anterior uveitis, intermediate uveitis, posterior uveitis and panuveitis. It causes redness, pain, blurred vision, dark spots in field of vision and sensitivity to light (Wakefield & Chang, 2005). Uveitis is polygenic and has complex inheritance patterns. Genes involved include HLA, TNF, Interleukin-1 cluster, IFN, Chemokine and Chemokine Receptor Genes, and CARD15 which has an AD impact (Martin, Kurz & Rosenbaum, 2003).



Figure 10: Uveitis (<http://www.mohaneyeinstitute.org/images/uveitis.jpg>)

2.6 Understanding and Healthcare

Studies have indicated an overall lack of knowledge about conditions such as albinism, resulting in three different outcomes, namely pity, rejection or acceptance of the individuals with the condition (Estrada-Hernández & Harper, 2007). A lack of comprehension of the physical and psychological effects of VI creates negative social support. This is demonstrated by the under- or over-estimation of visually impaired individuals' abilities or restrictions. Some members of society feel that visually impaired individuals are helpless, possibly leading to inadequate understanding of their desire for independence (Verena & Boerner, 2005).

According to Szybowski, *et al.* (2007), when children are diagnosed with a genetic condition, the parents/guardians are generally the ones to receive the medical and genetic information and the situation is often emotionally overwhelming. Thus, it is reported that parents may be unable to comprehend and remember the information at a later stage, and so they cannot convey it to their offspring. Consequently, many young adults have been found to have little knowledge and understanding about the genetic and medical aspects of their condition and how it is linked with their family (Gaigher, Lund & Makuya, 2002).

Making the transition from child/adolescent healthcare, where parents often accompany their children, to adult healthcare can be difficult and leave individuals feeling abandoned and unsure about their future health. Young adults may not want to conform to the setting created by adult health professionals, consequently impacting their future health (Blomquist, 2006). As individuals with visual conditions will need ongoing eye-care and other health services throughout life, it is important to assess a young adult's comprehension of his/her genetic condition. Having accurate knowledge and appropriate understanding of their condition could equip them to effectively interact and communicate with healthcare professionals (Guerette, Lewis & Mattingly, 2011).

In addition, healthcare services, prosthetics and up-to-date technologies are not often available and may be expensive and challenging to obtain. The appearance and types of devices used to enhance aspects such as vision can have extreme psychosocial implications for VI youngsters, some of whom have already been marginalised (Groce, 2004).

Evaluating their information needs can assist with their transition to adulthood (Szybowski, *et al.*, 2007). Another way to fill information gaps regarding genetic conditions is via genetic counselling. It can be described as a psycho-educational process which aims to assist the young adult to understand the genetic condition, so that the information can be used by him/her in a meaningful way to strengthen personal control and diminish psychological anguish, and in this way individuals are usually able to adjust to the disorder better. This could help to minimize challenges to young adults' transitioning to an adult healthcare system (Szybowska, *et al.*, 200; Middleton, *et al.*, 2017). According to Weil (2000), citing Carl Rogers' client-centred

approach, healthcare professionals such as genetic counsellors can assist visually impaired young adults by using unconditional positive regard, empathy and genuineness to make them feel more at ease when providing genetic and medical information about the disorder. This could empower young adults to make more informed choices, anticipate the consequences of their inherited visual condition, and thus have more control about how they manage the future challenges they may face.

2.7 Procreative Beneficence and Decisions Regarding Reproduction

One method by which people create meaning in their life is by having children (Hotke, 2012). However, in the early 20th century, the eugenics movement strongly felt that individuals with various forms of deafness and blindness that may create suffering, should abstain from procreation. This resulted in the sterilization of 375 000 people with hereditary blindness or deafness, and physical deformities, amongst others, which continued throughout the 1900s (Hotke, 2012). While the eugenics movement has been largely discredited (Sandel, 2012), in a study to determine the reproductive choices of parents who have had a child affected with a genetic disease, it was found that 65.5% avoided further pregnancy, whereas 34.5% wanted to have more children (Kelly, 2009). According to Kelly (2009) these individuals experience ambivalence regarding the prospect of having more children due to their perception of the various decisions they need to make and consequences of their choices. The options that these individuals face include not having prenatal or preimplantation genetic testing and taking the risk of having an affected child, having a prenatal test and terminating an affected pregnancy, or accepting an affected child. To avoid the challenges of choosing between disturbing options, a substantial amount of individuals decide not to have future children.

The two methods that can be used to have children free of genetic based conditions are:

- Firstly, prenatal selection, which is performed by undergoing chorionic villus sampling or an amniocentesis during pregnancy (Hotke, 2012). If these tests indicate that the fetus has a genetic abnormality, the pregnancy could be terminated, which, according to the choice on termination of pregnancy act of SA, can be conducted until twenty weeks if medical or psychosocial circumstances permit or after twenty weeks if a

substantial medical reason is identified by healthcare practitioners (Choice on termination of pregnancy act, No. 92 of 1996, 1996:s2).

- Secondly, post-conception selection involves Preimplantation Genetic Diagnosis (PGD), in which embryos are tested *in vitro* and those found to have genetic abnormalities are discarded (Hotke, 2012).

According to the principle of procreative beneficence, when a couple decides to have a child, and there is availability of selection, then they have a moral obligation to choose a child whose life is expected to be best of all the possible options. The notion is that individuals who wish to have a child but are apathetic about whether the child will prospectively have a good quality of life, have a moral flaw (Savulescu & Kahane, 2009).

Sandel (2012), however, argues against this, stating that individuals cannot select their children and therefore parents should not be held accountable for the type of child they have, even if they are somewhat conscientious. For parents to perceive their children as gifts, they should accept them as they are. The greater the capacity to manipulate genetics and the outcome of child bearing, the greater the burden becomes to be perfect and to perform well. Instead, regardless of a child's disabilities, the parents should be willing to nurture and accept him or her (Savulescu & Kahane, 2009). According to critics from a disability rights perspective, assumptions about the quality of life of a disabled individual and how disability affects the family, dictates termination practices (Kelly, 2009).

Genetic conditions generally have an impact on the wellbeing of an individual, therefore, prospective parents may want to obtain all the genetic information about disease susceptibility of their future children (Hotke, 2012), as this awareness and understanding of the inheritance risks is an important contributing factor in making choices (Kelly, 2009). However, according to Smith and Aguirre (2012) many individuals only have a basic understanding of genetic knowledge, which often leads to misconceptions about hereditary conditions. In some instances, individuals appear confused about their chances of having affected children, as they have been given a variety of risk statistics from different informants. This was validated by Winkelstein *et al.* (2010) by looking at a different group of individuals who have conditions such as X-Linked Agammaglobulinemia, in which having a good comprehension of their risks

meant that most males did not feel that their disease influenced their decisions about procreating.

Genetic and medical factors as well as psychosocial considerations all impact individual thoughts about having children. Some experience with impairment might link individual choices to societal discourses regarding disabilities (Kelly, 2009). For instance, Rosenblum, Hong & Harris (2009) found that visually impaired parents are concerned about being able to look after their children. While some had concerns for the child's safety, how to transport the child and the reactions of society, others felt that their disability would teach a child to be more compassionate and accommodating. In addition, Oliver (2004) mentioned that what parents learn about disability from society has an impact on their perceptions and feelings about their offspring being born with an impairment, whereas Winkelstein, *et al.* (2010) suggested that the absence of reproductive burden may also be due to the affected individuals' effective adjustment to their disease.

A study by Petersen (2006) found that some decisions were justified in terms of the prospective wellbeing of a child and fairness towards him/her, while others wanted a family so badly that they were willing to take risks. Despite this, Hershberger *et al.* (2012) states that in their study it appeared that people have a genuine concern for their future children and are aware of the seriousness of their decision, regardless of what they choose.

2.8 Models of Disability

Two models of disability have been proposed: the medical model and the social model.

The medical model attributes the difficulties related to disability on the individual's medical condition alone. Treating, rehabilitating or curing an individual is thought to help them to reach a higher level of 'normality'. The community does not have an obligation to accommodate disabled individuals, instead, they should get medical assistance to adjust themselves to present circumstances. Once all medical concerns have been addressed, all the other problems will disappear (Sullivan, 2011).

On the other hand, the social model of disability, theorises that society incapacitates disabled individuals. It is said that disabled people do not experience social exclusion due to their impairment, but by the feedback of society towards people with impairments. In 1988, an organisation called “A Wider Vision” advocated for the usage of this model to provide services for visually disabled individuals. The social model refuses to view particular challenges in seclusion from the entirety of disabling environments. For example, interventions for unemployment do not only focus on how the labour markets operate, but also involve aspects such as education, transportation and culture. In addition, the attention is on problems caused by barriers and incapacitating environments, instead of functional limitations. Individually based interventions, such as medical, educational or rehabilitative are not seen as counterproductive when approving this model (Oliver, 2004).

2.9 Psychosocial Aspects of Visual Impairment

Young adults with disabilities have the same hopes and aspirations as their peers. They also want to obtain education, gain financial status, live independently and have meaningful relationships.

2.9.1 Social Functioning and Relationships

Visually impaired individuals may be more isolated and have reduced social networks as many sighted individuals are unsure and uncomfortable about communicating with people with disabilities, and thus tend to exclude them from social activities (Hersen, van Hasselt & Segal, 1995). Prejudices from peers create barriers to forming friendships and partially or non-sighted youngsters are often teased and bullied. Despite these issues, young adults with disabilities regard friendships as an important aspect of life (Wiegerink *et al.*, 2006).

It is noted that communication breakdown, due to the reduced ability to identify non-verbal cues and see objects which are referred to, is often experienced in conversations. Individuals become increasingly demotivated to remediate such situations, subsequently they withdraw from social situations (Hodge & Eccles, 2013). Thus, Wiegerink *et al.* (2006) notes that young adults without disabilities tend to become increasingly socially active as they mature, while those with impairments have been found to become less active. This is further validated by

Wallhagen *et al.*, (2001)'s statement that a decline in mobility may lead to reduced social interaction, leading to social isolation and loneliness. This has a negative impact on one's psychosocial wellbeing which in turn may lead to a reduction in mobility and an amplification of social isolation (Wallhagen *et al.*, 2001). A generally positive finding is that relations with parents and siblings have been reported to be good, however, in some instances close relationships with parents have been found to hamper the shift to peer-centred relationships (Wiegerink *et al.*, 2006).

Many reasons can contribute to visually impaired people's challenges in having relationships, including the condition itself, stereotypes about disabled people, or even the fact that visual indicators are used by some sighted individuals to determine someone's interest in having a relationship, and thus might misconstrue the intentions of those who are visually impaired (Fichten, *et al.*, 1991).

Young adults can acquire interpersonal skills via dating and interacting with peers and by this means they discover their aspirations regarding future gratifying relationships (Wiegerink *et al.*, 2006). However, according to Pinquart and Pfeiffer (2012) this is not always an easy task as some visually impaired youngsters find it more difficult to approach potential partners at social events, particularly as they become increasingly self-conscious about their physical appearance.

Packman *et al.* (2012) found that young adults were anxious about the reaction of their partner/potential partner towards their condition and if they would be accepted. This could be due to the findings in studies such as that of Fichten *et al.* (1991) who noted that when a potential partner was known to have a VI, people tended to have more negative thoughts about that partner, the situation and their friends' reactions. When the public have negative perceptions and attitudes it has the potential to create the illusion that disabled individuals do not have the urge to establish a family, as they are sexually inactive (Priestly, 2001). Thus, challenges in forming and upholding romantic relationships are encountered amongst young adults with VI. (Gold, Shaw & Wolffe, 2010).

During a study conducted on individuals with albinism it was found that most individuals would not want a partner with the same condition. The reason for this did not include the factor

of genetics and producing affected children, but instead that they were unsure if they could support a family as they would likely struggle to find employment and that it would potentially amplify the problem. Those who wanted a partner with the condition, did so out of fear of not being accepted and loved by individuals without albinism (Gaigher, Lund, & Makuya, 2002).

Despite these limitations, gratifying intimate relationships have been found to emerge between visually impaired individuals and amongst sighted and visually constrained individuals (Fichten *et al.*,1991). This is particularly valid in instances where individuals have a positive self-esteem and believe that they are approachable (Wiegerink *et al.*, 2006).

2.9.2 Independent Living

The ability to live independently is a challenging, yet key target for many disabled individuals. Arnett (1998) notes that an adult status is defined as having financial independence, living in their own home and taking personal responsibility, by young adults. This is in line with a study by Clark and Hirst (2007), in which visually impaired young adults said that although they were content to live at home with their parents, most of them would like to live independently. Those who had moved out of their parents' home had no intention of moving back. Living independently can be very challenging for some visually disabled people and some individuals fear that they will not manage alone. A fraction of individuals wanted to be in residential care, as they felt it was a good opportunity to establish friendships. Due to similar views people with disabilities established the independent living movement in the 1970s to signify their right to be independent within society (Hendey & Pascall, 2001).

At times disabled young adults are treated as children for a prolonged period by certain family and society members, since they are perceived as vulnerable and have special physical needs (Hendey & Pascall, 2001). It is assumed by the above-mentioned individuals, that these youngsters should remain at home and be dependent on others. In countries such as Iceland, there is no expectation for young disabled individuals to reach adulthood, thus providing them with education, social interactions or preparing them for this new life phase seems unnecessary. Consequently, in cases where individuals are unable to make independent choices, it becomes increasingly unlikely that they will develop responsibility (Groce, 2004).

In other locations, such as the United States, acquiring daily living skills is such an important asset for visually impaired individuals that it has been integrated into the core curriculum for students. These include skills such as housekeeping, meal preparation, caring for clothing, organization, money management, and social skills. While investigating the level of independence of youngsters with visual restraints, it was found that although visually impaired children could do some tasks such as setting the table, washing and fetching snacks, they were incapable of sweeping, folding or hanging laundry, making their beds, or preparing a basic meal at the same level as sighted children. A possible reason for this could be that due to lack of vision, these youngsters might not understand how to perform the tasks and without opportunities to practice or instructions on how to successfully engage with these duties, a delay in competence occurs. This creates foreseeable obstacles for young adults who wish to become independent (Lewis & Iselin, 2002). Another aspect which could possibly impact independence is the fact that some young adults have progressive vision loss, which means that gradually they become less capable of functioning alone and become dependent on family or friends (Clark & Hirst, 2007).

With regards to education, Reed and Curtis (2012) noted that amongst Canadians, the proportion of youngsters with visual disabilities who had degrees or diplomas correlated with the general population, which is an indication that they have the potential to be as capable as their sighted counterparts of successfully completing their studies. Prior to obtaining higher education some visually impaired youngsters believe that they will not obtain satisfactory levels of social and academic support, thus they are apprehensive to pursue further schooling. As it generally takes visually disabled students up to five times longer to prepare educational materials and with a high reading load, these individuals face multiple challenges. For those who struggle, the large and demanding workload often leads to eye strain and headaches. Another barrier is working in groups, as at times sighted individuals treat those with disabilities differently (Reed & Curtis, 2012).

It has been suggested that independence should not be defined with regards to physical capacity, but instead the control over one's own life. Therefore, the emphasis is on people formulate their own decisions and not necessarily whether they are able to implement it themselves (Priestly, 2001).

2.9.3 Employment

SA has implemented legislations and policies such as the Promotion of Equality and Prevention of Unfair Discrimination act and the Employment Equity act, to overcome labour force barriers which disabled individuals face. This ensured that in 2007 approximately 43 716 individuals with disabilities had temporary, part-time or full-time employment. However, this still implies that only 1.8% of the disabled society were employed (Maja, *et al.*, 2008). Although government policies are trying to equip disabled individuals to manage in an inflexible working environment, they do not modify the way work is executed, for it to become more accessible (Oliver, 2004). Even though there are instances where job reservations are made, often career advancement does not occur (Arora & Shetty, 2014). This illustrates the ongoing challenges faced by many of these individuals in establishing an occupation.

Globally, being unemployed, underemployed or earning a low salary is a challenge faced by a significant group of young adults with disabilities. Disabled adults encounter unemployment rates which are approximately 40-60 percent higher than for their non-disabled counterparts, however, this fluctuates in some countries (Groce, 2004). Within the work field, the nature and type of employment is much more limited for partially sighted and blind individuals than for those with normal visual capacity.

Generally, employers tend to be apprehensive about employing visually impaired youngsters, as they are afraid of possible impediments in attaining full productivity (Shaw, Gold & Wolfe, 2007). To combat this, it is helpful to have good insight into one's visual disability when making decisions about a future career, because being able to communicate information about the disorder might empower the individual to assure an employer of their potential (Guerette, Lewis & Mattingly, 2011).

Inaccessible public transportation and facilities have prevented people with disabilities from being hired by some of the prestigious organisations in SA (Maja, *et al.*, 2008). In addition, during the transition to adulthood, employment accessibility affects self-identity and obtaining a salary frequently influences social identity. Being unemployed means, one becomes financially dependent on others, which leads to a decrease in control over relationships. These factors can be concerning to visually impaired individuals as they begin to prepare for the working environment (Priestley, 2001). However, a study by Shaw, Gold and Wolfe (2007)

concluded that, although young adults encounter barriers such as employer attitudes, restricted resources, job requirements and lack of tolerance by colleagues and the public, they were optimistic that they would conquer these obstacles

2.9.4 Mobility

Mobility is challenging on many levels for visually disabled individuals. While finger touch can be used to obtain the necessary information to move around the home, this is usually not the case for outdoor mobility. The terrain can become a hurdle while walking around when there are potholes, uneven surfaces, steps and crowds. Furthermore, crossing streets is daunting for those who are unable to see (Arora & Shetty, 2014).

Being able to obtain a driver's license and drive often represents independence and contributes to quality of life. Thus, employment, social interactions and routine daily activities are, at time, impacted by restrictions on driving (Owsley & McGwin, 1999). Depending on others for transportation can be frustrating and in developing countries, public transportation is often a large asset in providing mobility. However, announcements are not always made during the journey nor at the various stopovers and often busses do not stop at the designated areas, which can result in individuals being unable to estimate where they are or need to be throughout their journey. The differences in height of bus steps and train station platforms are also challenging because disabled individuals rely on touch and memory to navigate (Arora & Shetty, 2014).

Printed bus and train schedules or bus stop signs are often not useful to the visually impaired as they are too small to read. They also cannot see route numbers or names on busses when they are near, let alone when they are at a distance. Even though it is very useful when such information has been enlarged, visually impaired individuals still sometimes find it challenging to read or decipher. This is particularly the case for blind individuals who use braille as a means of reading. For those who are unable to read efficiently, the preferred means of obtaining information about transit operations are auditory messages (Golledge, Marston & Costanzo, 1997).

2.9.5 Psychological Wellbeing

An essential factor regarding the quality of life of young adults with visual impairments is the extent to which they can cope with their reality. Depression is quite common amongst individuals who have vision loss. It is said that the extent of visual degeneration does not influence prosperity. However, studies have found that those who are more independent have fewer indicators of depression. Social support is an effective source to minimize depression and other mental or emotional reactions, but overprotection can lead to a decline in mental wellbeing and non-existent or negative support exacerbates the process of depression (Papadopoulos *et al.*, 2014).

2.10 Chapter Summary

In chapter 1 and 2 different aspects of literature were reviewed, the motivation for the study was highlighted and the aim and objectives were described.

Chapter 3: Methodology

3.1 Introduction

In this chapter, the methodological framework used to conduct the research will be described. This includes the research design, participant population and size, and the research setting. Furthermore, the data collection and analysis process will be outlined. The chapter will conclude with a description of the ethical issues that were taken into consideration.

3.2 Research Design

This study is based on a qualitative design, utilizing a phenomenological approach. According to Finlay (2011:8) “*qualitative research illuminates the less tangible meanings and intricacies of our social world*”. It takes place in a natural setting, where the researcher does not implement any form of manipulation. Qualitative research focuses on discovery and understanding (Macfarlane, Veach & LeRoy, 2014). It examines the way individuals construct personal meaning, amplifies the voices of respondents, and describes the diversity of their experiences (Bless, Higson-Smith & Sithole, 2013). Such meanings may include the respondents’ thoughts, feelings, notions and actions (McMillan & Schumacher, 2001). It allows healthcare professionals to explore what health and illness means to individuals by investigating their experiences (Finlay, 2011). Qualitative studies compliment the field of genetic counselling, as the clinical practice revolves around interacting with individuals around multifaceted and sensitive topics, and the skills used are similar to those needed in conducting interviews (Macfarlane, Veach & LeRoy, 2014).

Phenomenology is a common qualitative approach utilized in healthcare (Al-Busaidi, 2008), where occasions or situations are described by the researcher through an individual’s experience (Finlay, 2011). This requires implementation of bracketing, a process by which any prejudices the researcher might have about the phenomenon are eliminated (Al-Busaidi, 2008).

No follow-up interviews were required in this current study and all participants were interviewed within a period of three months. Unlike longitudinal studies, this form of research is more appropriate for this postgraduate degree. However, the disadvantage is that no accounts can be made of data changing over time.

3.3 Research Population, Sample and Setting

3.3.1 Research Population

Young adults who have VI associated with or because of genetic factors were recruited for this study. The individuals have VI as either the primary result of genetic factors, such as CC and RP, or due to eye abnormalities manifesting in association with genetic conditions such as albinism. Individuals could participate in the study on the grounds that they were eighteen to twenty-three years of age at the time of the interview, as this age group targets people who are in the first few years of young adulthood, when their period of transitioning from adolescence to adulthood is prominent. The visual ability of the individuals is within a spectrum from limited vision, with the capacity to see, to legally blind.

Individuals who are visually impaired due to non-genetic factors were excluded from the study. To ensure that the participants do not unexpectedly or unwittingly learn that they have a genetic visual condition during the interview, and possibly get traumatised by the information, they needed to know that their VI is due to a genetic factor in order to participate. To maintain a non-biased stance, no preference was made with regards to gender or ethnicity.

3.3.2 Research Setting and Recruitment

Initially four institutions were identified as suitable sources for recruiting participants, however only three of them agreed to get involved in the study. Namely:

- Athlone School for the Blind: a boarding school for children with visual impairments. This was the main institution used, as a relationship had already been formed between the school and the University of Cape Town (UCT) by means of different research being conducted (HREC REF: 226/2010).

- The League of Friends of the Blind (LOFOB): a non-governmental organisation for visually impaired individuals.
- Retina SA: a non-profit organisation focusing on retinal degenerative conditions.

The school and LOFOB host individuals that are visually impaired due to a wide spectrum of reasons, which include environmental and genetic factors.

Meetings were held with each institution to discuss the research and method of recruitment. The researcher addressed members of Retina SA at their 2016 annual general meeting, where a brief outline of the research was given and an invitation was extended to any individuals who might be interested in participating. Permission was obtained from Athlone School for the Blind and LOFOB to source participants from their institutions and to conduct the research on their premises (Appendix E). A nurse at Athlone School for the Blind went through the data folders of individuals to identify who was suitable for the study. A meeting was held in which potential participants were briefed on the research study and invited to participate. On the day of the interviews the researcher was given the folder to read, to ensure that inclusion criteria (discussed below) were met. LOFOB opted to recruit the participants without input of the researcher. They were supplied with information sheets and consent forms to distribute within their organisation. They then went through their records to identify any potential participants and approached them with the study information. Braille forms were developed for blind individuals who preferred to read the documentation themselves, instead of having somebody read it to them. The forms were also available in English or Afrikaans depending on the participants preference.

Interviews were held at the various locations or at a place specified by the participant as the setting is known to influence the information divulged by the respondent (Marshall & Rossman, 1999). The aim was to ensure that they felt comfortable and familiar with the environment. This was particularly important in this instance as navigating the environment takes time, effort and can be somewhat challenging for some visually impaired individuals.

3.3.3 The Sample

Purposive sampling was used for this study. Participants who meet the predetermined criteria set by the researcher, are selected, as they would be suitable to address the research question (Given, 2008). The individuals should have experience with or be well-informed about a specific phenomenon and convey it in an expressive and insightful way (Palinkas *et al.*, 2015).

Seventeen participants were sourced for the study, however two of them were excluded from the research due to either not meeting age requirements or having VI due to a non-genetic cause. Thus, the final sample consisted of fifteen participants. Fixed guidelines have not been instated for the sample sizes used within qualitative studies, but instead it is determined by factors such as the duration within which the research needs to be conducted and the depth of the interview (Al-Busaidi, 2008). In qualitative studies, the goals are focused on in-depth and highly contextualized understanding of phenomena, which is well suited to small samples.

More importantly than the size of the sample, is obtaining saturation. Data saturation is achieved when no new information emerges during the data collection process. Indicating that it is not necessary to collect more data (Given, 2008). In qualitative research data saturation is more difficult to obtain than with quantitative research, thus complete saturation in this instance was not entirely possible. After the initial interviews were conducted, a period was given in which any other potential participants could opt to participate. Then the researcher inspected the interviews to determine if more data was required. Once this process was complete and no other willing individuals surfaced, it was concluded that enough data was collected. Thus, in qualitative research, sample sizes are smaller than those in quantitative studies (Al-Busaidi, 2008)

3.4 Data Collection

Thirteen of the interviews were conducted at Athlone School for the Blind, one interview took place at LOFOB and one at the Department of Human Genetics at the UCT. A private and quiet room was used that created an environment in which the participant could feel comfortable and safe.

Prior to the commencement of the interviews, the interviewer made sure that the participants met the required inclusion criteria and a consent form was read to them, which highlighted aspects such as the aims of the study, the need for audio recordings and the measures taken to ensure confidentiality, for informed consent to be taken (Appendix A). A copy of the signed consent form was given to everyone who was interviewed. Thereafter they were required to complete a short questionnaire consisting of ten closed-ended questions (Appendix C). The purpose of the questionnaire was to obtain socio-demographic information which was used to assist in understanding the phenomena. Due to visual constraints, the participants opted to have the questions read to them, instead of being given the form to complete by themselves. The researcher practiced cultural sensitivity by determining if she could ask about each participant's ethnicity. Starting the process with basic questions assisted in establishing a form of rapport prior to discussing sensitive information (Denscombe, 2003).

The next step of the data collection process was to conduct face-to-face interviews using an interview guide (Appendix D). Face-to-face interviews are beneficial as they allow the researcher to detect various social cues. The purpose of the interviews was not to obtain specific answers or to test a hypothesis, but instead to gain insight into the experiences and perceptions of visually impaired young adults. In this study, it allowed the researcher to make observations on both the non-verbal and verbal behaviours of the participants, which further enriched the data (Opdenakker, 2006). The researcher conducted all the interviews personally.

Many phenomenological studies use in-depth interviews as a means of data collection (Finlay, 2011). Legard, Keegan and Ward (2003) agree that one of the primary techniques used in qualitative research is in-depth interviews. Therefore, this study utilised this interviewing style. In-depth interviews allow participants to provide thorough accounts of their experiences, while the researcher has the flexibility to probe interesting areas that emerge. Conducting interviews which do not have a rigid structure, nor complete uncertainty allows the researcher to obtain in-depth information on the topic of interest without predetermining the outcome. In-depth interviews consist of open-ended questions and probing instead of closed-ended questions which result in short and direct answers (Given, 2008). Thus, the interview guide consisted of open-ended questions, which allowed the participants to divulge information to whatever extent they felt comfortable with, while permitting the researcher to ask probing questions to

elicit additional data if necessary. The questions were designed to allow the interviewee to express his/her experiences and perspectives extensively (Turner, 2010). The open-ended questions provide an opportunity for participants to use their own words to communicate their views, feelings and experiences about a situation, without creating boundaries (Terre Blanche, Durrheim & Painter, 2006).

The interview guide was reviewed by three independent supervisors, namely Professor Greenberg, Ms Leisegang and Ms Dusterwald to ensure validity and determine that all necessary questions were included. The quality of a study is improved by means of test studies (Hazzi & Maldaon, 2015). Therefore, a mock interview was conducted with a co-supervisor, Ms Leisegang, for the researcher to become acquainted with the interviewing process and then a test interview was conducted to determine if there were shortcomings or errors in the interview design (Turner, 2010). Thereafter, appropriate adjustments were made, such as refining a few of the questions and identifying additional issues to explore, before continuing with data collection.

The participants were given the option of doing the interview in English or Afrikaans as the researcher is competent in both languages. If another language was requested, arrangements would have been made to include a translator in the interview, however this was not necessary as all the interviewees were satisfied to participate in the two languages noted above. The interview sessions proceeded for a minimum of twenty-five minutes to an hour and were voice recorded, which permitted intense observation and engagement between the participant and the researcher, instead of rigorous note taking. However, a few notes were taken throughout the interviews to complement the recordings.

3.5 Data Analysis

An alpha numeric code from P1 to P15 was assigned to each person to ensure confidentiality. All names that were mentioned during the interview were replaced with fictitious names for anonymity for the individual named as well as the possibility that the name might result in the identification of the participant. During and after the interview, notes were taken, which

highlighted observations regarding the setting, participant's nonverbal behaviour, and any other factors which might have had an influence on the interview.

The interview recordings were transcribed verbatim by the researcher. This process occurred concurrent to the interviews. After the first three interviews were transcribed, one of the co-supervisors inspected them to discuss and provide guidance where needed. There were instances in which a few of the participants and the researcher spoke in Afrikaans during the interview. These sections were transcribed in Afrikaans and then translated to English. To ensure that no distortions occurred during this process, back translation was performed, in which a bilingual individual translated the English back to the original language.

In qualitative research, the goal of analysis is to describe and interpret participants' perspectives (Vaismoradi *et al.*, 2016). Raw data alone is not informative and further steps need to be taken to organise the material into descriptive and meaningful data (Creswell, 2012). Thematic analysis was used for this study. It is deemed to be a functional research instrument which supplies a comprehensive account of the data. The advantage of thematic analysis is that it is not strictly bound to a specific theoretical framework, which means it can provide more attainable types of analysis. The analysis process is not linear and therefore one might move back and forth through the phases as it gradually develops (Braun & Clarke, 2006). This method was selected for the researcher to implement content extraction, coding, and to create themes which are used to portray the individuals' experiences of hereditary VI.

The first part of analysis is to familiarise oneself with the data. To accomplish this the researcher immersed herself in the collected data by listening to the recordings and repeatedly reading the data. Listening to the recordings was a useful tool to highlight aspects such as vocabulary, tone and way the participant interacted throughout the interview, which can get lost when investigating the transcripts only (Braun & Clarke, 2006).

The next phase was to generate codes and categories. *"Codes identify a feature of the data that appears interesting to the analyst, and refer to the most basic segment, or element, of the raw data or information that can be assessed in a meaningful way regarding the phenomenon"*

(Braun & Clarke, 2006:18). Themes and sub themes were created from grouping the codes together. Connections were formed between the data and prior literature findings. By creating associations between concepts and various literature the researcher could investigate how the data interacted, in hopes of discovering the greater meaning of the data.

The researcher found it more beneficial to code manually by means of making notes, labels and highlighting sections. Therefore, coding and data analysis was conducted by highlighting information via colour codes or creating margin notes. Large posters were created which contained the different themes, whereby various information from the transcripts were added and connections were made amongst certain topics.

3.6 Validity and Reliability

For a study to be useful, those who are engaging in it must trust in its sincerity. The way in which data is gathered, investigated, interpreted, and presented can combat the concerns related to validity and reliability (Merriam, 2009). In many instances, it has been noted that the terms reliability and validity are better suited for quantitative studies and have been exchanged with the terms trustworthiness, transferability, credibility and dependability (Polit & Beck, 2012).

3.6.1 Trustworthiness

Studies which display a comprehensive internal logic and the suitability of the research question, study design, data collection and analysis methods, signifies high credibility, also known as internal validity (Bless, Higson-Smith & Sithole, 2013). One of the strategies to obtain credibility is triangulation. In this study data triangulations, which is indicated by comparing and cross-checking data collected from different locations and from individuals with diverse perspectives, was enhanced by obtaining data from several sources such as individuals belonging to a support group, those associated with a school for the visually impaired and an organisation providing a variety of resources for such individuals (Merriam, 2009).

Furthermore, to increase the trustworthiness of the study, verbatim accounts were created of the audio recordings and these direct quotations have been used to present the research findings

(Bless, Higson-Smith & Sithole, 2013). The process of back translation, as explained previously, assisted in demonstrating the validity of the data.

Reflexivity, which necessitates that a researcher should be aware of their distortions, assumptions and biases regarding the research, and reflect critically on the self as a researcher, should be taken into consideration. In order to ensure that interpretations are valid, ongoing self-reflection should be maintained, thus the researcher engaged in various methods of reflexivity throughout the study, such as creating journal notes and having discussions with the supervisors and research colleagues (Polit & Beck, 2012).

Member checking, a process by which the researcher returns the data back to their participant for affirmation, was considered. If done correctly, the trustworthiness of qualitative data and results is evaluated and confirmed via this method (Birt *et al.*, 2016). However, due to the participants' limited vision, which leads to them requiring the assistance of an external individual to read information, a decision was made not to use this technique as it could lead to ethical and other complications.

The degree to which the findings of the study correspond with similar situations is known as transferability or external validity. The provision of accounts about the researcher, his/her rapport with the participants and the context involved in data collected, permits researchers to assess the corresponding factors within various settings (Bless, Higson-Smith & Sithole, 2013).

3.6.2 Dependability

Dependability, otherwise known as reliability is the degree to which the research findings can be replicated. When a clear research scheme is meticulously described and followed, a dependable study is produced. (Bless, Higson-Smith & Sithole, 2013). To ensure this, all the initial interview questions were asked in an identical manner and all the interviews were conducted by the researcher herself. Furthermore, all the transcripts were coded in the same way.

3.7 Ethical Considerations

3.7.1 Ethical Approval

The study was granted ethical approval by the Faculty of Health Sciences Health Research Ethics Committee of UCT (HREC REF: 376/2016).

3.7.2 Consent

It is very important that participants can make a conscious, informed decision to participate in the study or not. This is achieved by providing them with adequate information about the research, determining their comprehension and indicating the voluntary facet of participation (Polit & Beck, 2012). Prior to obtaining demographic information and the commencement of the interview, each participant was required to give written consent. Due to visual constraints, an option was available to give verbal consent and for a witness to sign on their behalf. This method of witness-signed consent was performed for three of the individuals.

The interviewer read the consent form, which explained the purpose of the study as well as the terms of participation, to each participant. No coercion or persuasion was used to encourage individuals to participate. The individuals could ask questions before agreeing to sign the form and were assured that participation was voluntary, thus they were able to withdraw from the study at any point (Legard, Keegan & Ward, 2003). The consent included the participation in the study and for the interview to be audio recorded.

3.7.3 Confidentiality

Confidentiality stems from the principle of respect for autonomy and ensures a participant that no information will be disclosed without their authorization and their identity will remain unknown to the public (Wiles *et al.*, 2008).

The consent forms, questionnaires, researcher notes and audio recordings were kept locked away and on a password protected computer. The information could only be accessed by the researcher and supervisors. Alphanumeric codes were assigned to each document and recording, to assure confidentiality. Where participants used the names of an individual in their

family, community or of themselves, a fictitious name was assigned to ensure the anonymity of those mentioned. The data will be kept safe for usage in journal publications and will be discarded thereafter.

3.7.4 Risks/Benefits to the Participants

When conducting any research, the benefits to the participants and society should outweigh the potential risks (Polit & Beck, 2012). A few of the interview questions were of a sensitive nature, which could have evoked emotional responses. Thus, the discussion of sensitive information was a potential risk to participants. This risk was minimized by displaying empathy and cultural sensitivity towards the participants and allowing them to withdraw. Provision was made for an individual to be referred to a genetic counsellor or a psychologist, if he/she requests or if the researcher felt that the situation warrants it. By interviewing people who are unaware that their visual disability has a genetic aspect, it could have led to emotional and psychological trauma if they became conscious thereof during the interview. Thus, in order not to breach the ethical rule of do no harm, individuals without knowledge of the genetic component of their visual disability were excluded from the study. It was made clear that they would not benefit financially or medically from the study. A research-related bodily injury risk assessment form was completed and submitted to the Faculty of Health Sciences Research Ethics Committee of UCT, to indicate the lack of physical harm to participants whilst conducting the study. This form indicated that this was a low risk study (UCT: Form FHS022).

In contrast, being able to speak about their feelings and being in a situation where they felt encouraged to share their story and emotions might have had a therapeutic impact. Participants also benefited by receiving the opportunity to ask questions after the interview and were given information regarding genetic counselling services, organisations for visually impaired/blind and support groups if they requested. The long-term benefit of the research is to use the findings of the study to improve genetic counselling and support services for young adults with hereditary VI.

A small financial compensation was offered to the participants for their time, effort and travel expenses. In a few instances, it was requested that a snack would be provided for the participants in place of the money compensation.

3.8 Chapter Summary

This study aims to gain insight on the perceptions and experiences of young adults who have genetic based VI. This is best expressed through thoughts, feelings and narratives; therefore, a qualitative method was implemented. In this chapter, the entire research process was described, from recruiting participants to analysing the data.

Chapter 4: Results

4.1 Introduction

This result chapter begins by outlining the socio-economic and cultural information of all individuals who participated in this study. Thereafter the findings of the study are documented in conjunction with extracts from the interview data.

4.2 Socio-Economic and Cultural Information of Participants

Fifteen participants completed the short questionnaire and answered all ten questions. In Table 1 the socio-demographic information is outlined. Figure 10 illustrates the various conditions found amongst the individuals and Figure 11 outlines the inheritance pattern according to each individual. A summary of the socio-economic and cultural information of the participants can be found in Table 2.

Characteristics	<i>N</i>	%
Gender (<i>N</i> = 15)		
Males	10	66.7
Females	5	33.3
Age (<i>N</i> = 15)		
18	4	26.7
19	7	46.6
20	3	20.0
21	0	0
22	1	6.7
Mean age	19.1	
Ethnicity (<i>N</i> = 15)		
Black	9	60.0
White	0	0.0
Coloured	5	33.3
Indian	1	6.7
Current education level (<i>N</i> = 15)		
Grade 10	2	13.3

Grade 12	11	73.3
University	1	6.7
None	1	6.7
Employment (<i>N</i> = 15)		
Employed	0	0.0
Unemployed	1	6.7
Student	14	93.3
Relationship status (<i>N</i> = 15)		
Single	13	93.3
Informal relationship	2	6.7
Number of children (<i>N</i> = 15)		
None	14	93.3
One	1	6.7
Vision status (<i>N</i> = 15)		
Partially sighted	13	87.0
Blind	2	13.0

Table 1 Socio-economic and cultural characteristics

The average age of the participants was 19 years. The two thirds of the sample were males (10/15) and one third were females (5/15). Most participants were black (9/15), followed by coloured individuals (5/15) and only one Indian (1/15). No white individuals volunteered to participate in the study. All but one participant had no children. One of the individuals had a two-year-old son, who was not displaying signs of a visual condition, this participant is the only one that is not currently part of the education system, and is currently unemployed. Thirteen were in a secondary institution of which nine were completing their final year and one individual was studying at university. Two individuals mentioned that they were in a relationship, the other thirteen stated that they were single. Thirteen of the participants were partially sighted, of which the degree of vision was variable, whereas two were blind. The various conditions of each participant will be outlined below.

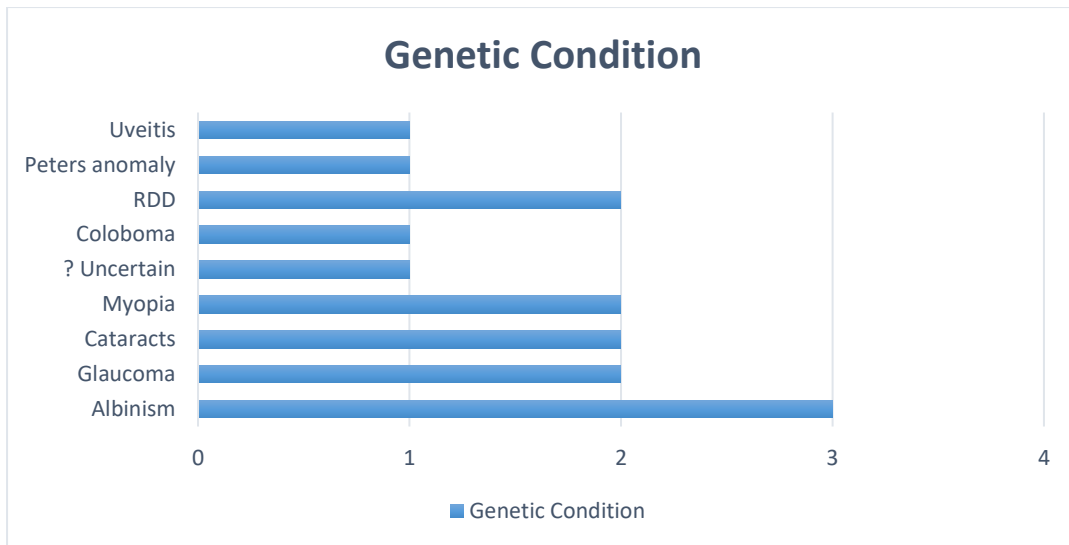


Figure 11: Genetic conditions

Some of the participants were unsure of the exact diagnosis or name of their condition. Others had become aware of it by asking individuals at the organisations they are associated with. In cases where they were unable to give an answer, the information was taken from their folders or documentation containing their personal details. Three individuals had albinism, two had cataracts, two had myopia and another two had glaucoma. RDD was noted in two participants, one of which was unclear as no final diagnosis had been made and the situation was under investigation, while the other individual had cone-rod dystrophy. One youngster had coloboma, one had Peters anomaly, one had uveitis, and the diagnosis one participant was not very clear.

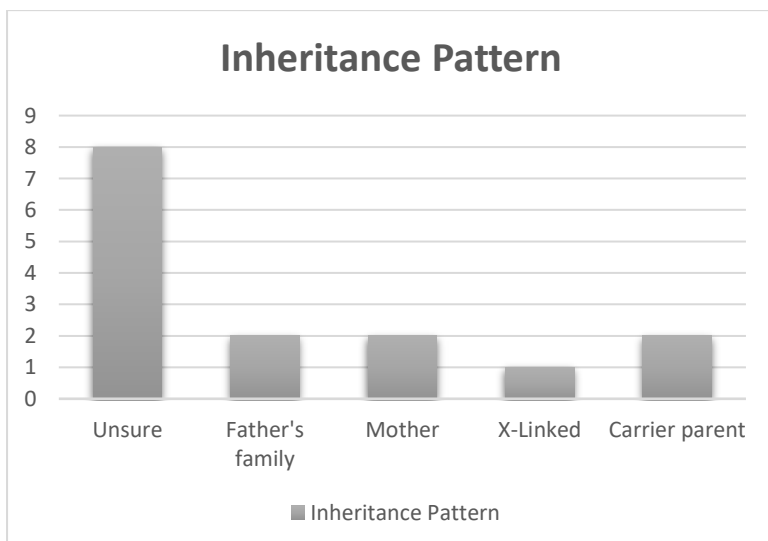


Figure 12: Inheritance pattern

As per Table 2 eight of the participants did not know how they had gotten the condition. One of these had mentioned that she was ‘born with it’ but did not know anything else. Four of the individuals could give an indication from which side of the family they thought the condition came. P1 was the only one who used a genetic term such as X-Linked to define the pattern of inheritance, whereas P7 and P9 didn’t mention a mode of inheritance but did mention that their Albinism came from a carrier parent/s. This information will be explored in more detail in one of the themes below.

P Code	Sex	Ethnicity	Age	Education	Vision status	Genetic condition	Inheritance pattern
P1	Male	Indian	22	University	Partially sighted	Retinal dystrophy	X-Linked
P2	Female	Black	19	Gr 12	Partially sighted	Peters anomaly	Born with it
P3	Male	Black	18	Gr 12	Partially sighted	Congenital glaucoma	Unsure
P4	Female	Black	18	Gr 12	Blind	Bilateral Uveitis	Unsure
P5	Male	Black	19	Gr 12	Partially sighted	Albinism	Unsure
P6	Male	Black	20	Gr 12	Partially sighted	Myopia	Unsure
P7	Male	Coloured	18	Gr 12	Partially sighted	Albinism	Carrier parent
P8	Female	Coloured	19	Gr 12	Partially sighted	Congenital cataracts	Unsure
P9	Female	Black	18	Gr 12	Partially sighted	Albinism	Carrier parents
P10	Male	Black	19	Gr 12	Partially sighted	Congenital cataracts	Father’s side
P11	Male	Black	20	Gr 12	Partially sighted	Cone-rod dystrophy	From mother
P12	Male	Coloured	19	Gr 12	Partially sighted	? Uncertain	From mother
P13	Male	Coloured	20	Gr 10	Partially sighted	Myopia	Father’s family
P14	Female	Coloured	19	Gr 10	Blind	Bilateral coloboma	Unsure
P15	Male	Black	19	None	Partially sighted	Glaucoma	Unsure

Table 2 Summary of socio-demographic information

4.3 Themes Identified

Through examination and in-depth analysis of the transcripts, several themes were identified. Table 3 summarises the themes and sub-themes. All quotes have been documented in English. At times, these might be the translated version of conversations in Afrikaans.

Theme	Sub-themes
Theme 1: Understanding	<ul style="list-style-type: none"> ▪ “I don’t understand who I am” ▪ “My eyes will become weaker” ▪ “They have no clue” ▪ “Without understanding no one will know”
Theme 2: Procreation: What about children?	<ul style="list-style-type: none"> ▪ “It is possible that they have the visual condition” ▪ “Not willing to take the chance” ▪ “My child can live with it” ▪ “How will I raise them?”
Theme 3: Social Interaction	<ul style="list-style-type: none"> ▪ “I am always alone” ▪ “They make fun of me... They feel sorry for me” ▪ “It feels like I am blaming them” ▪ “Going to struggle to get the right one for me”
Theme 4: Mobility	<ul style="list-style-type: none"> ▪ “I Can’t Drive” ▪ “The bus...the train...I can’t see it” ▪ “Moving around is difficult”
Theme 5: Abilities	<ul style="list-style-type: none"> ▪ “It did affect me academically” ▪ “The advantage of a disability” ▪ “I can’t do everything by myself” ▪ “Most of us are handled like eggs” ▪ “I can do what the other people can”

Table 3: Themes

4.4 Theme 1: Understanding

This theme focuses on understanding at a variety of levels. Understanding has emerged to be a core factor within this study. The thoughts and comprehension these young adults have regarding what their condition is, where it came from, as well as how they perceive others to understand their situation plays a large role in determining their view of the implications of their visual conditions in their lives.

4.4.1 “I don’t understand who I am”

When the individuals were asked about their conditions they were all able to explain their symptoms and visual abilities with ease. However, there was a significant amount of confusion about what exactly their condition is and the reason behind it. Some had minimal or no understanding of what their disorder is, apart from the fact that they are unable to see efficiently.

“I don’t understand my condition miss, it is like, I don’t know how to explain it but it is just, I don’t understand. Like I know that if I don’t want to wear my glasses, I won’t see far. That’s all I know miss.” (P10)

The fact that these individuals do not know what their condition is and do not have knowledge about their situation, results in them having many questions, which often are left unanswered and this can lead to a feeling that they have done something which caused the disorder. An internal struggle is faced in which they try to come to terms with their condition by understanding why this has happened to them.

“There are those kinds of doubts that you, that come in your mind, like what I done wrong? Why do I have to have this kind of thing? Why me? You see those kinds of questions. Why this condition cannot go to somebody else, not me?” (P15)

This confusion does not only have an impact on the youngsters’ view of their lives, but it also impacts the way they explain the information to others. If one does not understand one’s own condition, then how is one to explain it to somebody else. Consequently, they resort to providing explanations that provoke the least amount of questioning. This is something that was particularly noteworthy for Participant 4 who became blind during infancy, however, she tells people that she was born blind, because this is an answer which they easily accept.

“I don’t know the genetic disease even. So, I don’t know anything basically about it. I am not aware of what caused it, what caused me to be blind or anything. I just know that I am blind, that’s it. So, I don’t have any information about it. Even when people ask me why I am blind, then I just say I was born blind, because I don’t know the condition of me being blind, I don’t know what happened. Because I was still young, around about four years.” (P4)

A few of the participants did have some understanding of their condition and mentioned that they had conducted research to try and gain insight on the disorder. The three individuals with albinism could explain the condition relatively well and two of them were even able to state that they had carrier parents.

“My understanding is that there is a lack of a certain pigment or something in my skin and in my eyes. That’s what gives me this colour, complexion something and that’s what makes my eyes weak.” (P7)

“They were both carriers of albinism genes.” (P9)

The method by which they acquired the condition was a confusing matter to most. There was a consensus that it is associated with their families, however, a few of the participants such as participant 3 and participant 11 did not understand exactly how this occurred.

“I don’t understand how it is inherited because I double checked and the Internet they said it may be inherited. Which is in my family somebody that had it, it’s not my father but it’s his

father now I think maybe it skipped one generation or two. So, I think obviously now it will skip this generation, maybe that one now maybe, I don't know. I don't get it.” (P3)

“I got it from my mother. I think like she has like the X. The X chromosome she have, it was a small thing and then when it came to me, maybe when they made me it doubled up twice and that's why it gets like extra with me. I think that's the cause.” (P11)

4.4.2 “My eyes will become weaker”

Having a decreased visual capacity not only has a short-term impact, but also creates uncertainty over the future ability to see. Some were unsure what their future vision would entail, whilst others felt that their vision would gradually decrease.

“My eyes will actually get weaker and it has gotten weaker.” (P7)

“Maybe it will be getting worse or getting blind.” (P11)

Some felt that they may eventually become blind, which would create many challenges and they would need to readapt and learn to cope under new circumstances.

“My eyes will become weaker and I think that will be a huge change in my life. I would have to adapt to being blind and all that. I will have to accept that I'm blind and I will have to go to places to relearn alphabets” (P2)

The reason for having weaker vision was thought to be due to the way in which they care for their eyes. Participant 10 mentioned that his eyes are weakening because he feels too embarrassed to wear his glasses. Whilst another participant believes that the preservation of vision will be acquired by means of sufficient eye care and by following management instructions.

“I think that if I like take care of my eyes very well, then it will be fine. But if I like strain my eyes then as years go by they will be weaker.” (P9)

In some instances, awareness that a decrease in vision is possible was an unpleasant thought and provokes anxiety in the individuals when they think about it. Managing with decreased vision was already challenging, yet alone having a weaker or absence of vision in the future.

“I think it will impact my life in a very bad way because I am struggling already to cope with being partially sighted, if I get blind or something like that, I don't know if I can cope with it.” (P12)

Participant 15 is mindful that he has a risk of becoming blind. Having a need to be prepared for such an event, he has started performing activities blindfolded, to become acquainted with what blindness entails and to learn to adjust and cope without vision.

“I will blindfold myself and move around. Just to be in the same... like with everybody. I must have the idea about being totally blind because that’s something that are going to happen in the future.” (P15)

4.4.3 “They have no clue”

Most of the young adults in this study felt misunderstood. They verbalised this at least sixty-one times. As per the statement by participant 10.

“Some people miss, some people they don’t understand miss.” (P10)

Furthermore, family were also not very understanding at times and this was found to create tension or conflict.

“Like many of the people in my family don’t understand me, you see miss. My sisters, maybe I am sitting in ah in front of the tv and then they will shout at me like “why must you do that” and that, because they don’t understand me.” (P10)

In addition, many indirect references were made to the lack of understanding they receive. This indicates the large impact it has on their day to day life and often affects their social interaction.

In different cultures, there are a variety of beliefs regarding disorders and disabilities. These views create frustration, fear and disbelief amongst the youngsters. Some of the beliefs that they have heard of included the fact that people believe witchcraft is responsible for the disorders, whereas others believe that encountering an individual with certain conditions will benefit them in some way. All three of the individuals with albinism had exposure to such beliefs, as well as many of the other youngsters.

“As you know, Africans think it’s witchcraft” (P4)

“There are stories that are made regarded to persons with albinism. And some of them are stories like we are not people. And then there are stories like we don’t die, we just disappear.” (P5)

“Then like they would literally give you money out of the blue. They believe that if they made me happy, then they would have luck. And I don’t understand, I think like they don’t understand why I am white and stuff, because if they did they wouldn’t give me their money and hugs only

because they want to get luck from me. How can I make luck for them? I don't have luck myself.” (P9)

They had experienced a greater sense of understanding whilst being amongst other visually impaired individuals within the school setting.

“You see here in school it is fine because we understand each other” (P10)

“Like especially at school, at school I have people with like that have been here for many years that understand.” (P4)

4.4.4 “Without understanding no one will know”

There was a sense of desire for an enhanced understanding from the community and family. This was portrayed directly and indirectly. The longing for others to understand what they experience visually and what their condition is, was noted when participants brought props to the interview to explain the VI. Participant 1 had paper glasses which were designed to demonstrate what individuals can see depending on their type of RDD, which was given to the researcher to examine. Participant 3 brought pictures of individuals with congenital glaucoma and a diagram of how the condition originates. In addition, participant 2 had notes which explained the condition in more detail. Verbal statements were also made with this regard. This was expressed at least sixty-four times, excluding the previous amount mentioned in the sub-theme above.

“Umm just for them to understand me and fully support me... it's just the basic thing is to have a good understanding.” (P14)

When asked if they would find it useful if somebody explained their condition to them, most of the youngsters felt that it would. There were two main reasons for this. Firstly, they wanted to have a greater understanding about themselves, to educate others about their condition. This continues to indicate the desire to be understood by the community.

“Ja that will be better, ja then maybe I can also teach them on my condition Can everyone also to know about it.” (P3)

Secondly, they wanted to have more knowledge to promote their health and quality of life and to reach a level of acceptance and develop a sense of identity.

“For me it’s okay, like to tell me the truth what is happening on my eyes. Because if maybe I didn’t know, then maybe I will be questioning myself. Not even accepting myself the way I am.” (P11)

“I need to know more about my condition and maybe there can be like a cure. And maybe that person can help you” (P12)

Those that did not want the condition explained to them, felt that it would not change their circumstances and that information would not eliminate or minimize the condition and their feelings. For Participant 10, knowing that he will have an ongoing eye condition provokes a feeling of disinterest in anything anybody tells him as he does not want to accept the condition.

“I feel like it won’t change anything miss. Even if it explains it. I won’t take it serious, it’s like I will stay like this miss, all that’s in my mind is I will stay, like even if you can like put how many words, you see miss. Like my parents always try to convince me, like keep on telling me ‘don’t worry God won’t give you this if you can’t manage’. I like, yes miss you can talk that but the main point is I won’t change miss and I think that makes me don’t care about what they say.” (P10)

Participant 14 explained that it would be useless for somebody to explain the condition as she doesn’t want to be in the situation she finds herself in and felt that people would give her information merely to try to make her feel better. A similar view was mentioned by participant 6 who did not want others to determine if he would have affected children, however, he was satisfied to gain more knowledge about himself.

“There is one thing that I just think whenever they mention it to me. I just think that everything is useless, yes. Because I feel that the person is wasting their time and whatever they say to make me feel good, doesn’t make me feel good, because I have my own opinion to what I feel about it. And it won’t make me feel any better, because my own opinion being blind will be my opinion and their opinion will be theirs, because they will just say everything that’s positive, trying to make me feel good. And my heart and my mind won’t tell me the same thing, so they will be wasting their time and I will be, just be okay whatever.” (P14)

4.5 Theme 2: Procreation: What about Children?

The main implication of having a genetic based VI according to this sample is procreation. In many regards the psychosocial aspects outweighed the fact that the condition is genetic, however, the hereditary aspect had a large contribution with regards to having children. This theme focuses on objective four of this study, by exploration of the thought process utilised to decide about having children in the future, as well as if and how they would manage to raise

children whilst being visually disabled. Regardless of the opinions evolving around future children, all the young adults felt that having children would have an impact on their lives.

4.5.1 “It is possible that they have the visual condition”

The uncertainty about the health of their future children has created an internal struggle for these individuals, in which they need to determine if they want to take the risk to procreate and if their current situation would warrant having children. For some of them, this thought process was driven by their interpretations of the risks of having a child with the same condition. For participant 14 trying to follow an inheritance pattern within the family lead to the conclusion that possibly the disorder skips generations.

“Maybe if I have kids they will have the same problem, not really, but one of them maybe. It depends because my mother... my granny have an eye problem, but my mother don't have it. I have it. So it might be skip one. Maybe my kids won't have it but their kids will have it. It will maybe be like that.” (P14)

On the other hand, participant 3 was not concerned about having children as his understanding was that they would not be affected, regardless of being informed about genetic conditions at a prior stage.

“My condition, it doesn't actually affect your sperm cells and stuff, so it has nothing to do with that. So everything is like fine to me. The problem it's all the things like the drainage of the eye, it occurs in your eye, so everything is in my eye that was the problem so does not actually affect other organs in the body. So that's why I didn't actually worry when they were here because I knew my kids will be normal.” (P3)

Participant 1 had more information about genetic conditions and methods of inheritance, therefore his thought process was more technical.

“I was also told that... by my doctor, that since it is X-linked and my mother is a carrier, if I have daughters they would have 80% of contracting that condition.” (P1)

4.5.2 “Not willing to take the chance”

The fact that they could pass the condition to their children was rather unsettling to a few of the young adults and they did not want to take the risk and expose their children to the same life situation that they are in. The decision not to have children was most significantly expressed by one of the participants who said that he would rather die alone than burden his children with the condition.

“I don’t want no kids miss. Because I am scared they will be like their dad, you see. That’s a goal I want in life miss, I want no kids. I believe that it is going to be unfair to me to give my kids this. Like, I want to die alone, I want to be alone miss. I don’t want to bring other people to this, see that is what I want in life.” (P10)

For one of the blind individuals (P4), the idea of having visually impaired children was not problematic. However, a fear provoking thought was the possibility of them being blind. She felt that this would be a huge barrier, as they would then be unable to assist her. and that she needed sighted children to be her eyes. Another blind individual (P14) also did not want children, however her reasoning was that she would be unable to assist them efficiently.

“I am afraid of getting a blind child, that’s the problem. If the child is visually impaired that’s fine, but not blind. Like partially sighted. The problem is being totally blind like me, what if the child is also totally blind and the second one also blind. That will cause much more problems because then your children won’t be able to help you even, because they will also maybe be blind.” (P4)

For participant 1 having his own children was not an option, however, due to him having more knowledge about various options, he was contemplating having children via artificial insemination. He had not mentioned if he would want to use a donor sample or PGD. The idea of needing to find other methods of having children was also expressed by participant 13 who said he would want to adopt a child instead of having a biological child.

“Artificial insemination is one option. I know it’s not a fun way of going about it, however I am not going to repeat my grandmother and mothers mistake. Because there is technology now and it can be prevented.” (P1)

4.5.3 “My child can live with it”

Over half of the young adults would accept their children if they had the same condition. Although some of them were not particularly keen on having a visually impaired child, they would still have children and would accept them as they are and would make sure their child receives all the necessary assistance. Having a visual disability created a sense that they would not reject or avoid having such a child, because they themselves have the condition and this would mean that they are rejecting themselves.

“I will just accept it the way it is. The only thing I can do is to accept it. Because I can’t take it back and say, ‘hey go back, I don’t want you because you are visually impaired like me’ I won’t like throw him away, because I know the situation, I come from that situation.” (P11)

Interestingly the three individuals with albinism were most accepting of having a child with their condition. To them it was not a challenge and it would not have any negative impact on their lives.

“I wouldn’t have a problem with that because it is something that comes from my family, so I wouldn’t mind if I can have children with my condition.” (P5)

Not only would participant 9 accept a child with albinism, she would prefer it.

“It never had a setback on me. Like ‘I don’t want to have children, what if my children they look at me, like they are like me.’ No I never like had that. Like I do want to have children one day, and I wish to have two twins. I wish I can have twins, girl twins that are also with albinism.” (P9)

4.5.4 “How will I raise them?”

Having visually impaired children was one factor that impacted the individuals. In addition, another aspect which they thought might create challenges is their own visual disability, with regards to looking after a child. Although this topic was not the most important concern regarding offspring, it was noted that they thought perhaps their children might disobey rules because the parent could not see, or that harm would befall their child and they would have difficulty determining this.

“If the child is young they walk around and maybe I won’t see that the child has got something in his mouth or something. And like medicine, I can give the child wrong medicine and not see properly.” (P12)

For some of the individuals, no physical implications were noted, but instead an emotional connection. They were concerned that the children would be embarrassed to be seen with them or would be too young to understand their lack of sight.

“And some of the children will be embarrassed to take me to school because when it is meeting, the parents must be at school. Some of them won’t want to take me to school.” (P2)

For this reason, they would need assistance from a sighted partner, as someone who could see would be able to do things for the children which the visually impaired counterpart would struggle with. This has an implication on the type of partner these individuals can have, as their selectivity regarding a companion’s health creates limitations to relationships.

“I plan to have children when I am married. And I am sure the husband that I will marry one day will don't have visual impairment. Because like we can't do that with the children. Because I am sure it is going to be worser. So, it will be a challenge for me but I plan to have someone who will help me to help them.” (P9)

Participant 15 who already has one child admitted that the toddler was being raised by his parents, as he did not have the capacity to provide the sufficient care that is required. He felt that his parents were doing a good job of raising the child and that he would hinder the child's progress.

4.6 Theme 3: Social Interaction

Social interactions seem to be one of the hurdles faced by each of the young adults. Humans are social beings and have an inherent longing to interact with others. Although it remains the same in this instance, having a VI has influenced the way in which these individuals merge into society and the dynamics of engagement does not correlate with the accepted view of social interaction. In this theme, the experiences of the young adults are explored with regards to their social and intimate relationships.

4.6.1 “I am always alone”

Being isolated from society, friends and sometimes even family is often the case amongst these youngsters.

“I don't really interact; I am not always around people. Coz I have that kind of a shield, like, I like being alone because I don't know how to express myself in front of people because I am so different.” (P2)

They find it increasingly difficult to engage with friends whilst not being able to see the object which is being discussed or not being able to partake in an activity because it requires a high visual output. Having to either pretend to see properly or to be excluded from the conversation is an isolating feeling. This results in avoiding friends and rather being alone. These feelings are amplified when they find themselves near individuals who are fully sighted, because they tend not to take the visually impaired individual's condition into consideration during interactions.

“They see things that I can’t see and they laugh at things that I maybe can’t see what they laughing at so I feel out sometimes, and I just go sit at home. I don’t like to be around normal people. I’m always in the house because sometimes I can’t stay with my friends because that things make me angry when they see things that I can’t see well.” (P10)

One of the participants was thinking of his friends, rather than his own wellbeing when deciding to isolate himself. He feels that his condition is putting people who associate with him, in an awkward position and to avoid such situations he detaches himself from social engagement.

“If I go with people or if I am with people and then there are people that are passing I am causing that tension to them. Like ‘look at him’. I think that most of the time I am ashamed to be with people because I am gonna, like maybe I am gonna cause them a tension. So, most of the time I prefer to be alone.” (P5)

They feel embarrassed about their condition and do not want others to notice the physical aspects thereof. One participant refuses to wear his glasses around sighted individuals, which decreases his vision and creates friction in his social interactions. This individual spent a substantial amount of the interview sitting with his eyes closed. Another participant has started wearing contact lenses to avoid wearing glasses, in this way he is improving his vision whilst hiding the impairment from others. One of the blind individuals had noted that she has learnt to act as sighted people do and therefore most people do not easily notice that she is unable to see.

“I have to wear glasses every day, and that’s the thing I can’t do. I can’t, I don’t want, no I don’t want to wear glasses, miss.

Interviewer: And why do you say you can’t wear glasses?

“Miss, I can’t, I feel embarrassed, I feel bad, I don’t feel right, you see. Because I am used to go out to normal people and many of my friends don’t know I have this disability. I keep on hiding it away. If I wear my glasses they will see, you see miss. I don’t want them to see that I am like this.” (P10)

Other than the fact that social relationships become unstable and the youngsters find themselves with fewer friends or engaging in social activities much less than their sighted peers, being isolated has a negative impact on their mental health. It evokes feelings of frustration, anger, sadness, depression, and even suicidal thoughts.

However, for participant 9 and 12 friendships are easy to attain and maintain, thus they did not experience the isolation as explained above.

“For me it’s easy to get along with people. I get along with people normally and making friends is easy.” (P12)

In the instances in which they had good friendships, it was due to the mutual understanding they had. However, satisfactory social interactions were mentioned minimally.

“I don’t have a problem with my friends, because they understand umm my condition and we get along.” (P9)

Society is often judgemental or unempathetic and alienate visually disabled individuals by their means of interaction or treatment of these individuals.

“We always get people that will like try to distance themselves from you and will always try to take advantage from you.” (P11)

Occasionally friendships are terminated because people do not feel comfortable having a visually impaired friend, do not know how to interact with them, or do not want to accommodate the needs of a visually impaired individual.

“I lost most of the people that I was spending time with, I have lost them because of my condition. we had to social network, so there had to be a change, because now I am using something like talk back. So that simply means that they have to write in full, none of those writing in nice short cuts... So that also put some people off from me.” (P15)

Furthermore, they have encountered that the community view them as mentally challenged. These inaccurate perceptions from society are frustrating and can often lead to the individuals feeling alienated from the environment they are in.

“When they see a blind person, they see a stupid person. They see this person that sometimes deaf also or can’t speak because most of the time, maybe when I am walking with my friend or my mother they will prefer asking questions to them more than me.” (P4)

4.6.2 “They make fun of me... They feel sorry for me”

Various reactions are produced by society towards visually impaired youngsters, which these individuals do not find appealing. Being bullied is a common occurrence amongst these individuals and has a negative impact on their psychological wellbeing. Participant 12 recalls being bullied while he was at a school with learners that do not have disabilities and his

achievements were hindered by this. Participant 3 explained the amount of sadness bullying could evoke.

“It was like being an outsider and kids making fun of me and that. Like they pulled me down. They took me away from performing as well as I wanted to.” (P12)

“If your self-esteem is a bit low, I would say you will cry a lot.” (P3)

The emotional impact can be more detrimental in some instances, which leads to a need for professional psychological management. This indicates the possible extent of the implication of being continuously persecuted for having a disability.

“I had depression before, it was basically because of my eyes and like I have been bullied at the communities. So it made me feel very one sided and it led to depression. Which I had to go to hospital for three months basically, for treatment to help me like get counselling and stuff.” (P2)

On the opposite side of the conundrum is the pity received from people. The young adults struggle to feel empowered and to integrate swiftly into society when others constantly feel sorry for them and make them aware of their visual circumstance as if it hinders them from living a fulfilling life.

“All the people look at me like ‘shame look at him’ and so on. I don’t want them to look at me like that. I want them to look at me like a normal person because I am a normal person.” (P12)

4.6.3 “It feels like I am blaming them”

The genetic component of the visual conditions has a direct impact on family relationships. Although many of the individuals did not mention any negative feelings aimed at their family for endowing them with the condition, in some cases a form of hostility and blame was apparent. Participant 12 was holding his mother accountable for his condition as the condition was believed to have been transmitted from her.

“Most of the time I thought that it was my mother’s fault.” (P12)

While growing up participant 10 thought that his parents purposely created him with the condition, which he could not understand or accept. His mother refuses for him to undergo surgery to improve his vision. This has enhanced his perception that she is trying to punish him and has amplified his resentment towards her.

“I really thought that my parents made me like this, I thought that they hated me... The doctors say they can operate me and then my mother keep on saying no, and that makes me and my mother not to like each other miss.” (P10)

4.6.4 “Going to struggle to get the right one for me”

A variety of feelings surfaced regarding intimate relationships and finding partners. Although the genetic aspect of the condition did not seem to impact most of the opinions regarding the way they thought a perspective partner might view them, two of the participants believe that a man would not want to marry them, in fear that their child will also have the condition. This was explained by participant 2. Whilst others, such as participant 12 believe that a disability in itself will create barriers to finding a partner.

“Some guys may be afraid to be with me because they are afraid that their child will end up the same and stuff.” (P2)

“I think it is going to be hard because most girls don’t want to take guys that has a disability or something like that.” (P12)

In contrast, one individual thought that finding a partner would be easy due to the condition being familial, instead of a medical cause or having been injured in some way. Needing to attend hospital for health reasons or having complications associated with visual impairment was more significant than genetics, therefore she felt that her condition would not negatively impact a relationship.

“I think it will be quite easy for me to find a partner and be in a relationship, because I don’t have any causes of being blind. It’s just a family thing.” (P14)

Having a companion who was also visually impaired or has a genetic condition was not appealing, because as mentioned previously, they were concerned that they would have children who have a visual condition.

“I don’t want a person who also has a genetic disease because that will increase the chances of me also getting offsprings that will also have this genetic disease or condition.” (P4)

In some instances, the reluctance to have a visually impaired partner was due to an emotional or physical reason. Being able to perform tasks within the household, especially with regards to the management of children, requires the assistance of a fully sighted individual. Furthermore, needing psychological support meant that a companion should not undergo the same emotional challenges.

“I don’t want to date a disability child. I don’t want, I need a shoulder to cry on. I can’t date a girl with same problem as me, where will I cry? I need a shoulder; I need a normal person.” (P10)

4.7 Theme 4: Mobility

Difficulties relating to mobility were revealed by all the participants. Theme four examines mobility from a visually impaired young adult’s frame of reference. Frustration was expressed with regards to the numerous obstacles faced while trying to navigate from location to location. Besides these challenges, attempting to be mobile independently creates many safety concerns, which hinder these individuals from pursuing activities they would engage in, had they not been visually impaired. Being unable to do a standard task such as driving is saddening and is discouraging to some of the individuals.

4.7.1 “I can’t Drive”

Driving independently is a task which is exceedingly desired amongst this sample, however, many of the partially sighted participants noted that they are unable to own a car or drive. The inability to drive renders them dependent on others. For some individuals such as participant 7 this can be a hindrance as people are not always accommodating or willing to assist with transportation.

“I can’t drive a car. Umm I have to ask people to drive me certain places and some people will say no, when it’s urgent or something like that.” (P7, Appendix G quotation 1)

Participants 5, 9 and 10 had a dream of owning a car and being able to drive. The inability to drive and having that dream torn away from them, has caused them to feel hurt and unhappy and has impacted their emotional wellbeing. Participant 9 explains the process of needing to undergo a special optometric test which indicates if one’s vision is sufficient to obtain a driver’s licence, and how she is hurt by this situation.

“It’s painful because as I mentioned that you can’t see and there will be actually obstacles that are in our way because of the visual impairment. For instance, driving. Like we have to pass a certain test in order, so what if I did not pass the test, then my dreams of having my own car and driving it will be smashed. So that’s why I say it’s painful in a way.” (P9)

Whereas for participant 1, the desire to drive evoked a determination to find methods to drive. He mentions visual aids which can assist with driving

“I obtained a referral letter to see the optometrist in Kuilsrivier, for driver’s license optic glasses. So that will help me obtaining a 90% vision. Ja so that’s one umm medium term goal.” (P1)

4.7.2 “The bus...the train...I can’t see it”

With the inability to drive, these individuals have no choice but to rely on public transportation. However, this is a challenge as most of these youngsters cannot engage with the public transportation system as the average sighted person does. The largest obstacle for them is the fact that they are unable to see the proposed destinations of the busses and trains. The trains and busses need to be extremely nearby before they can distinguish whether it is the correct one which they need to take.

“When I go to the train station. I can’t see the train when it comes. I only see it when it’s you know (puts hand in front of face). Then it’s already gone. I can’t even see it,” (P2)

Participant 2 continued to explain the same occurrence with the busses and the inability to read the destinations labelled on the anterior of the bus. As public transportation proceeds according to a schedule, regardless if everybody has occupied their seat, this often results in the individuals missing their bus or train.

“The bus, golden arrow, when they come you have to look there right at the front is the name, so you have to check what kind of bus this is, where it goes to. I can’t see that, so the only time I see is when it is nearby, then it’s gone, then I have to wait again for another bus.” (P2)

It was thought that a method to solve this hurdle would be to have auditory systems which would transmit the transport information in substitution of reading.

“Maybe the government can make, like a technology bus or something. Maybe like a bus that when it’s more like 10 centimetres way or something, the name that’s on the bus it will say it loud... ok this is Mitchells Plain this is Rondebosch or whatever. Ja ok you will hear the bus and then you will stop the bus.” (P3)

Boarding the correct bus or train is not the only challenge. If this step is successfully accomplished, the next task is identifying when to disembark. A misjudgement occasionally occurs and they find themselves at the incorrect location.

“I took a wrong bus. Luckily, I had my ticket with me. So I told him I couldn’t see, Now it is this area I am not familiar with, now the bus went to Blue Downs, okay now they stop there, they stop at the mall there. Now ok, now I told the driver that I am not supposed to be here, I am supposed to get off in Mitchell’s Plain already. But he said the bus passed there already. Now I told him ‘sorry sir but I couldn’t see, maybe you drove too fast or what, I didn’t see what actually happened’.” (P3)

An aspect that discourages them from using public transport is the attitude from people they encounter along their journey. People’s reactions towards their visual disability makes them apprehensive of asking for assistance.

“Sometimes I try taking a taxi and it didn’t work very well, because the change that they gave me, sometimes people give you change and then you must count and then I was doing that and some people were making jokes and you know.” (P6)

“If you are going somewhere or if you have to take a bus or taxi, then it is not easy, because you can’t see the golden arrow bus when they are writing there. And most people, if you ask them they are going to ask ‘can’t you see the writings there’. If you are telling them like ‘no I can’t see, my eyes are a problem’. ‘Oh wow you are so young to have a problem with your eyes.’ and all that stuff. Then it’s like tough for those people that are shy to ask. They won’t even try to travel to places because they are scared to ask people.” (P11)

Safety was another concern, as it was felt that using public transportation is not very safe.

“Public transport, it’s a bit dangerous because those people don’t accommodate for people with blindness and disabled people.” (P14)

4.7.3 “Moving around is difficult”

Transportation is not where the challenge with mobility ends. Walking around, be it to get from one destination to the next, to go to the shop or to visit friends is described as a dangerous, scary and unpleasant challenge. Concerns were voiced with regards to crossing streets, as vehicles cannot be seen clearly. Participant 7 explained his frightening experiences of crossing the street.

“Going over the road, I should be injured right now because of the amount of times I got knocked over. I have been hit by bakkies and other vehicles a bunch of times. I could have been badly injured.” (P7, Appendix G quotation 2)

The fear of venturing outside to unknown places creates a barrier, as the youngsters tend to remain indoors or in areas which they are familiar with. Various aspects provoke their concerns, such as being in an unfamiliar environment in which conflict occurs and they are unable to navigate away from the situation.

“I stay at home sometimes, because I am afraid of going out and while I am still in the streets, like sometimes I can walk alone there... like move around there on my own, so I am just afraid of gangsters, what if the gangsters fight and I am still there roaming around.” (P4)

The amount or assistance from the community, or the lack thereof and the fear of what society would think if they were observed trying to navigate from place to place, also influences the confidence and keenness to become active outside.

“Like at the community umm I am always indoors because I am afraid what people will think of my eye problem because after when I walk down the street the people start talking about what is wrong with that girl’s eyes, can’t she see.” (P2)

When assistance is provided from the community, the task of navigation becomes less strenuous or unappealing as they feel safer and less scrutinized. This was the instance for participant 10 and although he must return home before sunset, as he has very limited night vision, he was less wary of leaving home during the day.

“When I am walking to the shop alone someone will come in my community and say come let me guide you home you see. Like that one you see I am struggling with something, especially when I am going to pass the road. There will be always someone from the community who helps me to go past the road.” (P 10)

4.8 Theme 5: Abilities

It is no secret that VI affects routine activities which can be performed and the achievements made by the individuals. However, the extent of this was variable amongst the individuals and various life experiences dictated their specific belief regarding their abilities and the impact of their disability on their future.

4.8.1 “It did affect me academically”

To gain an education, a level of visual capacity is required within most educational contexts. The participants that were attending Athlone school for the blind were very content with their educational circumstances. Being in a school in which educational settings and resources are tailored to fit the visual needs of the individuals’ made a huge difference in their learning experiences and their educational achievement.

“Here at school they understand us, the teachers are helping us, giving us text book with big names, so there is no problem at school, they give us like everything like we need.” (P 10)

This was not the case with the participants that attended a mainstream school previously. Their education was negatively affected by the harsh circumstances of being surrounded by learners and educators that performed at an average pace. As per the shift to the school for the blind, it is evident that they were unable to keep up with the sighted learners and maintain satisfactory progress.

“I started in the normal school and when I go on with my studies then I struggle to, to continue there.” (P5)

Attempting to read information from a black/white board was one of the main hurdles, due to the far distance, the wording was not clear and the individuals could not make appropriate notes in the given time frame.

“We were reading and I cannot see. I would end up writing the wrong things because I couldn’t see properly. She (the teacher) was busy writing and she divided the board into three. And she would write this side, write on that side and write on the third side (explains with hands). When she is done on the third side she would erase the part on the first side. When she would erase it I am like still struggling like ‘what is the first sentence’. She would erase everything and start over and then I remember this day I burst into tears because I couldn’t see and she kept on writing and even my peers were carrying on writing, and I was stuck.” (P9)

Tertiary education was an appealing thought to most of the youngsters and most of them had the ambition to attend university at some stage, in addition to one individual who was already completing a law degree. Regardless of wanting to obtain further education, the thought of attending university, a system which they are not accustomed to, was frightening. They anticipated many new obstacles, such as orientating themselves in the new environment and thought that they would not receive the same support and resources, which would create a setback in their journey to obtain new knowledge.

“In future, maybe I am going to be out of Athlone school for the blind and go for university. It is going to be lots of people there and then the lecturers won’t be able to go and enlarge one paper for you. So, it is going to be tough because everyone he have to do everything by yourself.” (P11)

4.8.2 “The advantage of a disability”

Employment consumes a large portion of adulthood, and for those wanting to be independent, having a job would be necessary. This however, was not one of the concerns to most of the young adults. They were confident that they would acquire a job without effort, as being disabled placed them at an advantage above others. Via various sources they had ascertained

that a certain percentage of the workforce should be disabled, which meant that they would be hired.

“They said that we come first in the business, I think in the business about 15% there must be disabled people, so I don't actually worry because I know there are acts that are protecting us or wants us to be in the workplace” (P3)

The fact that they thought they could easily obtain a job was reassuring in many instances. However, one of the girls who also believed that individuals are employed due to having a disability thought that it was not an advantage to be employed purely based on such a quota. She seemed insulted by the idea that she would only be good enough to work by default.

“I don't like that when they say like ‘okay we will hire you because like we need this number of people with disabilities.’ I don't take my albinism as a disability so don't hire me only because you want to add to that number, but hire me because I have that qualifications and I am passionate about this job. Don't do me a favour, do it because you need me.” (P9)

The specific occupation that can be obtained is however a limiting factor. Many professions require a sufficient vision ability, such as becoming a doctor or pilot, or a job that requires a rigorous amount of reading or strenuous eye activities. This means that they are unable to be as selective about their career path as their sighted peers. Although there are many possible occupations and they did name a field which they were interested in pursuing, they would possibly in some instances have been interested in a different field of work.

“I can become maybe a journalism, I can become a teacher. So to become a doctor it's hard because, eish to be a blind doctor, you gonna like put something in the wrong place and all that stuff. And to become a lawyer and all that stuff, I can become. So it's easy for me to get a job, but some jobs you will always have disadvantage about that.” (P11)

Having a successful career did not seem challenging to those who knew what they wanted to do for a living, and felt that their vision would not drastically hinder their capacity to perform efficiently within that field.

“I want to do IT and it doesn't affect me. But sometimes it does because I need to look at small things and sometimes I can't see and then I need to go ask someone to help me. But I like doing IT and I can do it.” (P12)

4.8.3 “I can't do everything by myself”

Having a lack of visual capacity generally results in a lack of independence. In this instance, the level of vision was a contributing factor. The blind individuals are more dependent on others

and often do not experience the desired level of privacy. This was noted as a significant hurdle for these young adults, as they are moving into a life stage where they want to make their own choices and partake in activities that under normal circumstances should not be supervised.

“I must have a person who is with me most of the time. Sometimes I can’t go to places that I want to go that somebody won’t know. So, your life is not, you don’t have privacy.” (P4)

“I am always inside the house, I am not used to going, like now that I am blind, I never went alone somewhere.” (P14)

The partially sighted group felt relatively confident in their ability to be independent and were excited about the fact that young adulthood meant that they could finally make autonomous decisions.

“I would manage living alone because I am good at working alone and I know how to work with my money and how to like live alone.” (P12)

“I want to be independent. I don’t want to be dependent on anyone because my whole childhood I have been dependant to people to help me. Now I think it is my turn to help myself.” (P2)

However, being partially sighted creates a sense of dependence in certain circumstances. This is especially the case in new environments, where they are unsure of the surroundings and need assistance from others. Sometimes creating awkward situations, in which they are required to ask for assistance from strangers. Family and friends are occasionally enablers of dependence, as they take over the responsibility of the individual or try to perform routine tasks for them.

“I get fed-up to be around them because I don’t even get my chance of being independent. Because each and every thing that I do, they want to do it for me. Even when I stand up, even if it was possible that somebody could go toilet for you, and then you just have to sit there and they take over.” (P15)

4.8.4 “Most of us are handled like eggs”

Apart from the limitations that are routinely faced, a constant reminder continuously lurks with regards to what they are able to achieve. In general, the community underestimates them and continuously points out the tasks that are thought to be impossible for them to perform or they are undermined when they do attempt something.

“My mom will be saying that ‘oh my gosh, you can’t cook and stuff’.” (P2)

“If I explain to them my condition and what I can do, it’s like they don’t believe, they think that whatever I say it’s not true. Like when I say ‘I will be able to do this’ they say ‘no its impossible for you to do that because you are blind’. They think that there’s lots of things that I am not

capable of doing. But there is, because I mean I am just blind, there is nothing wrong with me actually. It's just that I don't have sight to see.” (P14)

The youngsters find themselves in situations in which their capabilities are disregarded even before they have had the opportunity to dictate whether they are able to engage with the situation or not.

“When you take somebody, who is fully sighted to go to the shop with, and you are the one who's going to buy, but most of the time whenever they ask questions they won't ask the question directly to you. Even if you are the one who will be taking the money in your pocket, they will be asking the person who is fully sighted.” (P15)

Although the individuals like their school, some felt that they were being nurtured too much, which would lead to difficulty once they leave the secondary educational system. They propose the solution to this would be to be assigned more personal responsibility so that they are able to acquire the necessary skills.

“If we like at normal institutions the teachers like don't really explain the work and put copies and stuff. I think the school should make me more prepared by allowing me to do my own stuff basically. To make my own copies, to use the laptops to write, that's how I will be more strong enough to face the outside world and working areas, if I will be too dependent to people outside then I won't be able to cope.” (P2)

4.8.5 “I can do what the other people can”

A very strong sense of desire to be successful and to prove to the community that they will succeed, emerged. They want to reach the same goals as sighted young adults without allowing anything to get in their way.

“We also want to be at that level where everyone is, you know.” (P6)

This desire to acquire the same achievements as their sighted counterparts stemmed from either wanting to fulfil their own dreams and internal goals.

“I do my best to get to where I want to be. I am not going to let anyone stand in my way of getting to where I want to be. So, ja, I am going to get to where I want to be, but with this disability.” (P12)

Or on the other hand, the main aim was to prove that they are not inferior and should not be looked down upon.

“If maybe I can study and study so that I can get into the level that everyone is getting, so that I can maybe show some people, that also people with albinism can reach that level.” (P5)

This urge for accomplishment was not dictated by their particular visual capacity, as the blind youngsters expressed similar wishes than those who have less severe visual loss. The strategy to reach a life victory included obtaining tertiary education, finding a job, owning a house and living independently.

“Next year I will study of course and I will get a job and I will buy myself a house and live on my own and on my own money.” (P2)

They tried to rationalise their disability by concluding that God created them with a visual condition in order for them to make a success.

“The reason is to succeed in life, even although things are a bit hectic, even although things are challenging. God wanted me to be somewhere, but in a different way, not the same route that people were using. So that I can be different and like more successful than other people are. Like being most successful,” (P15)

Failing to excel and triumph in the goals that they themselves, their families, friends and community set out, was not an option. Regardless of the challenges, they believe that success must be reached by one means or another.

“So you have to make sure, like you succeed in life.” (P9)

4.9 Chapter Summary

This chapter specified the socio-economic and cultural information of all the participants. The five themes that emerged from the data collected during the interviews were explored by means of sub themes and quotes from the young adults.

Chapter 5: Discussion

5.1 Introduction

This chapter is dedicated to discussing the themes and various findings which were presented in the previous chapter. Where appropriate, existing research has been integrated into the interpretations. Each of the themes interlink with one another and although some aspects are more prominent, the connections found between them demonstrate the importance of how each facet affects visually impaired young adults' lives.

5.2 Understanding

One of the main aspects that became evident throughout this study was the level of understanding experienced by the participants and the affect it had on some of their perceptions of their visual disability. Understanding of a genetic-based medical condition is difficult for many individuals. The main aspects that people seek more explanations on are the diagnosis, cause, the effect on vision, management and progression of the disease (Clarke, *et al.*, 2015). In a study conducted of adolescents with genetic disorders, it was determined that they had a desire to understand more about their disabilities, namely, the way it affects them, condition specific information and how it should be managed (Szybowska, *et al.*, 2007). These findings support the information provided by the young adults in the current study who have a longing for knowledge of their condition, as it is clear that many of them do not understand or have sufficient knowledge about their condition, which has created a considerable amount of confusion, uncertainty and anxiety for them over the years. This illustrates the importance of education on these disorders. According to Clarke, *et al.* (2015) individuals have hopes that being seen by a genetic eye specialist would lead to answers. The young adults who participated in this research also felt that being seen by a healthcare professional to explain their condition would assist them. For those who seek understanding from others, this seemed like a good solution to obtain the correct information to use when needing to explain the condition. This leads to the importance of GC.

GC may assist to integrate information into these young adult's self-concept, which might prompt them to think about the possible future impact and meaning of their genetic condition. Adolescents who had undergone genetic testing had previously shared the same confusion about their condition and found that GC was very useful and that it helped them to navigate and manage life better. In an interview one of the adolescents had said *"I definitely gained a better understanding of what I had and why I had it...if I had a bunch of unanswered questions about my disease then I would feel even more lost. So, I think through the genetics session I learned a lot about the disease, and I think that's a good start to coping and adjusting"* (Pichini, *et al.*, 2016:887). If the visually impaired youngsters in this study were to attend a GC session, they would possibly have similar remarks, as they were rather positive about a promising outcome of such a session. These individuals with hereditary visual conditions will undergo a transition to adult healthcare. To do so they likely need to have a better understanding of their condition to gain increased responsibility over the management of their condition, which GC would assist with.

As per the current research, a study by Clarke *et al.* (2015:353) demonstrates that individuals were also concerned about a decrease in their sight and future vision loss. One such person had said *"Oh, my God, I hope they find something quickly before I do something with my other eye."* Furthermore, they were also trying to find methods to prepare for the diminishment of their sight. It seems that individuals in the previous and current study are aware that a decrease of vision can have a significant impact on their life and that it would require many new adjustments, which they are unsure they could endure.

Identity development is an important developmental stage according to Erikson (1959). This stage occurs during adolescence, which Erikson (1959) defined as a period in which children slowly transition into adulthood. Identity is defined as an individual having a good concept of who he/she is. Having a sense of identity means that an individual is content in their own body, has a sense of life direction and feel that they are a contributing member of society. Role confusion however, can distort the view of oneself and the perceived perspectives of others and may lead to the extensive questioning of oneself (Sokol, 2009). Based on statements, sometimes disabled individuals find themselves in communities and families consisting of able-bodied individuals, which creates an amplification in the complexity of gaining an identity, as they possess some unique traits in comparison to everyone else (Weeber, 2004). This portrays the dilemma the visually disabled young adults in the current study are

experiencing. The lack of knowledge and understanding about their genetic condition creates obstacles to attaining who they are, why they are disabled and what it means for their future. This may hinder their process in obtaining an identity. When observing the experiences and perceptions of participant 9, one of the individuals with albinism, it became apparent that the perception and understanding of a condition correlates with identity development, as she had knowledge and comprehension about her condition and was very confident, content, proud and reassured about herself.

The individuals in this study felt misapprehended, much like a group of adolescents with genetic conditions who had voiced consternation about possibly being misunderstood or rejected by their peers (Pitchini *et al.*, 2016). Often sighted individuals do not comprehend the extent of the visual condition and what a person is able to see (Lourens, 2015). In a different study, many of the disabled participants did not bother to produce explanations, because they did not expect any form of understanding from others, as they thought that their disability was not understood (Brouwer *et al.*, 2008). There seems to be a consensus amongst the visually impaired individuals, regarding feeling misunderstood. A variety of instances and interactions with people, creates an atmosphere in which they believe people do not know what they are enduring nor what their condition entails. Many beliefs are formulated about the visually impaired. A few of these include the view that blind people are evil, that they are being punished, they possess special powers or produce good luck (Papadaki & Tzvetkova-Arsova, 2013). With the variety of cultural contexts in SA, it is not surprising that these youngsters find their condition being linked to various beliefs.

Deaf individuals have a deaf culture and although they function in the community, they gravitate towards other hearing-impaired individuals. In contrast, unique experiences are shared by visual impaired individuals which are associated with visual disability, however, they do not have a specific blind culture with which they are able to identify (Weisleder, 2012). The sense of belonging encountered within such a culture is the opposite of the lack of empathy and sense of alienation reported within the general society in which disabled individuals are raised in (Weeber, 2004). Previous studies indicate that individuals feel that they have a form of blind community because they were isolated from others, which created a sense of cohesion, and only when they ventured out of their 'blind' surrounding did they lose the sense of who they are (Lourens, 2015). In many instances when the young adults in the current study spoke of being understood by their peers, they were referring to their friends which were also

attending the school for the blind. Although they feel more comfortable around these individuals, they still experience a sense of confusion, displacement and a need to integrate into the sighted world, unlike people within a community that form part of the deaf culture. The individuals who displayed the greatest sense of belonging were the three youngsters with albinism. They had a good sense of their condition and how it originated. They could relate to individuals within their immediate surrounding, as well as to external groups that formed to create an albinism community. Although these youngsters also faced many obstacles and had their own albinism related challenges, they seem to be in a position that was more reassuring.

5.3 Reproduction

The procreative implications of a hereditary visual condition were highlighted by the individuals in this present study. It was found by Kelly (2009) that parents of children with genetic conditions thought of reproduction as unpredictable and risky. In a different study, it was noted that most individuals that understood risks made decisions on those grounds, however, people that were not aware of or lacked understanding also indicated that they would avoid procreation due to the risk (De Pina-Neto & Petean, 1999). Seemingly, the South African young adults with their own visual condition also felt that having children was risky and they were unsure what the outcome would be. This could possibly be because they do not have correct knowledge about the inheritance risks of the condition, which means that they tend to formulate risk perceptions from limited data. It has been noted that risk perceptions constructed by patients are derived from their family's or own medical and psychosocial experiences (Klitzman, 2010). This statement seems somewhat appropriate within the current setting, as some of the individuals who were struggling most to understand, adapt or cope with their condition, formulated a reproduction risk which was high enough for them not to attempt having children, whereas those who felt more comfortable with their condition had created a less threatening risk perception for themselves. In this instance, uninformed decisions regarding reproduction may lead to a few of these young adults not bearing children, even though they are at low risk of having an affected child or visa-versa. This highlights the implications of not having sufficient information about inheritance risks.

In addition, experiential knowledge, which is a product of direct interaction with a condition, is a mediator in the decision-making process surrounding reproduction. Experience as means

of decision making can be useful in making choices, however, it might increase uncertainty about what they feel might be the correct conclusion (Boardman, 2014). Two views might form due to experiential knowledge.

Firstly, when one has endured life circumstances which have brought about many challenges, uncertainties, and psychological turmoil, it becomes increasingly difficult to envision or accept that somebody else should be subject to such an experience. A participant in Pitchini *et al.* (2016:588) study explained this view “*And just being the mother, like having that instinct that I don’t want my kid to go through the feelings I’ve gone through.*” The fact that some of the young adults in the SA study do not want to have children with a visual condition is not surprising, as they do not want their loved ones to endure the same hardship. It was found that blind parents did not want their children to feel responsible for them (Pagliuca *et al.*, 2009). Interestingly, this was not the case for participant 4 who wanted children to look after her. One study highlighting parent’s decision-making processes with regards to reproduction found that many participants did not want children, in the fear that they would have a child with a disorder. The method to insure this was surgical sterilization (Kelly, 2009). Although none of the individuals in the current study had mentioned this idea, some had also thought of ways in which to not have biological children, whilst still becoming a parent.

On the other hand, some individuals experienced their condition as something that can be overcome and that creates a more fulfilling life. Others felt that they could endure such a disease and therefore trying to eliminate it from future generations would be an indirect insult to themselves. They are willing to accept and have children who have the hereditary condition (Boardman, 2014). In this current study, those that did want children were inclined to feel that they manage with the condition and therefore their children will too and that they are in the same situation therefore they should not wish it away. Amongst these individuals that have a yearning to be understood, accepted and included in society, by rejecting the idea of having a visually impaired child, they might feel that they are fulfilling precisely the things that they accuse society of doing.

Genetic counsellors might find such information on experiential knowledge useful when counselling individuals with hereditary visual disorders, with regards to their experiences of their condition and how it influences their perception of future children. Also, it could indicate who might be more distressed, unsure and in need of extra support (Boardman, 2014).

Pagliuca *et al.* (2009) had received feedback from blind parents who experienced difficulty with activities such as bathing, feeding and giving medication to their children. In the current study, they did not mention any of the above, nor any other baby related tasks, possibly because they had not yet had children. They did, however, have concerns about older children and that they would have challenges providing adequate safety for their offspring and that they may be disobedient, misbehave or be psychologically affected. Visually impaired parents have reported on similar experiences with their children. They noted unsafe encounters which they had with their children, the fact that they hand out chores which are left undone, difficulty in transporting them, assisting them with everyday activities and their offspring's embarrassment towards them. A comment from one mom was remarkably similar to what participant 2 was anticipating, she had said *"The girls used to be mortified that I squint and look different from the other moms. I couldn't drive them to the mall like other moms"* (Rosenblum, Hong & Harris, 2009:85). Throughout the interview process it seemed somewhat as if the youngsters had not given extensive thought to how their own visual condition might influence raising children. There are a few reasons that are associated with this. Firstly, because most of them are not blind, they feel that they have enough visual capacity to conduct necessary child rearing activities. Alternately their strong belief in success and that they can achieve many things, creates a sense that they will be able to overcome any parenting related obstacles. Or because they aim to have sighted partners, they believe that he/she will be able to do the tasks which they themselves cannot do and therefore the tasks that they cannot manage are not actually important or problematic.

5.4 Social Interactions

5.4.1 Friends, Family and the Community

Having VI has had a large impact on the young adult's social interactions. Visually impaired individuals have limited social networks that consists of few friends, especially those that are sighted (Huurre, Komulainen & Aro, 2009). In a pilot study conducted on individuals with illnesses and impairments it was found that regardless of being born with a disability or acquiring it at some stage in life, the participants felt a sense of aloneness (Weeber, 2004). In a study by Pitchini, *et al.* (2016) numerous participates felt isolated due to their condition. They thought that no other people shared their disease and thus they felt resentful. Both the above-mentioned studies concur with the alienation revealed by this current group. Furthermore, it

highlights the blame and hostility that individuals such as participant 10 and 12 experienced. Adolescents and young adults are often in a stage in which they are self-conscious about their physical appearance. Those with visual disabilities, especially those which are easily physically noticeable, find it increasingly difficult to seek out social interactions, which limits their social skills and generates social isolation (Hodge & Eccles, 2013). A few of the young adults in this present study have undergone great lengths to suppress or conceal their impairment, to maintain friendships and be accepted into social circles and included in activities.

As per the findings in previous research, this group of young adults also noted the negative reactions from the community towards their impairment (Weeber, 2004). These reactions triggered the feeling of isolation in multiple studies. Adolescents in Pitchini, *et al.* (2016) study noted that they were bullied and that their sense of loneliness was intensified when receiving different treatment from family and social networks. To avoid revealing their disability and being tormented about it, they chose to withdraw from social activities. Multiple studies have mentioned the bullying visually impaired individuals endure, which includes name calling and being ignored (Hodge & Eccles, 2013). As with these individuals, the participants in this current study have been exposed to bullying, which as expected, has left them with emotional scars. To avoid this from recurring they attempt to escape vulnerability, exposure, and other unpleasant situations by avoiding social interactions. This however, amplifies the feeling of isolation. In contrast to being mistreated, many stereotypes emerge by various nondisabled individuals, in which they label the visually impaired as unintelligent, or they constantly display pity towards them (Lourens, 2015). The continuous sympathy and words of pity that sighted people provide is not always helpful. It tends to expand the gap between being 'normal', like the average member of society and being disabled. In the study by Lourens (2015) similar comments were made as in the SA study, regarding not wanting to be pitied as they felt that their situation did not warrant pity.

Adolescents with genetic conditions had a desire to be recognized as 'normal' by their peers in order to be included in everyday activities (Pitchini *et al.*, 2016). This is the exact portrayal of the young adults too. Being accepted and welcomed into a social circle is perhaps an important aspect of their sense of belonging. Acceptance within society is often easier to obtain when one is thought of as being the same as those who need to authorize the inclusion (Lourens, 2015).

It was found that 76% of visually impaired individuals had access to social media and used this for means of interacting and forming or maintaining friendships, which was useful to those who found it difficult to have physical encounters (Hodge & Eccles, 2013). These young adults in the current study have experienced some obstacles in this regard. Friends do not want to type out full words so that the voice program can read it out correctly or the appropriate voice technology was not available. The participants did however report some advances in their supportive devices, which might make their future social media interactions more effortless and pleasant.

The social circumstances of the young adults have resulted in a decrease in their psychological health. Many situations occur in which the visually disabled are excluded from social activities because people are uncomfortable to engage with them, or do not want to seek assistive methods to interact efficiently. Lack of sufficient support has been linked with loneliness. It is the quality rather than the quantity of the support which is valued and required most (Hodge & Eccles, 2013). Psychological wellbeing is related to good relationships with peers and family, as it creates a sense of acceptance. Therefore, isolation due to rejection can result in depression (Huurre, Komulainen & Aro, 2009)

5.4.2 Intimate Relationships

Individuals with disabilities have goals to establish intimate relationships, as with any person in society. The difference however is that these individuals encounter social situations informing them that they are not suitable romantic partners.

Many of the young adults in this study were seeking non-disabled companions who were able to assist with daily activities and care for them. A clinical psychologist found supporting information on disabled men who were also seeking such a partner (Gill, 1996). However, not wanting to have a visually impaired companion means that the young adults become selective of their partner, which creates a challenge to be in a relationship, as they tend to interact with visually impaired individuals on a more regular basis than they do sighted people. Gaigher, Lund & Makuya (2002) noted that prejudice regarding visually impaired individual's disability might reduce their chances of finding a sighted partner. At times, non-disabled individuals avoid intimate relationships with visually impaired individuals, as they get frowned upon by others or are labelled as being defective for not having a 'normal' partner (Gill, 1996). If this

is the case the youngsters are in a challenging predicament as they will avoid starting relationships with individuals with visual conditions, but when they find a sighted individual whom they are interested in, they might be rejected.

In the study by Gill (1996) it was stated that non-disabled men might reassess a relationship and marriage with disabled woman on grounds of the risks for future children. This concept is mentioned by females in this SA study. However, more interestingly such views were more readily expressed by the male and female visually impaired individuals themselves.

In this study, sighted individuals were thought to be better equipped to provide more support, and fulfil relationship needs. However, the opposite was found in a study by Gill (1996) in which woman that have experienced unsuccessful relationships with non-disabled partners tend to be more positive about disabled partners, as the mutual understanding seemingly deepened and strengthened the intimacy. Not enough information was given by the SA participants regarding previous relationships to know if this could perhaps be because a relationship with a sighted individual had not previously occurred. Disabled individuals who choose to have a partner with a similar impairment often face judgement and are often told that the relationship will not be successful or that they are merely in the relationship by default (Gill, 1996). Although this aspect was not highlighted in the current study, it is possible that a partial reason for wanting sighted partners is connected to their desire to fit in with society and not to be thought of as incapable and thus if they have a sighted partner, nobody can say that they are in a relationship because they could not do better.

5.5 Mobility

Having the incapacity to drive independently has a global effect on the quality of life of individuals, including the young adults who participated in this research. Individuals who are unable to drive were found to be frustrated and discouraged and felt that their freedom was curtailed (Golledge, Costanzo & Marston, 1997). The disappointment the youngsters portrayed when speaking about not having a car or driving is indicative of their hopes to be able to reach that milestone and the heartbreak and despair of having such a dream shattered.

Safety within the South African setting differs from that of the United States and European countries. In a study by Golledge, Costanzo and Marston (1997) the participants found it safe to use public transportation in California. However, due to high crime rates the general population of SA tend to be concerned and fearful of their security and safety on public transport. They feel that their safety is threatened whilst walking from one location to the next, waiting at the stations and during transit on the bus, train or taxi (Kruger & Landman, 2007). This feeling would be amplified by those who are limited visually, thus, it is not surprising that some of the youngsters are apprehensive to attempt being mobile and instead stay at home.

As with the young adults in the current study, such as participant 7, a visually impaired individual from a different study also explained his/her frightening encounters while navigating across the road, *“very often when I cross the road, a bicycle passes by very closely or I hear the brakes very near”* (Brouwer, *et al.*, 2008:471). Furthermore, a group investigated by Golledge, Costanzo and Martson (1997) also experienced challenges and a lack of safety when attempting to cross the street. Which prompted them to request auditory traffic lights. This indicates the extent of insecurity they encounter each time they need to get from one public location to the next and how unsafe and fearful many visually impaired pedestrians feel.

Efficient mobility is often acquired via training and mobility courses. However, even after learning orientation and mobility skills, it can remain a physical and mental challenge (Soong, Lovie-Kitchin & Brown, 2001). The sample from this study did not mention that they are receiving orientation and mobility training. However, from dealing with the school the researcher is aware that such training is available. Perhaps these individuals have not found this training effective enough to assist them with the challenges they face. Or perhaps SA does not provide the correct infrastructure or circumstances to implement these skills. For example, since 1999 references have been made in which the Cape Metropolitan Council approved policy proposals that aim to deliver appropriate transportation assistance for individuals with disabilities (Snyman, 2002). Now in 2017, many years after these proposals were approved, the transportation system has not changed drastically enough for individuals to report that they struggle less with mobility.

In many countries, a variety of systems and technologies have been implemented to assist disabled individuals. One such example is Vienna, Austria which has a public transportation system which is reliable, safe and cheap. The train stations are equipped with elevators, to eliminate the difficulty of using stairs. Furthermore, the tramway, subway and bus stations have a tactile guidance system, along with an audio information system and visual guidance system. Indoor navigation systems are available, which use tactile systems to calculate routes for the individuals to follow. Other applications also exist which provide the visually disabled and blind with specific information regarding their mode of transportation and route. This contributes to Vienna continuously being awarded the top city for quality of life in general (Emberger *et al.*, 2013). These forms of assistance are not available for visually disabled individuals in SA. Therefore, the young adults need to determine where they are and how to reach their destination without any external aid.

Various interactions with the public, which the youngsters encounter during their journeys, has the potential to be rather unpleasant. Mehra *et al.* (2015) explained that during a study on mobility it was noted via questionnaires and observations, that, without assistance, major difficulties were faced by visually impaired individuals to access busses and such individuals found that seeking assistance occasionally lead to them being misguided and influenced their social interactions. This supports the experiences noted by the young adults in the current study. Having to explain themselves and their situation, hearing comments made about their vision or being stared or indirectly laughed at, leads to these youngsters rather struggling by themselves or avoiding certain forms of transportation. During the interviews, they had suggested audio announcements, to be able to identify the busses. In the study by Mehra *et al.* (2015) a bus identification system was implemented, in which the visually disabled individual had a device which they could use to gain information regarding the busses. The information verbalises the destination or bus number, the distance between the individual and the bus when the enquiry is first made and the distance again when the bus is stationary. Furthermore, it informs the bus driver that a disabled individual is trying to embark. This was found to have a significant impact on the successful mobility of the visually disabled and of 100 attempts to embark a bus, 94 were completed successfully. Which verifies that the young adults in the SA study make very valid suggestions regarding the transportation systems they use and that such systems would greatly minimise the obstacles faced amongst these individuals.

5.6 Abilities

5.6.1 Education

VI has created educational setbacks and obstacles for these participants. An investigation on children with albinism in the Limpopo region had found that many prefer to attend mainstream schools (Gaigher, Lund & Makuya, 2002). This is contradicted in the current study; in which the youngsters are more content to be in a special needs school and they feel that venturing into a mainstream educational system would be a challenge. In environments where assistive devices were not provided efficiently, visually impaired individuals are unable to read normal text on notes or in books, nor see information written on boards or displayed on projectors. Which hampered their learning and ability to perform well (Lourens, 2015). By attending mainstream schools, this is what the young adults in the SA study experienced, which negatively impacted their learning and lead to their preference of attending a special needs school where their educational needs are met.

When making the transition from school to university it is seen that individuals who have attended schools for the blind experience more challenges with integration, than those who attended a mainstream school. This is due to the ample amount of support and assistance provided by special needs schools. Therefore, when visually impaired youngsters embark on tertiary education they are ill prepared to operate within the system (Lourens, 2015). The young adults in the current study do seem to feel that they are not prepared to attend university, even though they desire to, and the fact that they know they will not be in a contained environment makes them more anxious. The awareness that they are not equipped to venture into a new educational setting is conceivably why they want to be given more responsibility and opportunities to gain more control prior to university.

Universities do offer disability support; however, this can only be provided when individuals disclose their disability and request assistance. Lack of disclosure often relates to how they wish to be perceived by others and how they view themselves. Some individuals who are visually restricted have attempted to minimize their disability by not sitting in the front of the class, not asking for help and hiding their assistive devices, to be viewed and treated in the same way as the other students (Lourens, 2015). This is an important aspect within this current

group, as they have already expressed that they want to be seen as ‘normal’ and to be treated the same as others. This could have a negative implication for their future education and could create unnecessary extra burdens.

5.6.2 The Truth about Employment

In recent years’ individuals with disabilities have been accommodated more readily within the workforce due to the various acts that have been implemented, in an attempt for disabled citizens to be treated equally in society. The visually impaired individuals in this study believe that they will be the person to be employed, without taking into consideration that other disabled individuals may be competing for the same position. However, employment and being offered a vacancy is much more complex than it appears. According to Trading Economics (2017) the unemployment rate amongst youth in SA was 54% in 2016. This indicates that there is already a significant barrier to job opportunities and the probability of acquiring a position, without even taking disabilities into consideration. Elsewhere in the world, it was documented that only approximately one third of visually impaired individuals had some form of employment.

It was found that young adults in general tend to be over optimistic about employment, regardless of the less desirable labour force prospects. This might be linked to their incapacity to assess their worth to companies (Roberts, 2015). The visually impaired youngsters in this study spend time with sighted peers who possess these views and thus they may try to compare themselves with these individuals and consequently are influenced by this reasoning. This, along with the false hope that the equity act might evoke, conjures the absolute certainty that they will be employed without effort.

The young adults in this study wanted to become doctors, pilots or soldiers, yet they realised that this was not possible and they needed to aim at an occupation that they had the physical capacity for. Similarly, a group of adolescents with albinism were found to have high ambitions of becoming healthcare professionals, soldiers or even soccer players (Gaigher, Lund & Makuya, 2002). Understandably, this would be an unpleasant realisation for the participants, however, it seems that their confidence of finding a job suppresses any negative feelings they

might have. Although these youngsters are aware that they may have limitations with the type of career they pursue, Roberts (2015) describes that caution should be taken, as elevated levels of optimism regarding employment could create a pursuit for jobs in which success is unlikely.

Some disabled youngsters do realize that they might have a disadvantage when competing for vacancies and felt that their able-bodied peers are more likely to be employed as it would be less effort and cause less uncertainty about the person's ability to perform satisfactorily (Hendey & Pascall, 2001). This current study did not produce any views such as this. Even though some individuals felt limited, none felt that their sighted peers would obtain jobs instead of themselves. This is perhaps because they feel that they have an advantage above non-disabled individuals and that their sighted peers would in fact have more challenges to establish a career.

5.6.3 What they Can and Cannot do

It has been said that the attitude towards visually impaired individuals are observed in three manners. Namely:

- Non-acceptance, which is related to social interaction.
- Having the belief that visually impaired individuals are helpless and are unable to performing day-to-day tasks.
- Thinking that those with sight limitations must be assisted by sighted individuals, without determining which visually impaired individuals do not require help or areas in which aid is not necessary.

Living a regular life becomes increasingly difficult, due to the above attitudes (Papadaki & Tzvetkova-Arsova, 2013). The youngsters in this study desire a level of independence, as would any sighted individual. They wish to have the control over their lives in which they dictate how they live and what they do. Studies indicating the desire for being independent contributing individuals of society, supports the findings of this current study. The types of comments found in previous research included having dreams to drive, own a home, be like others and feeling satisfied when being able to do things without needing assistance or burdening others. These views are illustrated by an individual that said, *"I wanted to be earning a living, to be independent... not relying on other people to provide financial support for you,*

but standing on your own two feet.” (Hendey & Pascall, 2001:39). This is very similar to the comments made from this SA sample.

In some instances, visually impaired individuals are easily deemed incompetent to perform certain tasks or partake in specific activities, without allowing them the opportunity to explore their capacities and options. An example of this is when they, unlike their sighted peers, fail at a job or subject they are deemed as unemployable or unable to acquire education (Groce, 2004). This could be one of the reasons why the youngsters in the current study feel that they need to succeed, as they may be aware that they will be stripped of any further opportunities if they fail at a task. They express that people assume that they are incapable and somebody always tries to take over and does not allow them the opportunity to do things. They find this frustrating, especially when they can perform the task.

The young adults in this study had a strong desire to triumph in everything in life. Salminen & Karhula (2014). indicates that a few youngsters in their research expressed that they could succeed independently, whereas their parents reported that the individuals were not managing, regardless of them always giving the impression that they were. Reports of various findings on individuals with chronic illness indicated that individuals tried to restrict the effect their disability had on their social relationships by reducing the distinguishability of their impairment. However, due to the absence of visual indicators, individuals did not validate their true restrictions. When integrating into a community such as the deaf community individuals had increased faith in their true capabilities and became less secretive about their confinements (Weeber, 2004). This SA study demonstrates their belief about having a capacity to be independent and to succeed in everything and more than their sighted peers, regardless of what anybody else thinks. This can be due to the feeling that they need to succeed to prove their worth and to earn a place amongst other members of society, or it could simply be because they truly feel that they can achieve all the goals they set out for themselves.

One of the factors associated with independence is mobility. Challenges with mobility and transportation results in a person becoming dependant. Furthermore, those who rely on others are unable to be mobile on their own, which in turn links to their social interactions (Hodge & Eccles, 2013). As with the results and discussion one can see how all these aspects have overlapping facets.

When looking at the seven-stage hierarchy of needs derived by Maslow (1943), it is clear that the visually impaired young adults in this study are experiencing some of these needs (Figure 12).

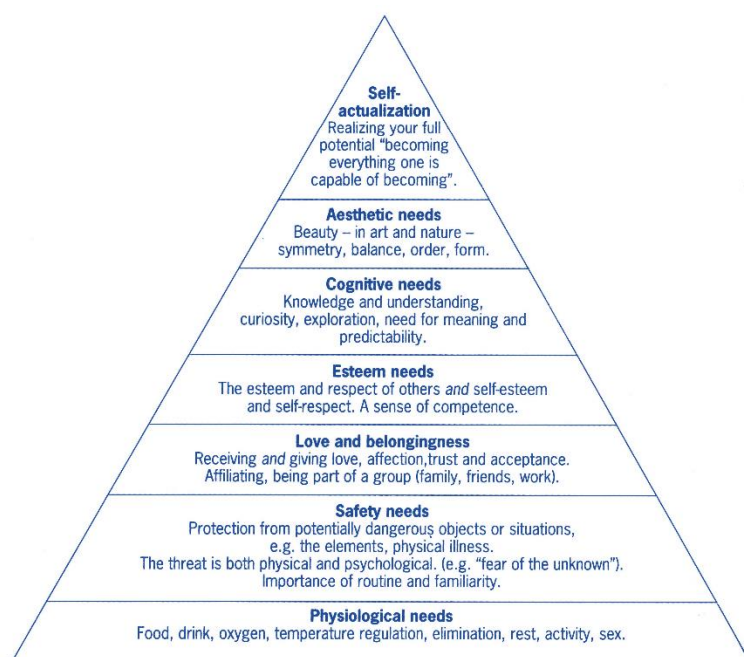


Figure 13: Maslow's hierarchy of needs <https://samirajamali.wordpress.com/2015/06/08/maslows-hierarchy-of-needs/>

The principle of this theory is that it is necessary for individuals to acquire the lower level needs before obtaining higher needs, such as self-actualization. It is said that a stepwise approach is not always followed and sometimes people shift up and down between levels and that generally one aspect is not completely fulfilled before reaching a new level, but instead, that individuals find themselves partially satisfied with some needs as well as unsatisfied and it is expected that the lower levels have a greater level of satisfaction before the higher level (Maslow, 1943). For these youngsters, the main challenges are safety needs, love and belonging, esteem needs, and cognitive needs. Some have experienced difficulty with their visual safety and will regress to this level if their visual capacity decreases or they encounter a threat to their vision. Almost all the individuals are entangled in the challenge to find belonging,

however, at the same time they seek competence. It is likely that they will not have the capacity to fully engage with their esteem needs if they have not resolved a larger quantity of their social situation. In this instance, the cognitive needs seem to overpower the other needs, as much of the conflict with the other levels is due to lack of knowledge and understanding. Nonetheless, as they shift between these four levels one can see that the implications of their visual impairment are creating a barrier to achieve self-actualization, which means that they can still live a fulfilling life, however they experience obstacles in reaching their full potential.

5.7 Chapter Summary

In this chapter, the five themes highlighted in the result section were discussed in collaboration with previous research findings that assisted in the understanding and interpretation of the research findings.

Chapter 6: Conclusion, Study Strengths and Limitations, Recommendations for Future Research

6.1 Introduction

The final chapter summarises the core findings of this study and highlights the strengths and weaknesses of the research. Furthermore, the implications of this research is noted and recommendations for future research is provided.

6.2 Conclusion

The aim of this research was to explore the experiences of visually impaired young adults living in Cape Town, and their perceptions of the implications of their hereditary visual disorder. To do this, various aspects were investigated, namely, the knowledge and understanding of the condition as well as the risks for their offspring. Furthermore, their thoughts and decisions about procreation were explored as well as their views about independence, social interactions and relationships, and other aspects which they feel have impacted their lives in some way. It was envisioned that the findings of this research could be utilized within health-care and genetic settings and various organizations for visually impaired individuals. Fifteen young adults, with various genetic visual conditions were interviewed. The data was analysed via thematic analysis and five themes were identified. Many of these aspects link to each other in some way.

Through this study it was found that most young adults did not know much about their condition and because of the lack of understanding they felt lost and confused and had many unanswered questions. Furthermore, this produces anxiety about their vision in the future, which caused some to expect the inevitability of blindness. Not only did they not comprehend their own situation, in their opinion the community, including close contacts, do not understand them. Consequently, they have acquired a longing to be understood by others as well as by themselves. Genetic counselling is a way by which they feel they can gain enough knowledge to attain control of their condition and to educate those around them. It has become apparent

that one of the main implications of having a genetic based visual impairment is a lack of understanding, which will persist until more clarity has been obtained, this creates obstacles in their identity development.

Secondly, having a genetic-based condition, means that there are implications for their future offspring. Having a lack of understanding of their conditions creates an uncertainty and confusion as to what the exact risks are, if any at all. Mixed feelings arose regards having children, for some possibly putting a child through the same situation as themselves, is not an option, whereas others would accept a child if they had the condition and think that the child would manage in the same manner as they have. There was some contemplation over their capacity as a visually impaired individual to raise children. No major concerns were voiced, however, in some instances, it was thought that the safety of the children might be jeopardised if they could not identify danger, or that their offspring might feel ashamed of them.

Thirdly, their social interactions are drastically impacted by their visual disability. They choose to protect themselves from the stigma, bullying, exclusion from activities or conversations, or being pitied, by withdrawing from social situations. Rejection from friends or family is not uncommon and in some cases the visual disability has cost them some friendships. They do want to be a part of the community, have friends and be included in social situations, thus they become frustrated, sad, angry or depressed. They attempt to rid themselves from embarrassment and to be included into social interactions easier, by hiding their disability. Their visual disability also influences intimate relationships. They do not wish to have partners who are visually impaired in fear that it would increase the risks to future offspring, as well as provide less support. The same thought process was applied for possible sighted partners. Thus, creating more challenges to finding a suitable partner.

Fourthly, visual impairment in general creates implications for mobility. First and foremost, their dreams of being able to drive are shattered and they must find alternative methods of transport. This task is challenging to say the least, as public transportation presents with many obstacles. They are unable to see the information on the trains, busses and signage; therefore, they have difficulty embarking or disembarking at the correct locations. They are afraid to

venture beyond their immediate environment in fear of not being able to see danger or how to escape it. Furthermore, there is a fear of getting hit by oncoming traffic when attempting to cross streets.

Lastly, living with a visual condition has an impact on their abilities. Their prior education at mainstream school was negatively influence due to spending much of the time being unable to see the content being taught. Due to this experience, they feel that university will be a challenge and that although they do want to continue their education, they will face similar hurdles that they have previously undergone as well as new obstacles faced with new surroundings. Independence is an aspect which leads to mixed feelings as they feel that they have the capacity to be independent and want to have control over their lives and what they can do, however their visual ability does not always permit this and at times family or friends force dependence upon them by constantly performing tasks for them or dictating what they can and cannot do. This diminishes the chance for them to prove their capability and to learn to cope without assistance. These youngsters want to fit in to society and want to prove that they can achieve what others can. This desire stems from their own goals and dreams as well as a need to fit in with society's standards. One aspect that was seen in a positive light was employment. They felt that they would easily obtain a job, due to having a disability and although they must select careers which would accommodate their impairment, most had found a field that they are interested in, therefore they believe being visually impaired has a favourable outcome with regards to employment.

6.3 Strengths of the Study

- This qualitative research seeking to explore the perceptions of the implications of visually impaired young adults with hereditary visual impairment, is the first known study of its kind that has been undertaken in the South African setting.
- The interviews were conducted in the language of the participants choice. This was very beneficial for some participants who opted to speak in Afrikaans or who used English and Afrikaans interchangeably throughout the interview. This granted the participants the opportunity to express their experiences without any communication barriers.

- All interviews were conducted in person, which means that verbal and non-verbal data could be taken into consideration.
- Open ended questions were utilized and there were no predetermined themes, which allowed the participants to answer freely, without set boundaries.
- Audio recordings provided evidence of the research and provided a more accurate record than handwritten notes would have.
- As noted by some of the participants, the interview process and the opportunity to discuss their experiences was therapeutic.

6.4 Limitations of the Study

- Not many individuals came forward or were sourced from two of the organisations. Furthermore, one of the institutions was not interested in partaking in the study. Therefore, the majority of the sample was recruited from one institution. As a result, the data collected might not be an accurate representative of other visually impaired young adults' experiences.
- Although the participants met the age requirement of eighteen to twenty-three years, the majority were still completing their secondary education at a boarding school for visually impaired individuals. This means that they have not had some experiences that school leavers have faced, and could only share what they anticipated with regards to certain aspects.
- It must be noted that in some instances they might not have responded truthfully, or they might have reacted differently than usual, because of participating in the research and to provide socially acceptable responses.
- An extent of bias is produced from purposive sampling. The views of those who are unwilling or unable to participate are not included.
- As most of the interviews were conducted at the school and the vice-principle was present when the prospective participants were approached regarding the study, some may have felt obligated to participate, even though the voluntary nature of the research was stated.
- The interviews were conducted over a compact period due to the young adults' examinations and then departing for home. As a result of this time constraint, it was not possible to evaluate, reflect on or make improvements to the interviewing process after

each interview. Nor was one transcription completed prior to the next interview. Instead this process was conducted in batches, which means that the interviews were not conducted as smoothly as hoped and the researcher might have missed opportunities to probe further into aspects that were later identified.

6.5 Implications of this Research

A few practical applications have been identified by means of this research and recommendations have been made below.

- Better systems implemented to ensure that individuals with hereditary visual conditions get referred to genetic services.
- It is of significance for genetics specialists and healthcare services to be aware of and understand the perceptions and experiences of young adults with hereditary VI, as this will equip them to meet the specialised needs of these individuals and will improve service delivery.
- Creating connections between support systems during transition into adulthood, such as schools for the blind to organisations for the visually impaired.
- Development of more efficient public transportation systems or assistive devices.

6.6 Recommendations for Future Research

Based on the current research findings, the following recommendations for future have been provided, to guide and add value to future research.

- Conduct research on middle-age or elderly individuals who have hereditary visual conditions, to explore their experiences and perceptions of how the condition has affected their lives. Whilst presenting the research at the Retina SA meeting, to find any willing participants, a middle-aged gentleman voiced an interest in partaking in such a study, which triggered the idea that such research would be valuable and welcomed.
- Explore the perceptions of visually impaired young adults who have undergone GC, regarding the implications of their condition. This would be valuable to determine the impact of the GC process on their perceptions.

- Research on young adults with more specific visual conditions could be of value to determine their life stance from the same condition. When looking at individuals with the same conditions it could be determined if some information is condition specific or if it remains the same as with a mixed group, such as the sample in this study.
- As these young adults feel that others don't understand them and that they are treated in a less desirable way by society it would be of use to research the South African society's view on VI and those who have visual disabilities.
- Investigating the experiences of visually impaired young adults who have not attended a school for 'the blind' would be helpful to gain insight on their worldviews and how this differs or correlates with those individuals who have been exposed to such an environment.

6.7 Chapter Summary

In this final chapter of the research, the conclusions were presented and the strengths and limitations were highlighted. Lastly, the implications of the research as well as recommendations for future research were noted.

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Appendix A: Information Sheet (English)

Dear Prospective Participant

A research study is being conducted under the auspices of the Division of Human Genetics at the University of Cape Town for a minor dissertation which forms part of a Masters degree in Genetic Counselling. The aim of the study is to examine/explore the views of young adults on the impact and consequences of being visually impaired/blind due to a genetic cause. The study is being done in order to gain a better understanding of how young adults perceive their disorder and the way it will affect their lives.

By doing this study we hope to determine at what level healthcare should be provided to visually impaired young adults and what aspects healthcare professionals should consider integrating into their counselling and care, with the intention of improving these services. Another anticipated outcome is to obtain information which could assist organisations for visually impaired people to provide better support and guidance to schools for the blind about areas in which to help children in preparing for transition into adulthood.

The study is aimed at visual disabilities which are associated with/as a result of a **genetic** change/fault in your DNA, therefore you have been invited to participate in the study on condition that:

- You are 18-23 years of age.
- You are visually impaired or blind because of a genetic factor.
- You are able to identify what the genetic factor is that is responsible for your disability.

A personal interview will be held in which we will speak about your understanding of your disability, some of the aspects that might be influenced by your disability and some of your experiences of being visually impaired/blind. The interview will be approximately 1-2 hours long and will be voice recorded in order make sure that your answers are captured accurately. The recordings as well as any other information obtained from you will be kept confidential and your name will not be used anywhere, thus the information will remain anonymous.

The study will not provide any medical or financial assistance for you. R150 will be given to you for the time you have offered up to participate in the study and for travel expenses incurred. Your participation is voluntary and thus you are able to withdraw from the study at any point in time, if you feel the need.

If you wish to participate in the study, please read and sign the consent form that has been provided. If you are unable to sign the form due to your visual impairment rendering you unable to write, you can give verbal consent and have a witness sign on your behalf. If you have any questions regarding the research, please contact me on PPLKAL001@myuct.ac.za or via phone at 083 337 1171.

If you have any questions regarding your rights as a research participant, please contact Professor Marc Blockman, the chairman of the Human Research Ethics Committee at the Faculty of Health Sciences of the University of Cape Town at: 021 406-6338.

Appendix B: Information Sheet (Afrikaans)

Geagte Voornemende Deelnemer

‘n Navorsingsstudie word geloods onder leiding van die Afdeling van Menslike Genetika van die Universiteit van Kaapstad. Hierdie studie is n kleiner tesis wat deel uitmaak van die Meesters graad in Genetiese Berading. Die doel van die studie is om die mening van jong volwassenes te verkry oor die impak en nagevolge van gesigsgestremdheid/blindheid weens ‘n genetiese afwyking. Hierdie studie word gedoen om beter begrip in te win oor hoe jong volwassenes hulle gestremdheid waarneem en hoe dit hulle lewens affekteer.

Ons hoop om deur middel van hiedie studie te bepaal op watter vlak gesondheidsorg aangewend moet word vir gesigsgestremde jong volwassenes, asook watter aspekte professionele gesondheidsorgers moet konsidereer en integreer in berading en sorg, om sodoende hul dienste te verbeter. Ons verwag ook om sodoende inligting in te win wat steun verleen aan organisasies wat met gesigsgestremdheid werk, asook bystand en raad aan skole vir gesigsgestremdes/blindes verleen op gebiede waar kinders voorberei word op die oorgang na volwassenheid.

Hierdie studie is gerig op gesigsgestremdhede wat geassosieer word met n genetiese verandering in die DNA, daarom word jy uitgenooi om deel te neem op voorwaarde dat:

- Ju 18-23 jaar oud is
- Jy gesigsgestremd/blind is as gevolg van ‘n genetiese faktor
- Jy kan identifiseer watter faktor verantwoordelik is vir jou genetiese afwyking

‘n Persoonlike onderhoud sal gevoer word waar ons oor jou begrip van jou afwyking gaan gesels, sekere aspekte wat jou afwyking beïnvloed en jou eie ondervinding met jou afwyking. Die onderhoud mag 1-2 ure lank duur en sal elektronies (net ‘n stemopname) opgeneem word sodat ons jou antwoorde akuraat kan weergee. Hierdie opnames, sowel enige ander inligting wat ons van jou ontvang, is vertroulik en jou naam sal nerens genoem word nie. Dus bly jy anoniem.

Daar is geen mediese of finansiele voordele uit hierdie studie nie. Verversings sal verskaf word tydens die onderhoud. Omdat deelname vrywillig is, kan jy, indien jy so voel, enige tyd van die studie onttrek.

As jy deel wil neem aan hierdie studie, lees en teken asseblief die toestemmingsvorm wat verskaf word. As jy nie by magte is om te teken weens jou afwyking nie, kan jy mondelings toestemming aan ‘n getuie gee wat namens jou kan teken. As jy vrae hieromtrent het, kontak my gerus by PPLKAL001@myuct.ac.za of bel my op 083 337 1171.

As jy vrae het aangaande jou regte as deelnemer in hierdie navorsing, kan jy vir Professor Marc Blockman, voorsitter van die Humanistiese Navorsings Etiek Komitee by die Fakulteit van Gesondheids Wetenskap by die Universiteit van Kaapstad kontak op 021 406 6338.

Appendix C: Informed Consent and Information Form (English)

MSc in Genetic Counselling Research Project

Young adults' perceptions of the implications of their hereditary visual impairment/blindness: A Cape Town based study.

PARTICIPANT INFORMATION AND CONSENT FORM

STATEMENT BY PARTICIPANT

Iconfirm that:

1. I have been invited to participate in the above mentioned research project which has been initiated by the Division of Human Genetics at the University of Cape Town because I am 18-23 years old and I am visually impaired/blind because of a genetic condition.
2. I understand that the aim of the study is to explore the views of young adults on the impact and consequences of being visually impaired/blind due to a genetic factor.
3. I am aware that the study is being done in order to gain a better understanding of how young adults perceive their disorder and the way it will affect their lives.
4. I am aware that the interview will be voice recorded. The recording will be stored in a locked cabinet and downloaded on a password protected computer and that only the researcher and supervisors will have access to the data. The recording will be destroyed after the completion of the study.
5. I understand that all information collected will be regarded as highly confidential, and a code will be assigned to any information I give, therefore my name and the names of anybody I mention will not be used anywhere in the study.
6. I understand that the information may be used for a thesis, publications in scientific journals and presentations.

7. I willingly agree to consent in taking part in this study, and I understand that I am able to withdraw from the study at any stage and that withdrawing will have no negative impact on me or my family.
8. I understand that some of the questions may cause me to feel emotional, but the risk of harm is small and arrangements for counselling will be made if I require it.
9. I understand that if I agree to participate in the study without being fully aware that my condition is genetic, information may arise during the interview which might shock and upset me. This will cause the interview to be terminated immediately and I will be referred to a genetic counsellor.
10. I understand that there will be no medical and financial benefits to me from this study.
11. I understand that I will be given R150 for the time and transportation used to attend the interview.
12. I am aware that the possible outcomes of the study may include improvement in healthcare, better support from organisations and assistance for children that are transitioning into adulthood.
13. I have been given the researcher's as well as the chairman of the research ethics committee's contact details should I need any further information or want to ask questions about the study.
14. _____ has explained the information of the study to me in English/Afrikaans or any other preferred language and I clearly understand the information

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

Signed at:

(address) _____ on _____ 2016

Participant's name

Witness's name

Participant's signature

Witness's signature

MSc in Genetiese Berading Navorsings Projek

Jong volwassenes' persepsies van die implikasies van hulle oorerflike gesiggestremdheid/blindheid: 'n studie gebaseer in Kaap stad

DEELNER INLIGTING EN TOESTEMMINGSVORM

VERKLARING DEUR DEELNEMER

Ek.....verklaar dat:

1. Ek genooi is om deel te neem aan bogenoemde navorsingsprojek wat deur die afdeling van Menslike Genetika by die Universiteit van Kaapstad geloods is, omdat ek 18-23 jaar oud is en gesigsgestremd/blind is weens 'n genetiese toestand.
2. Ek verstaan dat die doelwit van die studie is om jong volwassenes se uitsig aangaande die impak en gevolge van gesigsgestremdhied/blindheid as gevolg van 'n genetiese faktor te verken.
3. Ek bewus is dat die studie gedoen word om beter begrip te kry oor hoe jong volwassenes hulle gestremdheid waarneem en watter effek dit op hulle lewens het.
4. Ek bewus is van die stemopname wat tydens die onderhoud gemaak gaan word, Dat die opname in 'n geslote kabinet sal gestoor word en op 'n rekenaar wat met 'n wagwoord beskerm is afgelaai word, en net die navorser en toesighouers toegang tot die data het. Hierdie opname sal vernietig word aan die einde van die studie.
5. Ek verstaan dat alle inligting wat ingesamel is, hoogs vertroulik is, en 'n kode toegestaan word vir inligting wat ek verskaf. My naam of enige ander name wat ek verskaf, sal nie in die studie gebruik word nie.
6. Ek verstaan dat die inligting mag gebruik word vir 'n tesis, vir publikasie in wetenskaplike joernale en aanbiedings
7. Ek vrywilliglik instem om deel te neem aan die studie en ek verstaan dat ek enige tyd kan onttrek sonder nadelige effek op myself of my familie.
8. Ek verstaan dat sommige van die vrae vir my emosioneel mag wees en dat daar 'n klein moontlikheid vir skade mag wees en berading vir my aangebied sal word as ek dit vereis.
9. Ek verstaan dat as ek instem om deel te neem aan die studie sonder om heeltemal bevus te wees dat my toestand geneties is, mag ek tydens die onderhoud inligting ontvang

aangaande my genetiese toestand wat my moontlik mag skok of ontstel. Die onderhoud sal dan onmiddelik gestaak word en ek sal verwys word vir genetiese berading.

10. Ek verstaan dat hierdie studie vir my geen mediese of finansiële voordele inhou nie.
11. Ek bewus is dat die moontlike uitkomst van hierdie studie verbetering in gesondheidsorg, beter ondersteuning van organisasies en bystand vir kinders in die oorgangstyd na volwassenheid kan beteken.
12. Ek die kontakbesonderhede ontvang het van die navorser, sowel die voorsitter van die navorsers etiek komitee, sou ek enige verdere inligting wou he of vrae vra aangaande die studie.
13.het al die inligting aangaande die studie in Engels/Afrikaans of enige gekose taal aan my verskaf, en ek verstaan dit duidelik.

EK VERKLAAR DAT EK VRYWILLIGLIK INSTEM OM DEEL TE NEEM AAN DIE BOGENOEMDE NAVORSINGSSTUDIE EN DAT DIE ONDERHOUD OP BAND OPGENEEM MAG WORD

Geteken by:(adres)_____op_____2016

Deelnemer se naam

Getuie se naam

Deelnemer se handtekening

Getuie se handtekening

Appendix E: Demographic Questionnaire

Instructions: Please answer the following questions below by ticking the appropriate box or by writing your answer in the space provided.

1. What is your gender

- Male
- Female

2. What is your ethnicity?

- Black
- White
- Coloured
- Indian
- Other

3. How old are you?

- 18
- 19
- 20
- 21
- 22
- 23

4. Are you blind or visually impaired?

- Blind
- Visually impaired

5. What is the diagnosis of your condition?

6. Is it genetic? Add the type of inheritance if you know what it is

- Yes
 - No
-

7. What is your relationship status?

- Single
- Married
- Divorced
- Informal relationship

8. Do have children?

- Yes
- No

9. What is the highest level of education you have completed?

10. What is your employment status?

- Employed
- Unemployed
- Other

Appendix F: Interview Guide

Main interview questions with possible probes (if not explored by participant in detail initially)

1. Tell me more about what it is like being a person who has a genetic visual impairment/blindness?

- Tell me about your life as a visually impaired person.
- In what way do you think that your genetic visual condition affects your relationships/social interactions with people?
 - Finding a partner and being in a relationship?
 - How do you think society views you?
- What influence has having this genetic visual impairment had on your education? How about in the future?
- How did/might your genetic visual disability impact finding a job?
- What are your thoughts about accommodation and independence with regards to being visually impaired?
- Living with genetic visual impairment, what will it mean for your future?
- Adjustments/adaptions?

2. What is your knowledge and understanding/interpretation of your genetic visual condition?

- How do you think you got the condition?
- What are the signs/symptoms?
- Do you think other people understand your condition and situation? Why do you say that?
- Have you had any difficulty understanding what your condition is?
- Will it be helpful to you if someone would explain your condition and the genetic factors regarding your condition to you? Why?
- What do you think will happen to your vision in the future?
- Tell me about healthcare services that are available/will become available to you?
- How do you think having genetic visual impairment might impact your health in other ways? E.g. mentally, physically.
- How do you think the fact that your disorder is genetic influences your life?

3. Can you describe how genetic visual impairment/blindness has affected your decision-making around having children?

- What would it be like to have a child/children with the same condition as you?
- What, if anything would you do differently in raising a child with visual impairment, compared to how you were raised?
- What challenges do you think you might face having/raising children while having a visual disability?

4. Tell me about your experience of the obstacles you have faced over the last couple of years? (Integration into adult world)

- It sounds like it was difficult getting around – could you explain a bit more
- What types of things have you found easy and difficult having genetic visual impairment?
- What do you feel could have been done by your school or society to better prepare you for the transition into adulthood?

5. What are your expectations for the future?

- How do you feel about your future?
- What else would you like to tell me more about?

Appendix G: Ethics Approval



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



Room E53-46 Old Main Building
Grootescheur Hospital
Observatory 7925
Telephone (021) 406 6626
Email: shunetta.thomson@uct.ac.za

Website: www.health.uct.ac.za/fhs/research/humanethics/forms

15 July 2016

HREC REF: 376/2016

Prof J Greenberg
Human Genetics
Department of Pathology
Level 3, Wernher and Belt North
IIDMM

Dear Prof Greenberg

PROJECT TITLE: YOUNG ADULTS' PERCEPTIONS OF THE IMPLICATIONS OF THEIR HEREDITARY VISUAL IMPAIRMENT/BLINDNESS: A CAPE TOWN BASED STUDY- LINKED TO 226/2010 (MSc candidate- Ms K Popel)

Thank you for your response to the Faculty of Health Sciences Human Research Ethics Committee dated 27 June 2016.

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

Approval is granted for one year until the 30th July 2017.

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

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

Please quote the HREC REF in all your correspondence.

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

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

The HREC acknowledge that the student, Kalinka Popel will also be involved in this study.

Yours sincerely

Signed

PROFESSOR M BLOCKMAN
CHAIRPERSON, FHS HUMAN RESEARCH ETHICS COMMITTEE
Federal Wide Assurance Number: FWA00001637.
Institutional Review Board (IRB) number: IRB00001938

HREC 376/2016

Appendix H: Permission from Athlone School for the Blind

ATHLONE SCHOOL FOR THE BLIND
Private Bag X1
KASSELSVLEI
7533
Athlone Street, Bellville South
Athlonerass. Bellville-Suid
FOUNDED 1907 GBSTIG



ATHLONE SKOOL VIR BLINDES
Privatebag X1
KASSELSVLEI
7533
TEL: 021 2234 • (021) 951 2233
FAX: 021 5118
E-MAIL: rcs@athlonetblind.org.za

To whom it may concern

The Division of Human Genetics at the University of Cape Town has approached the Athlone School for the Blind to be a partner in their research programme on The Genetics of Inherited Retinal Degenerative Diseases.

As a school, we are happy to be involved in this project, as there are benefits for our past, present and future pupils and their families. These benefits include diagnosis, potential treatments and interventions. Furthermore, knowledge of the inheritance patterns of these conditions could benefit other family members who are not aware of the risks of passing on their condition to future generations. Genetic counsellors at UCT have also committed to providing education to our staff and pupils which will improve our understanding of inherited causes of blindness.

Signed

Dr John Philander
(Deputy Principal - Psychologist)

Signed

Mr F Fisher
(Principal)



Date: 31 May 2010

Appendix I: Permission from LOFOB

MEMORANDUM OF UNDERSTANDING

between

THE LEAGUE OF FRIENDS OF THE BLIND (NPO 002-921)

(Hereinafter referred to as LOFOB)

and

Kalinka Popel Student MSc (Med) Genetic Counselling (UCT)

(Hereinafter referred to as the research student)

PREAMBLE

1. This Memorandum of Understanding (MOU) is the result of discussions between the Management of LOFOB and the research student.
2. This MOU reflects a spirit of cooperation between the parties and reflects the broad intent of the two parties.
3. This MOU will be effective as from date of signature and can be reviewed, by mutual agreement, whenever necessary.

PARTNERSHIP AND COLLABORATION

1. The two parties hereto undertake to partner and collaborate in the field of visual impairment and disability, and the discipline of Genetic Counselling.
2. The following general forms of collaboration will be pursued:
 - 2.1 Teaching and learning in the field;
 - 2.2 Exploring possibilities for, and conducting, relevant research through joint studies and joint publication;
 - 2.3 Promoting and sharing information generated in mutually agreed on ways, including teaching, sharing, learning to clients within the LOFOB programme;
3. The following specific forms of collaboration will be pursued:
 - 3.1 Introduce the research student to LOFOB and its services;
 - 3.2 Provide theoretical input to the research student on visual impairment, blindness and disability;

3.3 Accommodate the research student at LOFOB to conduct research on the mutually agreed upon, relevant research topic.

3.5 All publications produced as a result of the collaboration for the specific/ specified research project must include The League of Friends of The Blind (LOFOB) or designated representative and its clients as co-authors.

4. The following considerations are made towards voluntary participation:

4.1 All parties involved acknowledge that participation is on a voluntary basis and this MOU in no way constitutes an employment agreement/contract and/or a financial agreement.

4.2 Any additional volunteers to be associated with the programme or projects must complete a LOFOB volunteer form and be approved by LOFOB management.

CONDUCT

1. All students/additional students/volunteers involved with the programme or projects related to the programme will undertake to uphold and abide by the LOFOB 10 Point Code of Ethics and the LOFOB Service Standards.

PERIOD

1. This MOU will remain in force for a period of 12 months from the date of signing by both parties.

2. This MOU may be terminated at any time by mutual consent of the two parties hereto or by 1 months' notice in writing by either of the two parties.

3. If this MOU remains dormant for a period of not less than 3 months it will be deemed that it has lapsed and is terminated by mutual tacit consent of the parties.

4. Where the MOU continues to be active the two parties hereto agree to review it and to consider its renewal for another period of 12 months.

The Research Student:

Ms Kalinka Popel

(UCT)

Signed: 

Date: 07/07/2016

LOFOB Contact Person:

Mrs Benita Petersen

(Manager)

Signed: 

Date: 20160713

Appendix J: Original Quotations in Afrikaans

Quotation 1:

“Ek kan nie ‘n kar bestuur nie. Umm ek moet mense vra om my na sekere plekke to te ry, en ‘n paar van die mense sal nee se, wanneer dit dringend is of iets soos dit.”

Quotation 2:

“Going over the road, I should be injured right now because of the amount of times I got stumped. I don’t know what the word in Afrikaans, in English. Ek was al klom keer gestamp deur bakkies en al ander goeter. Ek kon regtig beseerd gewees het.”