

Exploration of the impact of genetic counselling and patient support group involvement on retinal degenerative disorders (RDD) patients: a qualitative study

By

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Abstract

This study explored the experiences of individuals within the clinical genetic service in South Africa, with inherited retinal disease.

Methods

This qualitative research was based on a grounded theory approach. Semi-structured interviews were carried out after obtaining approval from the University of Cape Town Research and Ethics committees as well as from study participants themselves. Ten willing individuals were interviewed using an open-ended question guide to extract information about their experiences with vision loss including but not exclusively, concerning participants knowledge about their condition, ability to make decisions and their adaptation experiences. Interviews were audio recorded, transcribed verbatim and a reiterative thematic analysis performed on the raw data.

Results

A range of themes and sub-themes emerged from the data that marked participants' experiences from the time of onset of vision loss. The main broad themes include "a roller coaster journey," "learning how to survive," "a genetic condition affecting the back of the eye, and "paving the way forward."

Conclusions

The four themes presented in an almost linear fashion, marking a journey patients experienced from the onset of symptoms to the time of the interview. The initial stage of their journey was marked by difficulties and challenges exacerbated by the psychopathological responses reported in this dissertation. This stage was perceived to be prolonged and often extended into adulthood. The messages from the data support a view that the diverse and extended nature of this initial period was due to the limited support this group of participants received from the low vision specialists they consulted with. A significant impression of frustration and disillusionment was experienced by participants on not receiving information and guidance from these health care providers. Various mechanisms of support and control positively influenced patients' learning how to cope and survive their journey with visual impairment. Coping, learning what their condition is, what the causes are and the risks involved for future generations, empowered the participants in this study to obtain a view of a future for themselves and to make relevant decisions and choices necessary to realise the way ahead. Taken together though, continued education was deemed worthwhile.

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List of Terms and Abbreviations

| | |
|------|---------------------------------------|
| AGM | Annual general meeting |
| ARMD | Age-related macular dystrophy |
| CGS | Clinical genetic service |
| CSNB | Congenital stationary night blindness |
| IRD | Inherited retinal disorders |
| LOFB | League of friends of the blind |
| LCA | Leber congenital amaurosis |
| MD | Macular dystrophy |
| NGS | Next generation sequencing |
| NHLS | National Health Laboratory Services |
| PCR | Polymerase chain reaction |
| RP | Retinitis pigmentosa |
| SA | South Africa |
| STGD | Stargardt disease |
| UCT | University of Cape Town |
| VI | Visually impaired/ment |
| WHO | World health organisation |

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Chapter 1 BACKGROUND

1.1. Introduction

The personal impact of a genetic diagnosis on an individual, is far reaching as it often redefines or labels the very nature of the person and in many instances, the family at large. The permanent nature of genetic disorders influences the physical, psychological as well as the social health of patients and their families. The impact, however, depends on the condition, the relationship to the affected individual, and individual personalities. To further confound the situation, each family functions with their own set of interpersonal dynamics. These factors can evoke a myriad of emotions including isolation, fear, guilt, and helplessness (Genetic Alliance. 2009, Garcia et al. 2017). A clear comprehension and grasp of the possible and expected reactions and responses to diagnoses can therefore be useful to direct a course of action or approach aimed to minimise any negative effects and maximise the ease with which patients adapt and journey to a place of acceptance and general well-being.

Patient support services from genetic counsellors, psychologists, social workers and patient support groups play an important role in addressing the issues facing the patient and family with a genetic condition. To this end, clinical genetic services (CGS) encompassing these support services, have been established for several inherited disorders in the Division of Human Genetics at UCT, together with the National Health Laboratory Services (NHLS). The inherited retinal disorders (IRD) genetic service, however, is the only one offered in close collaboration with the support group, Retina SA.

The focus of this study is the exploration of the experiences of patients with retinal disorders in the South African IRD clinical genetic service.

1.2. Inherited Retinal Disorders

The World Health Organization (WHO) proposed the following definition for blindness. “*A physical, psychiatric, intellectual or sensory impairment, whether temporary or permanent, provided that it lasts for a significant period of time, that limits the capacity to perform one or more essential activities of daily life and which can be caused or exacerbated by economic and social environment*” (Sihota and Tandon. 2015).

Inherited retinal disorders are a group of degenerative diseases which cause vision loss and contribute to a significant proportion of the disease burden. It is reported that approximately 161-259 million people worldwide suffer from visual impairment (Dandona and Dandona. 2006). Based on statistics South Africa's population census of 55.91 million, it can be extrapolated that this country could have approximately 600,000 people suffering from IRD.

IRDs comprise of a large number of conditions causing progressive degeneration of the light-sensitive photoreceptor cells of the retina, which causes visual impairment and may lead to total blindness. Depending on the specific clinical type of IRD, patients may manifest different symptoms such as night blindness, impaired vision, tunnel-like vision and loss of peripheral vision, light sensitivity, retinal detachment to total loss of vision (Sullivan et al. 1996).

To date several heritable forms of retinal vision loss have been fully characterised. As a group the retinopathies, are one of the most clinically and genetically heterogeneous diseases, and this complicates patient diagnosis (Gillespie et al. 2014).

IRD phenotypes are grouped based on the type of primary photoreceptor cells affected. Patients with primary degeneration of rod photoreceptor cells experience night-blindness and restriction of the visual fields i.e. the loss of peripheral vision, which translates to the patient experiencing tunnel-like vision. These diseases can be congenital or have a later onset, and are either stationary (e.g. Congenital stationary night blindness, CSNB), or progressive (e.g. Leber congenital amaurosis, LCA, or retinitis pigmentosa, RP) (Rivolta et al. 2002). Patients with primary degeneration of cone photoreceptor cells or 'macular dystrophy'(MD), experience loss of central vision with an early age of onset (e.g. the juvenile-onset Stargardt macular dystrophy, STGD) or later (e.g. Age-related macular degeneration, ARMD) (Ayuso and Millan. 2010). Reduced penetrance can occur resulting in intra-familial disease variability (Rivolta et al. 2002).

The phenotypic overlap between the different clinical forms of IRDs can delay a clinical diagnosis, which in turn makes providing a patient with a prognosis somewhat challenging. This is compounded by changes in the clinical presentation (phenotype) as the disease progresses. The uncertainty and labile nature of these challenges often results in a great deal of frustration and confusion for patients.

Characterisation, however, of the molecular basis of the disease (in support of a clinical diagnosis) in each family in which the disease segregates, can improve the understanding and management of the disorder for each member. A definitive molecular genetic diagnosis therefore supports and clarifies a clinical diagnosis (Gillespie et al. 2014). A confirmed molecular diagnosis is thus a crucial step in the genetic management of patients and families as it provides a sense of certainty and direction (Berger et al. 2010) and often a measure of control for the patient. A genetic diagnosis therefore provides meaning and opens up avenues for diagnostic, predictive and carrier testing for family members. Also, a molecular diagnosis is crucial, in light of the number of clinical trials and treatments currently being investigated and developed for the IRD group of disorders (<http://www.clinicaltrials.gov/>).

Genetic factors, however, which confound reaching a definitive molecular diagnosis for an IRD include different inheritance patterns such as autosomal dominant, autosomal recessive and X-linked, although isolated cases are common (Berger et al. 2010). Less common atypical patterns which include mitochondrial, digenic, tri-allelic and uniparental disomy are also seen (Rivolta et al. 2002). Furthermore, to date more than 190 different IRD genes have been identified (Daiger et al. 2010) and within this already complex genetic landscape of IRDs, locus heterogeneity, genetic heterogeneity, allelic heterogeneity and clinical heterogeneity have been demonstrated. It is therefore not surprising that it is reported that 50-70% of cases can only be assigned to known IRD genes (Nishiguchi and Rivolta 2012). South African IRD research findings (with an even lower detection rate), suggest though that patients in this country either carry novel mutations in known IRD genes, or that their causative genes are novel (personal communication with Dr Lisa Roberts, Ph.D., University of Cape Town). This situation makes confirming a specific clinical diagnosis more challenging within the South African context.

Recent technological advances in gene identification and cell biology techniques have facilitated a dramatic surge in the understanding of retinal diseases. As a result, so much more is known and understood now than a mere 20 years ago, about the genetic defects which underlie IRD as well as the molecular pathophysiological mechanisms (Francis. 2006).

Molecular genetic testing is therefore the crucial first step toward confirming the phenotypic diagnosis at the DNA level. A genetic diagnosis facilitates the much needed clarity patients and families often need, as it affords a more precise prognosis, accurate and informed genetic and reproductive counselling. In addition, registering patients with a confirmed genetic diagnosis establishes a base for future clinical trials eligibility.

1.3. Psychosocial Implications of vision loss

Genetic information is often more sensitive than other medical information largely due to what it holds for the future (it's predictive nature), its permanency, and potential implications for the broader family circle. Many genetic conditions may require continuous attention and lack cures or even treatments. So a genetic diagnosis for one family member may mean that other relatives may be at risk of developing the condition. Genetic disorders also present challenges on the emotional and reproductive levels. As a result a myriad of psychosocial issues could arise depending on the unique personal and family dynamics which may exist.

With respect to individuals who experience vision loss or impairment, it is reported that many experience challenges in their lives (Ishtiaq et al. 2016). The restrictions blindness causes could include the inability to act at one's own will, the ability to move around freely and in the exercise of personal control. This study showed that blindness or low vision therefore often precipitates psychological implications such as guilt, anxiety, sadness, depression and feelings of dejection and desolation. Socially, the visually impaired face several challenges and therefore often choose to live in isolation (Sacks et al. 1992). However, other studies do show a marked level of social capability, and an insignificant tendency to social isolation (George and Duquette. 2006).

In an interview with Dr Shahid (Fellow of the American Academy of Optometry), she says that the most common psychological impact, of visual impairment is depression. According to the Preventing Depression in ARMD trial (Arch Gen Psychiatry. 2007;64:886-892), 30% of patients with AMD will experience depression within a few months of their second eye becoming affected. Social challenges such as unemployment and divorce are not unusual. Fear and anxiety is also common due to the anticipation of future vision loss or blindness. Phobic anxieties, such as fear of open spaces, traveling in crowded areas, and being left alone are also commonly reported among the visually impaired (interview with Dr Shahid 2017, <http://www.medscape.com/viewarticle/773451>). Dr Shahid also says that the visually impaired “tend to have fewer years of formal education, lower incomes, and increased dependence on public-assistance programs.” These views with respect to the psychosocial impact of VI as expressed by Dr Shahid are supported in published literature (Kempen et al. 2012).

Since IRDs are usually slowly progressive in nature, visual impairment may only become a challenge in their daily lives, in adulthood. By this stage, many would already have families and they would have to deal with the loss of a very important function in their lives. It is therefore not surprising that the impact of blindness on an individual has been linked to a grief reaction, where the person mourns loss of the sighted self (Leo et al. 1999). The person therefore has to adapt to the new reality of vision loss, by going through the stages of grief as outlined by Kubler-Ross (1969), i.e. denial and isolation, anger, bargaining, depression and finally acceptance.

Patients with potentially inherited vision loss must therefore learn to adapt to function without normal vision, while coming to terms with the reality that they may have passed the underlying genetic defect, on to their children.

Dr Shahid therefore says, that the focus of appropriate interventions should be on helping patients establish and maintain an optimistic attitude, assisting patients to be proactive about helping themselves, and guiding them to attain a sense or measure of control over their lives (interview with Dr Shahid 2017, <http://www.medscape.com/viewarticle/773451>).

Most interestingly, Dr Shahid says that although clinicians and patients alike, are unable to control the course of IRDs, how they respond to changes during disease progression, is something they can control. Acceptance involves a pragmatic grasp of what the limitations created by vision loss are, and what real capabilities remain. It means that health care providers and support services play an important role in helping patients maintain a balance between independence and assistance.

1.4. Clinical Genetic Service: Inherited Retinal Disorders

The IRD CGS in SA employs a multidisciplinary approach to service delivery in an effort to best meet the needs of patients and their families. This approach networks the expertise and skills of medical and molecular geneticists, psychologists, genetic nurses, social workers, genetic counsellors and Retina SA patient support.

Most families faced with the challenge of retinal disease, seek for information relating to: the diagnosis, prognosis, treatment and presymptomatic detection of the disease (Ganesh and Keep. 2011). The CGS team aims to address these areas of concern (amongst others) by providing patients and families with:

- a confirmed clinical and molecular or DNA diagnosis, where possible
- genetic counselling, information and risk assessment pertaining to their diagnosis
- access to a predictive genetic testing programme for those at-risk of a confirmed late-onset diagnosis in their family
- prenatal diagnosis for women with a confirmed carrier or affection status
- opportunities to participate in research programmes (where appropriate)
- referral to additional healthcare disciplines such as psychology, physiotherapy and occupational therapy (where applicable)
- practical social support and awareness through Retina SA membership

Retina SA is a national, non-profit organisation established in 1980 to benefit and support patients and families affected with IRDs. This support is offered through a number of focus areas which include:

- financial support of the IRD research project at the Division of Human Genetics, UCT to find the genetic mutations in South African families
- Facilitation of genetic testing to identify those patients who will benefit from clinical trials
- Public Awareness of retinal degeneration

- Patient support including education, referral and intervention
- Collaboration with local and international agencies and researchers
- Fund raising to support this work

(retinasa.org.za)

The high number of genes known to underlie IRD creates several challenges for molecular testing with respect to cost, time and choosing an effective testing modality i.e. the DNA panel most likely to hold the gene causing the retinal disease in a given patient, and therefore to yield a positive result. For these reasons, Retina SA has made it a dedicated focus to drive the journey from patient and family clinical diagnosis to gene mutation identification i.e. genetic diagnosis for its members. In South Africa, this is achieved through a collaborative effort between Retina SA and the CGS at UCT. The ultimate goal being to facilitate the restoration of vision to those carrying gene mutations appropriate for available gene-based therapies.

The drive to have all its members tested is achieved through distribution of detailed information sheets outlining the process. Once an interest to test is indicated, a pre-test genetic counselling session is arranged in order to protect the autonomy of patients with respect to whether they want to be tested or not. The process of choosing an effective testing modality involves genetic counsellors taking a detailed history to ascertain the possible inheritance pattern. The patient then undergoes a thorough clinical assessment to review and document the retinal pathology so that the subsequent genetic testing results may be interpreted within the correct clinical context. With informed consent, Retina SA funds the genetic testing of its members either in full or partially, in order to expedite a genetic diagnosis for the patient and IRD community of South Africa as a whole. The funding of testing is a last resort for those who want to be tested but are unable to. If the member decides to proceed with testing, informed consent is obtained and bloods are taken and sent for testing at Asper Ophthalmics. Results are verified at UCT and a post-test genetic counselling session is arranged for delivery of the results which will include an explanation of the implications. Consent is again provided by the member for the results to be sent to Retina SA and documented on their mutation database (registry) for its members.

The motivation of Retina SA for driving the DNA testing of its members, is that mutation identification is the prerequisite for clinical trial participation. The ultimate goal being to

facilitate the restoration of vision to those carrying gene mutations appropriate for available gene based therapies. In addition, the group keeps the hope of a cure or treatment alive among its members through the distribution of newsletters, detailing research trends and discoveries, particularly in the area of IRD treatment modalities. Retina SA also promotes awareness through regular meetings and annual general meeting (AGMs). These platforms allow Retina SA to very effectively create awareness amongst its members of developments with regard to newly available IRD genetic testing gene panels. Retina SA also offers practical help with the daily needs of those facing similar challenges associated with vision loss. Patients' association with support groups can influence the uptake of management recommendations as well as the acceptance of his/her diagnosis.

The pre- and post-test counselling aspect of the CGS focuses on meeting the educational, guidance in the decisions patients are faced with, as well as the psychosocial needs of the client. The primary goal of pre-test genetic counselling is to guide the patient through the process of making an informed decision about whether to proceed with genetic testing or not and what is the appropriate testing option. For retinal disease the options are varied and can be challenging to navigate (Figure 2.3). During this session, the genetic counsellor will explain and outline the benefits as well as the limitations of the tests that are available. The potential testing outcomes and associated implications, both for patients and other family members are also discussed at this pre-test session. The post-test session covers the delivery of test results together with an explanation of the interpretation and personal significance. The possible implications for other family members would also be addressed. The genetic counsellor also offers support and assistance with any other decisions which are a consequence of the results (Ganesh and Keep. 2011).

It is therefore not surprising that reports have taught that in order to be effective as a genetic counsellor, a patient centred approach to assisting the patient in his/her decision making process is employed to ensure that the unique needs of each patient are explored, assessed, acknowledged and addressed. This is in keeping with the generally accepted definition for genetic counselling which states that:

“Genetic counselling is a communication process, which aims to help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions” Resta et al. 2006.

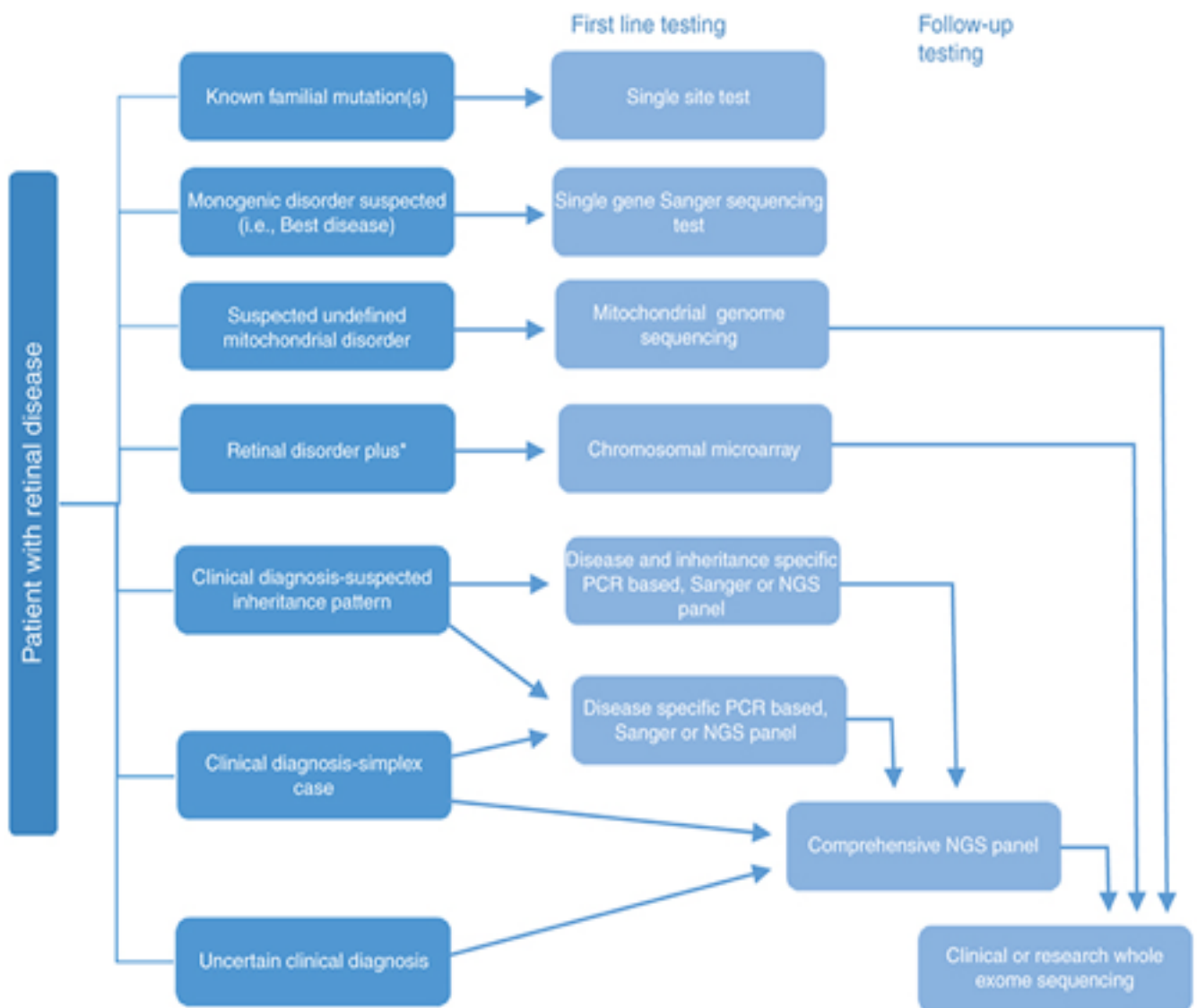


Figure 1.1. Genetic testing options for patients with retinopathies. Genetic testing for patients with retinal disorders should be considered on a case-by-case basis. This schema illustrates available options for initial genetic testing and follow-up analysis, if required. *Retinal disorder plus refers to patient with a retinal dystrophy in combination with additional systemic features, developmental delay/intellectual disabilities, and/or congenital malformations. NGS, next-generation sequencing; PCR, polymerase chain reaction. (Taken from Kristy and Seema. 2015)

It is of particular interest to note that inclusion of genetic counselling into CGSs improves patient uptake of management (Rutherford et al. 2014). As the process of genetic counselling is guided by the patient's needs, each patient experience is expected to be

subjective as well. In fact, Weil (2003), suggested that the psychosocial context of a patient be the guiding aspect of genetic counselling. Therefore, it is suggested that exploring experiences of a patient would best be investigated using a patient's perspective approach; hearing what they have to say (McAllister et al. 2016). The approach employed in the CGS offering to IRD patients and their families, aims to meet their needs and to guide them through the process of clinical and genetic diagnosis as well as the subsequent journey to acceptance of his/her new reality.

Taken together, IRD patients in South Africa, have the benefit of the collaborated efforts of Retina SA and the clinical genetic service which includes the services of qualified genetic counsellors.

1.5. Rationale

IRD patients' experiences dealing with the challenges of visual impairment, who have had the support and advocacy of a patient group such as Retina SA, which reinforces the goals of genetic counselling, with respect to psychosocial support, education and awareness, has to date not been explored. This study will therefore explore a group of patients' perspectives on their journey with this joint venture with respect to knowledge about their condition, reproductive behaviour, adaptation and coping while also allowing the participant to introduce themes of their own.

1.6. Research Question

How have IRD patients' experiences with the combined input of Retina SA and genetic counselling influenced their education, decision making and adaptation, with their genetic diagnosis?

1.7. Aim and Objectives

To investigate a group of IRD patients' perspectives on their journey with genetic counselling and Retina SA support. To specifically explore:

1. participants' understanding of the medical and genetic aspects of their diagnosis
2. how their counselling and support group experiences have influenced reproductive and life choices (decision-making)

3. how their counselling and support group experiences have influenced how they have adapted to their diagnosis

Chapter 2 METHODOLOGY

2.1. Research Context And Study Design

Study participants, who have had DNA diagnostic testing and genetic counselling, were recruited from the Retina SA membership. Personal interviews guided by open-ended styled questions, were conducted after informed written consent had been received for participation in the study. The data collected was recorded and transcribed. The transcribed data were then analysed. The research approach and methods followed are reported in this chapter.

A qualitative research approach was used in this study which is exploratory by definition rather than quantitative. Qualitative research simply means "any kind of research that produces findings not arrived at by means of statistical procedures or other means of quantification" (Strauss and Corbin. 1990). The research approach aims to explore an individual's perception and understanding of the world based on their experiences in a real world setting (Merriam. 2009). The approach exposes trends in thought and opinions and is designed to extract rich and detailed descriptions from study participants by allowing them to express themselves freely (Tuli. 2010). This approach was thus deemed best suited to answer the research question. The qualitative research approach used in this study was based on a grounded theory framework, which is an iterative process in the development of theories, concepts, ideas or themes based on observations. The approach uses a method of data collection best suited to the research question (Thornberg. 2012).

Some of the common qualitative research data collection methods include participant/ observations, focus groups (group discussions), and face-to-face interviews, (Kawulich. 2005).

The face-to-face research interview specifically explores the experiences, views and beliefs of individual study participants and therefore generates a 'deeper' understanding of the person's lived story (Silverman. 2000). This understanding is achieved because the method allows study participants the opportunity to express themselves freely and in so

doing, to highlight any themes not specifically probed for in the interviews. Detailed exploration of a single participant's reactions in the context of personal experiences is also possible without bias or influence from other participants, which may occur in a focus group situation. Furthermore, the private nature of the method makes it best suited for exploring sensitive topics which could be very personal, as participants may not feel comfortable discussing such issues in a group setting. The interview method for data collection was therefore thought to be most appropriate for this study which involves exploring the experiences of individuals with IRD.

Qualitative data collection methods usually involve the use of either structured, unstructured or semi-structured interviews. Structured interviews involve rendering a preset questionnaire with minimum deviation and scope for follow-up questions to prompt further elaboration on a theme of interest. They therefore are less appropriate if in-depth responses are required. Unstructured interviews on the other hand, are approached with no preconceived themes of interest and are administered with limited planning (May. 1991). This method is therefore usually very time-consuming and difficult to manage, in the absence of a clear area of interest. This method is therefore most useful when significant 'depth' is required, or if the topic is not well known. The final data collection method is the semi-structured interview which is guided by a set of questions designed to elucidate particular preconceived themes of interest. This method, however, allows flexibility for unplanned questions into areas where more detail may be deemed necessary during the interview (Britten. 1999). The guided approach aims to glean information concerning the same general predetermined focus areas from each interviewee, while maintaining a degree of freedom and adaptability in getting information from the interviewee. The attributes of this method often facilitates its use in healthcare research because this approach also lends itself to uncovering themes not preconceived by the researcher (Gill et al. 2008). The semi-structured interview method was therefore thought to be most appropriate for the current study in order to address the research question within the CGS setting which framed the context of this explorative study.

Free expression during interviews was further facilitated using open-ended questions within the semi-structured guided interview which allowed each participant to be comfortable to express themselves using their own style of language. The open-ended question approach allowed each participant the opportunity and platform to reveal as

much information with which they were comfortable, facilitating the collection of a set of rich qualitative data for further analysis. Furthermore, open-ended styled questionnaire responses or free text comments have been shown to be a valuable source of qualitative information which lends itself to subsequent thematic analyses (Rich et al. 2013). The open-ended questions give the researcher the flexibility to ask follow-up questions relating to a particular response.

Interviews were conducted at a location of the participant's choosing. This field-based method was used to ensure that participants did not feel intimidated or inhibited during the interview.

In qualitative research, reliability and validity is taken together as the credibility of the study design; Lincoln and Guba (1985) states that: "Since there can be no validity without reliability, a demonstration of the former (validity) is sufficient to establish the latter (reliability)." A number of terms are used to describe the concept of validity in qualitative research and these include quality, rigor and trustworthiness (Davies & Dodd. 2002). It is also reported that these concepts are dependant primarily on the ability and effort of the researcher conducting the research study (Golafshani. 2003). Furthermore, a mutual understanding between the researcher and participant of events and their significance also adds to the credibility of the study. Johnson (1997) suggested some strategies to enhance the validity of qualitative research studies and which were incorporated into this study design to establish a reliable picture of the realities of participants. These strategies included the use of multiple methods of data collection such as observation, taking of field notes during and after each interview (on observations, thoughts and ideas to help with subsequent data analysis), interviews and recordings. To reduce bias, the verbatim language of participants were used, the use of low-inference descriptors as well as unrushed and extensive field work sessions were embedded into the study design. Furthermore some data transcripts were re-analysed by a study supervisor at a different time and place in order to confirm and enhance the analysis and to get an alternative perspective on the themes present in the data to ensure rigour.

2.2. Participants

The study cohort was derived from the visually impaired population of South Africa, either diagnosed with an IRD themselves or their children, and who were registered members of the national support group, Retina SA. These participants had undergone pre- and post gene testing genetic counselling. The cohort of participants resided in Cape Town, Johannesburg and Pretoria.

2.3. Recruitment and Study Cohort

In adherence to the confidentiality policies of Retina SA, the researcher did not approach it's members directly. In consultation with Retina SA, an advert was therefore run in the regular newsletter (Figure 2.1) calling upon members to register an interest, to participate in the study. The researcher had no previous contact with participants. This course of action also ensured that participants were recruited without any form of coercion. Retina SA then emailed the names and contact details of those members who responded to the advert and who verbally consented to them, for the organisation to release their details to the researcher. On receiving the names and contact details of respondents, the respondents were contacted early in September 2016, via telephone or email to confirm their continued interest. At this stage the research study was described and the voluntary nature of their participation reiterated and emphasised.

Purposeful sampling (Patton. 2002) of participants was therefore used which is a qualitative research technique to obtain a group of individuals which met the inclusion criteria for this study. Purposefully sampling individuals who were members of Retina SA also provided participants who would produce information-rich data applicable for the research question of this study. The inclusion criteria for recruitment into the study were as follows: Individuals who were diagnosed with an IRD, who received molecular diagnostic confirmation during a genetic counselling consultation, and who were registered members of Retina SA. Participants were to all be older than 18 years of age and be fluent in english.

Retina SA Newsletter advert

We need 20 volunteers who have had genetic counselling, a gene test and results delivered to participate in a survey. This will form part of a master's degree for Rene Goliath, who was for many years a research scientist at UCT and is now doing a genetic counselling degree. Reply to this email and give us your name and cell or contact details, if you interested in helping her. Rene will contact you to arrange an interview at a time and place that suits you. The survey is entirely anonymous and no names will be published.

Figure 2.1. Retina SA advert calling for its members to register an interest in participating in the study. The advert was copied directly from the electronic newsletter in order to illustrate the large font used to accommodate the readership.

2.4. Research Setting

Each participant granted verbal permission telephonically, for interviews to be conducted at a private location (Table 1) (field method). Interviews were arranged away from other family members and associates. The interviews were conducted within the area of residence or employment of each participant. This was to adhere to the criteria for privacy with few or no distractions during the interview process so that each participant felt comfortable and uninhibited to express their experiences, views,

feelings and emotions freely. Conducting the interviews privately, away from family members who may be affected or at-risk could also promote freedom of expression without the risk of being heard and possibly hurting, confusing or offending a loved one in the process. Where interviews were conducted at the workplace, private board rooms were pre-arranged by participants. When conducted at home, the time of the interview was set for when family members were not home. The one interview conducted at a coffee shop close to her home, was during a quiet time of the day and in a “private lounge” section of the establishment. Besides private locations, interviews were conducted at a time which suited and was convenient for the participants to attend without influence by the researcher. To further promote convenience, the researcher used her own transport to travel to the participants’ chosen location and at the set times.

2.5. Data Collection

In order to explore the experiences and perspectives of each participant, it was a conscious goal to collect as much data as possible for later analysis. The interviews were therefore designed to generate responses concerning participants’ perspectives concerning ideas, opinions, and experiences, particularly but not exclusively in the areas of genetic education, decision making (choices) and adaptation. The face-to-face interviews were conducted to obtain the raw data. Guidelines for preparation for a qualitative research interview, as outlined by McNamara (2009) was followed. After initial greetings and introductions and that the use of the English language was acceptable, the purpose of the interview was explained in detail to ensure that the participant understood the study and the reason for their participation. The first two sections of the Information and Consent form (Appendix A); “Brief overview of the study” and “Participant information” was used to guide this discussion so that all the details were covered. This initial discussion was important firstly, because participants were partially sighted and while reading the information sheets were possible, it would have been too time consuming, and secondly, to establish a rapport and working alliance with the participant in an attempt to create a cooperative relationship with the participant. The terms of confidentiality and consent for participation in the study was emphasised which included permission for the interview to be audio-recorded and for the researcher to make notes during the interview as well (Appendix A). Free text notes facilitated the opportunity to write down any observations made in terms of non-verbal behaviour and cues. For example, if

the participant became nervous at any stage, or if the verbal and nonverbal communication did not coincide. The reason for the recordings were explained to be important for the researcher to retain and have on record the exact substance and essence of responses; much of which would be lost if left to rely on the recall memory of the researcher alone. It was outlined who would get access to their responses (researcher and supervisors) and very briefly how their answers will be analysed. The significance to reduce bias and interpretation of the data was explained and hence the importance of the analysis of their own comments and the recording of verbatim quotes where necessary. The format, length (between 40 minutes to 1 hour) and type of interview (semi-structured) was explained and each participant was informed of the researcher's contact details in the event they would want to make contact at a later stage or to withdraw from the study. The contact details of the research supervisor and Human Research Ethics Committee was also provided and indicated to be at the end of Appendix A. Finally, written consent was obtained for each participant, in the third section, "Consent For Study Participation" of Appendix A. As participants were all partially sighted, the place on the form where the signature was required, was pointed out to them. This signed, "Consent For Study Participation" section was photographed on the researchers iPhone, and later saved onto a password protected computer to maintain good study records, and deleted from the iPhone. Each participant received and retained the entire Information and Consent form (Appendix A) for their own records. Before the interview started, the participant was given the opportunity to ask any questions about the study or the process.

The open-ended question guide designed for this study was used to guide responses (Appendix B) to explore, but not exclusively, the psychosocial areas of interest in this study of genetic education, decision making and adaptation. Closed-ended questions were used to obtain socio-demographic information about participants such as age, education etc. Prior to the start of this study, the open-ended questions (and project proposal) were reviewed by two independent supervisors, Dr Tina-Marié Wessels and Professor Jacque Greenberg, the reviewers of the University of Cape Town's Department of Pathology Faculty of Health Sciences Research Committee (DRC) chaired by Professor Richard Naidoo, and finally by the University of Cape Town's Human Research Ethics Committee chaired by Professor Marc Blockman, for approval (HREC REF: 422/2016).

Two test interviews were conducted to assess the process, the appropriateness and scope of the questions in addressing the research question. These test interviews were audio-recorded and on review, where necessary and appropriate, questions were merged, reworded and re-ordered into groups dealing with the major themes of this study. These adjustments were also influenced based on the experience itself and feedback received.

Interviews were conducted by the researcher and followed the guidelines and recommendations of McNamara (2009). Each question was explored individually and comprehensively in order to extract as much data as possible from participants' responses. The carefully worded questions were designed to be as neutral and unbiased as possible in that emotional reactions to responses were avoided, according to Patton, who suggests to act as if "you've heard it all before. Opportunities were allowed for probing questions if more information was deemed necessary. Furthermore, follow-up prompts were used to encourage further exploration of responses, using minimum encouragers like "uh huh"s, ok, good, a smile or a nod of the head, in appreciation of responses. This acknowledgement from the researcher can improve the quality and quantity of information the participant is willing to share. Encouraging gestures also helped the researcher to remain focused on the interviewee and to be present at all times. As McNamara suggests, the researcher remained aware of her own manner and disposition during the interviews so as not to influence responses or to inadvertently convey her own opinions. A professionally balanced approach was adopted; not too formal or too familiar.

2.6. Data Analysis

Immediately after the interviews, the recorded raw data collected, was saved according to a file name system based on its number within the sequence of interviews (Interview 1 to 10). The interview audio-recorded files were transcribed verbatim shortly afterwards and saved by file name, Transcript 1 to 10, so that file Interview 1, corresponded to Transcript 1 and so on. All free text notes were filed and labelled with a similar number system, Interview 1 to 10 in order to link each interview, transcript and set of free text notes. All personally identifiable information was removed to maintain anonymity, confidentiality and bias.

The anonymised transcripts were then used to conduct a thematic analysis (Braun and Clarke. 2006). The analysis of interview data were a process which aimed to "make sense" of the information collected from each interview. The first step involved coding the data

(DeCuir-Gunby et al. 2011). This process of coding is an integral part of the interview data analysis process. Codes are tags or labels allocated to units of raw data which convey its meaning be it descriptive or inferential information (Miles and Huberman. 1994). Based on this, in this study it was decided to focus on the level of meaning.

Qualitative researchers often discuss the use of a codebook (Fereday and Muir-Cochrane. 2006). De Cuir-Gunby et al. (2011), however, suggests that creating a codebook should be a team effort as it is complex and tedious, and involves various components, and can therefore become an overwhelming challenge if undertaken by one person. Creating a codebook was therefore not undertaken in this study due to time restraints and the small team involved in the study (1 researcher and supervisors). Although a codebook as described by Fereday was not developed, codes were developed through a repetitive process and the definitions assigned to each were continually revised as the data became more clear. Code definitions were written in simple, straightforward language. The study therefore followed the principles of Fereday's code development process.

Codes can be theory-driven, data-driven or structural (derived from research goals). Theory driven codes are developed a priori from existing theory or concepts whereas data-driven codes are generated directly from the raw data. Codes can also be guided by the project's research goals and questions (structural). However, most codes are theory- or data-driven (Ryan and Bernard. 2003). As the development of theory-driven codes usually involves the constant revisiting of theory, and whereas data-driven and structural codes necessitate repeated examination of the raw data; in this study, only data-driven codes were created based on meaning. Applying codes to raw data is a means to explore how or if the data supports the research question. Coding is, in essence, a circular process in that the data can be scrutinised and re-scrutinised as new insights are learnt during the analysis of each interview. See Figure 2.2 for an illustration of the coding process.

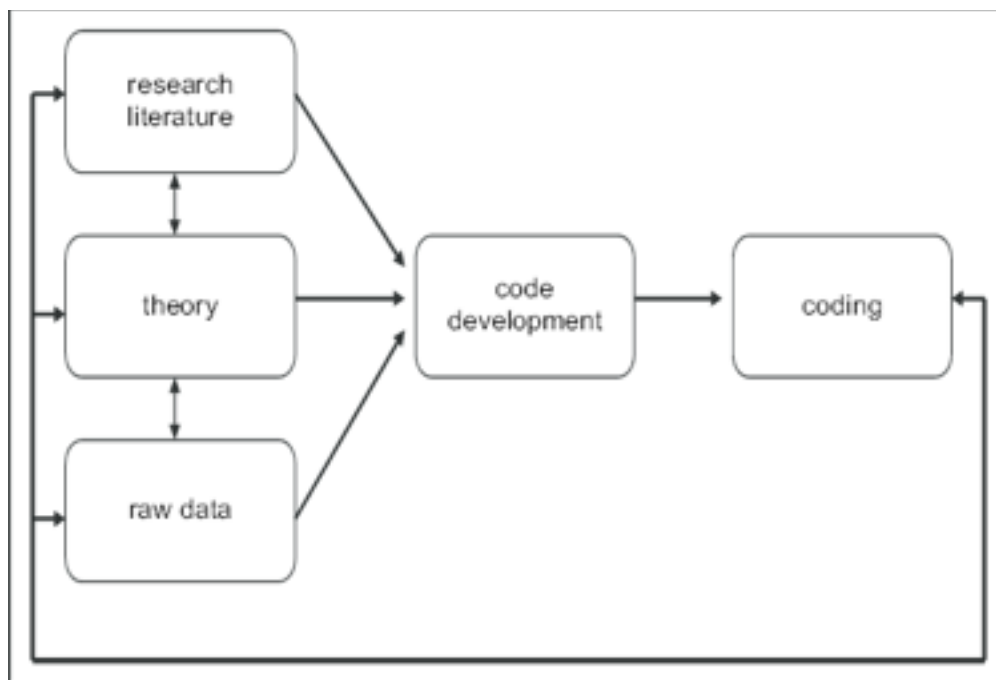


Figure 2.2. Circular process of coding (taken from DeCuir-Gunby et al. 2011).

To ensure that all labels were relevant, the codes were assigned to portions of data, such as expressions, statements, sentences, or paragraphs that were tied or associated to a specific context (Miles and Huberman. 1994). The final step used in developing data-driven codes was to determine the utility/reliability of the codes. This involved coding individually as well as with a supervisor. In addition, we discussed examples and non-examples of the codes. The focus of this triangulation effort was on both the number of codes (what) and the effectiveness of the codes (how).

The definitions of codes were recorded and compared across interviews for consistency of meaning. The information gained through this comparative process was then used to establish a way to capture their themes.

The themes extracted were patterns within the coded data related to the research question focus as well as any other themes that emerged. This thematic analysis involved grouping of the codes generated, into potential themes and examined each in order to substantiate the meaning, inferences and themes which emerged. The themes were defined, named and analysed. A theme is therefore an outcome of coding, categorisation, and analytic

reflection, it is a phrase or sentence describing more subtle and tacit processes (Saldaña. 2009). As an example, BEING ALONE was a code identified in this study, and SOCIAL WITHDRAWAL was a theme.

After conducting and analysing 10 interviews, it was decided to not engage in recruiting any further participants into the study because the researcher noted the recurrence of themes in data collected from the 10 participants' interview transcripts. It was therefore decided that saturation was reached particularly for the major preconceived themes described in this research. This decision was in keeping with qualitative based investigations which stipulate that the study cohort comprises about 10-15 participants or until data saturation is reached. Saturation is defined as the point at which the collection of new data will not introduce any new or relevant information to the study (Glaser & Strauss. 1967). The nature of qualitative research lends itself to produce big data and a large cohort is likely to become repetitive and therefore redundant. At this point, further data collections will not add any further value to the research question. A small sample size in qualitative research studies is therefore advised, in order to obtain relevant, in-depth and information-rich data (Marshall. 1996).

2.7. Ethical Considerations

The general principles and ethical considerations in qualitative research investigations within health care research was applied in the design of this study (Choo et al. 2015).

Institutional Approval

Ethical approval for this research study was granted by the University of Cape Town's Human Research Ethics (HREC) committee (HREC/REF 422/2016, Appendix C)

Consent

As explained, verbal consent to Retina SA was initially obtained from interested participants (responded to newsletter notice), to release their names and contact details to the researcher to contact them directly. On telephonic and email contact with each participant, a brief overview of the study was described. Dates were then set to meet for the interviews. Prior to the start of the interviews, formal written consent was taken (Appendix A) by the researcher, for participation in the study. This process of consent

involved the participants being guided through the information and consent form by the researcher. The voluntary nature of participation was emphasised and that if they wished, they could withdraw from the study at any time. The reason for the audio recordings was explained and contact details of the researcher, supervisors and HREC chair provided. Only once the participants understood the terms and purpose of the study, were signatures taken. The A signed copy of the form remained with the participant for further perusal at a later stage.

Confidentiality

Confidentiality of the views expressed by study participants was utmost importance and to this end, participants' names were blinded at the transcription and analysis stage of the study. Each participant was assigned a case number i.e. case 1 to 10. Once transcribed, audio recordings, transcriptions as well as the pictures of the signed consent forms of each interview were locked away on a password protected computer. Field notes were stored in private personal storage areas. Access was only granted to the researcher's supervisors. All audio recordings, transcribed documents and notes were destroyed at the end of the project. These precautions regarding confidentiality were outlined to each participant and they were ensured that no information would be shared with anyone except as outlined here, even family members. This is also why description of participants in this study does not contain personal details as a means to prevent them from being identified.

Risks

The risk of any emotional or personal discomfort to participants was avoided as the researcher was acutely aware and conscious that she presented herself in a sensitive, empathetic and non-judgemental manner towards the participants. In addition, culturally sensitive language was used. Participants were informed that they would be allowed to withdraw from the interview and study at any time in the event that they felt uncomfortable or compromised in any way. In the event that the researcher felt that the participant was in need of a counselling session, referral to a qualified genetic counsellor was arranged.

The ethical aspects of dealing with the stigma participants may have had due to their disability was considered. The word "stigma" has its roots in the Greek language and means a "mark" A stigma or a mark often inhibits communication between abled and

disabled people. It would not be easy for the disabled to say “I have unique needs that require consideration”, for example to take the arm of a visually impaired person or not, in leading the way (Behnke. 2009). The researcher was always aware of her behaviour so as not to disrespect the participant and risk compromising his/her dignity. Another example is that it was not assumed that the participant could not sign the consent form him/herself. Consideration and forethought about how the visually impaired person would prefer to be treated was thus undertaken and this enhanced the productivity of the interview session.

2.8. Benefits

The potential outcomes and impact of the study, was deemed to supersede the potential risks. The therapeutic benefit of “telling their story” during the interview process cannot be underestimated. Furthermore the data which emerged from this study brings into focus and highlight the patient and family benefits of a co-ordinated partnership between genetic counsellors and patient support services. The findings of this study will also influence patient care through reflection of actual patients’ views (de Salis et al. 2008). The data affirms approaches to patient care already in place.

Chapter 3 RESEARCH FINDINGS AND DISCUSSION

The purpose of this study was to examine patients with IRD perceptions and experiences with VI. The research question that informed this study was: how have IRD patients' experiences with the combined input of Retina SA and genetic counselling influenced their education, decision making and adaptation, with their genetic diagnosis?

Twelve Retina SA members responded to the Retina SA advert, 2 were lost to follow up and 10 confirmed their continued interest once contacted by the researcher. Table 3.1 outlines the demographics of the participants in this study. The 10 respondents each agreed to meet for an interview and a date, time and venue of their individual choosing was scheduled during October and November 2016, except for one interview which was conducted in May 2017. Of the 10 participants interviewed, 8 were affected with an IRD, and two were unaffected carrier mothers of affected children (cases 4 and 8).

| Case # | Diagnosis | Preferred meeting place | No. of children | Age range | Gender |
|--------|--------------|-------------------------|-----------------|-----------|--------|
| 1 | RP | home | 2 | 20-30 | female |
| 2 | STGD | workplace | 2 | 30-40 | female |
| 3 | STGD | workplace | 2 | 40-50 | female |
| 4 | LCA carrier | coffee shop | 2 | 30-40 | female |
| 5 | STGD | home | 0 | 20-30 | male |
| 6 | STGD | workplace | 3 | 50-60 | male |
| 7 | STGD | home | 2 | 40-50 | female |
| 8 | STGD carrier | workplace | 2 | 30-40 | female |
| 9 | STGD | home | 3 | 60-70 | male |
| 10 | RP | workplace | 1 | 30-40 | male |

Table 3.1. Participants' Demographics

Four males and 6 females participated in the study and each met the inclusion criteria outlined for the study. Seven participants were affected with STGD, 1 was a carrier mother whose children had LCA, 1 was a carrier mother whose children had STGD and 1 was affected with RP. (KEY: STGD: stargardts disease, LCA: Lever's congenital amaurosis, RP: retinitis pigments).

Seven participants were employed on a full-time basis, 1 was retired and 2 were unemployed. Five participants were English speaking Caucasians, 2 Afrikaans speaking Caucasians, 1 was of mixed ancestry, 1 of Indian and 1 was of Portuguese descent. All participants were fluent in the English language and could be classified in the middle to upper economic category. All participants had completed school and at least attempted to enter and pursue tertiary education. All participants were married and in stable relationships except one who was divorced but still living with her estranged husband. All participants were located within three major metropolitan areas of South Africa; 6 in Pretoria, 2 in Johannesburg and 2 in Cape Town.

During in-depth interviews, study participants described their experiences from the time of onset of symptoms to the present. They also discussed their personal responses to losing their vision and the accompanying emotional impact. Participants reported a range of subjects which mark their experiences. Analysis of the raw data collected in this study uncovered four broad themes. Within these four themes a number of related sub-themes emerged. Some of these sub-themes lent themselves to be classified into categories which capture the meaning of the sub-themes (Figure 3.1). While the themes and sub-themes are reported as being distinct, it must be noted that considerable overlap exists. Where participants' responses addressed more than one theme, the ideas based on meaning were placed where they belonged most logically.

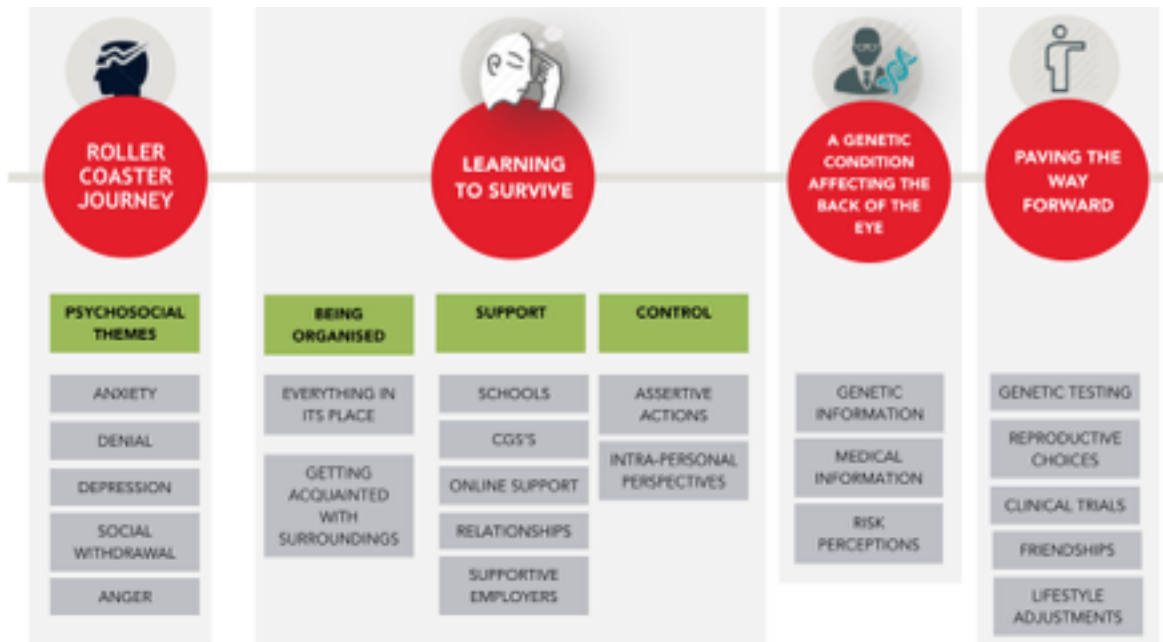


Figure 3.1. Illustration of the main themes (red circles), sub-themes (grey blocks), and categories (green blocks)

3.1. Roller Coaster Journey

The messages from the raw data revealed a “roller coaster” journey that most participants experienced. All participants started to experience disease symptoms very early during their junior school years and only started to adjust as adults. One participant spoke about learning to adjust to her condition for the first time as a young adult when she began her working career. Another said she began to understand what was happening to her during her late senior school years.

“It’s difficult, the only time I actually learnt to cope with it was when I started to work” (affected female, STGD).

“I only started to understand it in late high school, so for the most part of my condition I didn’t understand” (affected male, STGD).

Along this “roller coaster” survival journey, a number of psychosocial themes emerged from the data that participants adopted as internal survival strategies. Close analysis of these psychopathological responses to vision loss, resulted in the lumping of related

responses into broader themes including anxiety, depression, denial, social withdrawal and anger (Figure 3.2). It was interesting to note that these responses did not present in a linear fashion but instead in an almost chaotic manner, except for the anxiety, and hence the roller coaster type of emotional journey.

All participants reported a state of anxiety, particularly early on in their journey with vision loss. The sub-themes, which were lumped into this theme of anxiety encompassed “confusion,” “uncertainty,” “fear,” “shock” and “low vision specialists” (Figure 3.2). Participants gave feedback on feelings of being scared, confused and described that stage as a time when they lived in fear.

“Very scared and very confused...I have to say that I lived my life in fear” (affected male, STGD).

Participants related feelings of shock as they noted dramatic changes in the scope of their vision loss.

“After 3 months of summer holiday, I couldn’t read anything on the board...I couldn’t read my books, and then er that was a huge shock” (affected female, STGD).

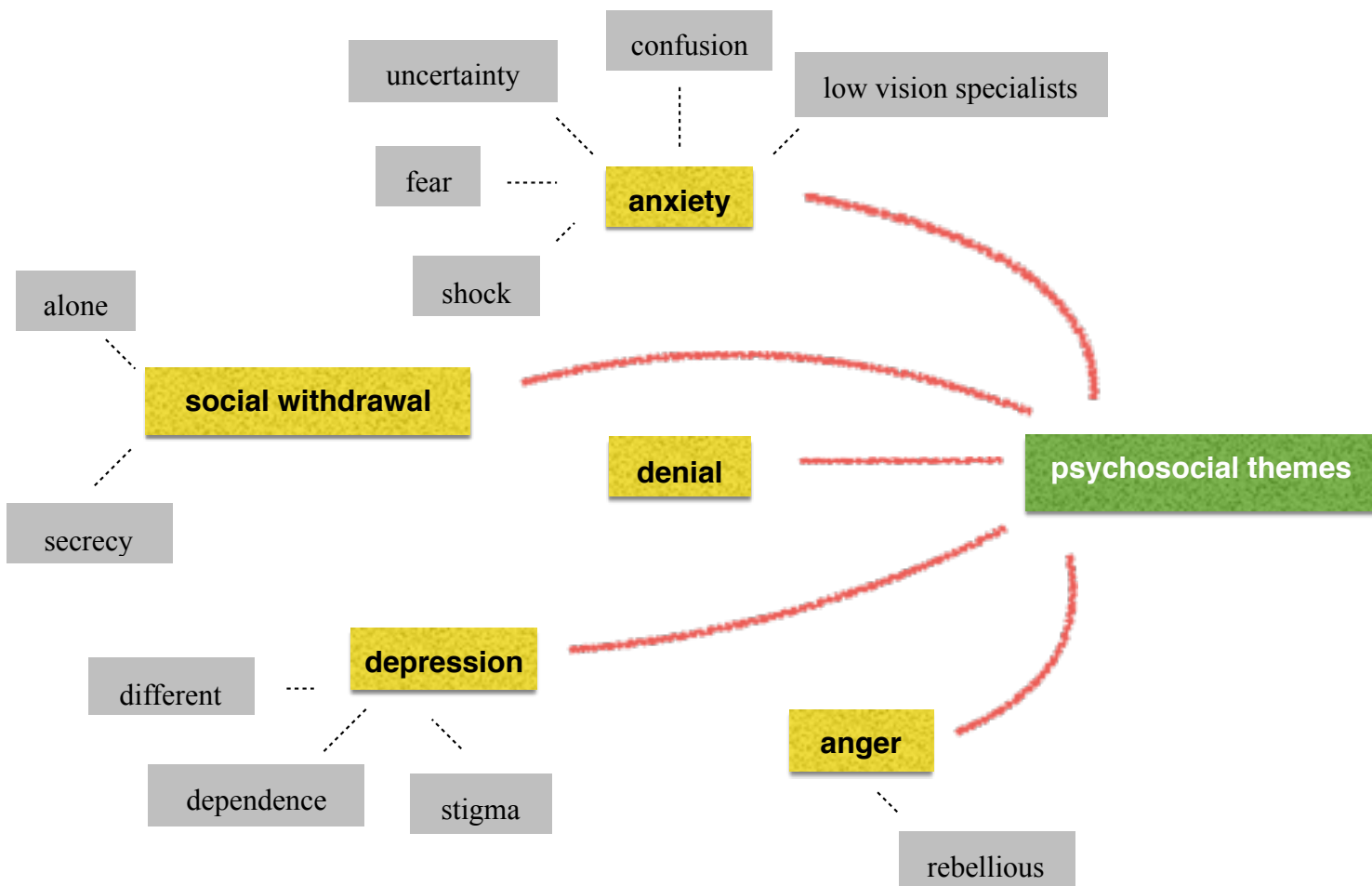


Figure 3.2. The roller coaster journey of psychosocial themes (yellow blocks) and sub-themes (grey

Feelings of shock were also said to be associated with the time when they were told that their sight would continue to deteriorate, not only for themselves but for their families as well.

“But for me it was just a bombshell” (affected female, STGD).

“As a family you get a shock and you don’t know how to deal with it” (affected female, RP).

Participants explained that much of these feelings were as a result of not knowing what was happening to them. They described that stage as a time of uncertainty and spoke about low vision specialists not assisting and supporting them.

“Nobody really explained, I think when being diagnosed they should actually explain to people properly that there is no cure, what’s in the pipeline for cures and stuff like that because people get their hopes up you know” (affected female, RP).

All participants therefore expressed varying degrees of dissatisfaction toward the health care providers specialising in the low vision area of expertise. Participants presumed that they would get the necessary support and guidance from these specialists. Instead they found these specialists were not abreast of up-to-date developments and basic information on IRD. As a result, participants were left to grapple with losing their vision, with little explanation and reassurance or comfort. Many therefore attributed their feelings of uncertainty, confusion, stigma, shock and fear that most experienced during the first years of their journey with vision loss to the lack of support and guidance from these specialists. Participants spoke about the lack of support and guidance from specialists in terms of outlining to them why they were experiencing vision loss or what the prognosis was.

“Yes because you don’t know what its all about because no one explained to me what RP was, they just said you’ve got some sort of a disease of the eyes” (affected female, RP).

As a result for many years from the onset of disease symptoms, participants were confused about what they were experiencing.

“For the most part of my condition I didn’t understand.... knowing would have made it easier” (affected male, STGD).

The views expressed by participants concerning the lack of support they received from specialists and the impact this had on their adaptation process, is supported by findings outlined by organisations which support people with vision loss (Royal National Institute of Blind People). The lack of support from specialists, influences how well the VI does at school as well as later in life. To this end, The National Sensory Impairment Partnership in the United Kingdom published a document on 30th June 2017 which outlines a framework to assist specialists and support services to align themselves with the management and care of the VI (natsip.org.uk).

It must be reiterated at this point that all participants were very young when they started to lose their sight, and this age factor compounded the trauma they experienced. One

participant expressed the negative feelings generally associated with that young stage of life and that having some clarity would have helped.

“In a time when you are very negative about life, knowing would have made it easier” (affected male, STGD).

Participants related that feelings of anxiety were compounded by parents and Doctors not believing them to have a problem seeing, leaving them to deal with the experience on their own, as young children. One participant told how after a school nurse said, “something was wrong,” that she was sent to an optometrist who told her parents that nothing was wrong and that she was seeking attention.

“He said he can’t see anything wrong and that I’m doing it for attention” (affected female, STGD).

This theme of anxiety is not uncommon and is therefore a well documented emotional response to vision loss (Adams and Perlman. 1970).

A few participants went through a stage where they did not want to accept that they were losing their vision. One participant verbalised having feelings of denial and an unwillingness to accept her condition which inhibited her from dealing with the practical realities associated vision loss.

“The problem was I couldn’t accept it” (affected female, STGD).

A carrier mother was also perceived to display a level of denial by her view that her children remain in a mainstream school. She chose to enrol them in a mainstream school because of their intellectual abilities and did not factor their physical disability into her decision. She suggested that schooling was about brain function and not their physical disabilities.

“They are in a mainstream/normal school because there is nothing wrong with her brain..she does well in school” (carrier female, LCA).

Adams and Perlman (1970) also report denial to be a known response to vision loss. Other studies also show that the VI will grieve their loss of vision and in so doing, they may experience the known phases of grief, which include denial, anger, depression, bargaining and acceptance (Kübler-Ross. 1969). The finding of denial as one of the stages of adaptation in the current study also supports the study by Stevelink et al.(2015) which

reports that people in denial of the consequences of vision loss attempt to continue to do things as they were previously accustomed to do which in turn results in feelings of frustration and irritation. In the current study this was particularly evident in those participants who continued in mainstream schools in spite of their deteriorating vision.

Not coming to terms with the reality of VI was a strong precursor to feelings of depression which was experienced by participants most intensely at the onset of vision loss. Depression featured prominently, evidently not only because of the deterioration of their sight but also due to the confounding psychopathological factors (Figure 3.3) they experienced at this stage of their “roller coaster” journey.

“I was very depressed” (affected female, STGD).

Reiterative analysis of the data and codes generated, revealed a few contributing factors related to the theme of depression. These included perceptions of being “different” and the “stigma” which accompanies this consciousness, the recognition of being constantly “dependent” on others and thus being a “burden” to loved ones (Figure 3.3). While it did not ever go away completely, over time, as they learnt how to survive their disorder, they found themselves fluctuating in and out of various states of depression. The triggers varied from an inability to go to the shop without assistance, to receiving test results and having moments of self pity. For example participants spoke about the frustration and emotional impact of not being able to drive.

“I get depressed but not for long” (about needing help to go out) (affected female, STGD).

“And then I was depressed again....” (after results were delivered) (affected female, RP).

“It’s much better now ...but sometimes I still cry but I learnt to deal with it.” (about affected children) (carrier female, LCA).

“I do feel sad, I always question ...why did this happen to me?” (on self reflection) (affected female, STGD).

“I never want to become that big a burden on them not being able to help myself” (being a burden) (affected female, RP).

“Those things do bother me ...depending on somebody.” (feeling dependent) (affected female, STGD).

Participants referred to the early stages as a period of fluctuating emotions until they could get some clarity and direction which was achieved through a definitive genetic diagnosis.

“So it’s been a big roller coaster until we found out exactly what she has” (carrier female, LCA).

“In that time I broke down alot, because how do you explain it? I didn’t know” (carrier female, STGD).

One participant related feeling depressed at the time she changed from a mainstream to a special school. At this pubertal stage in her life when little girls want to fit in to normal society, and when being different is not acceptable, it was difficult for her to deal with this change and feeling of being different.

“At the age of 11 ...I was very depressed...for me it was a big change to go from a normal school to a special school” (affected female, STGD).

Feelings of being different in turn resulted in participants feeling stigmatised. Participants’ feelings of being labeled as different were often due to their own intra-personal turmoil and self image. They spoke about feelings of inadequacies and therefore lacking the confidence to be a part of normal society.

“I knew we were different ...and for a long time I had a complex about it ...felt I was a lesser member of society” (affected male, STGD).

The stigma participants felt, however, was also influenced by insensitive actions of sighted people towards them. They related that in general, people treated them differently once they became aware of their VI. They said that sighted people made them feel like they had a contagious disease. This was very depressing.

“People treat you different” (affected female, STGD).

“The stigma of them still being labelled as disabled ...still affects them” (carrier female, STGD).

“To me, I’ve experienced it ...I’ll meet a guy and as soon as you tell them you have an eye problem, they are gone, its almost like you have this disease” (affected female, STGD).

Participants also spoke about the anxiety related to not getting clear information from specialists, contributing to their depression. They expressed having the perception that going to the Doctor is associated with treatment and a cure. However, when the Doctor could not make them better, together with an unclear reason, this predisposed them to depression.

“And its like oh no I’m going to get fixed and it’s not, and you know its quite emotional ja” (affected female, STGD).

Unaffected carrier participants’ feelings of depression also stemmed from feelings of not being able to connect with their children through the sense of sight. The absence of the value of face-to-face contact was almost tangible. A mother spoke of it in an emotionally intense moment during the interview.

“Because you know you can’t reach... your children (broke into tears) sorry!” (carrier female, LCA).

Literature on the psychology of vision loss report that depression is a common theme (Moschos. 2014). Furthermore, findings in the current study substantiates published reports that depression is usually experienced for varying durations and levels of severity. The unanimous response by participants of experiencing a depressed mood also echoes the report by Oehler-Giarratana and Fitzgerald (1980) that the psychopathological reaction to vision loss was more common and severe for those with partial sight loss when compared to patients who experience a total loss of vision. The report also outlines reactions of anger and hostility, which this current study also illustrates.

Wanting to be “alone” is a psychosocial theme that arose for most participants during the early stages of their vision loss experience largely because of feelings of being “different”, “stigmatisation” and “depression” as discussed earlier. Wanting to be “alone” was therefore a natural precursor to decreased socialisation. Another related sub-theme and contributor that emerged here was “secrecy” where participants reported not wanting to tell others about their condition (Figure 3.3). One participant said that she cut herself off completely, often even from family members.

“I closed up completely, I didn’t want to mix with other people, even with family” (affected female, STGD).

Participants outlined that they withdrew socially because of the differences they recognised within themselves compared to sighted people. They preferred to be among other partially sighted people.

“I was always withdrawn, I didn’t want to mix with other people ...never wanted to play with other kids if I’m not in my own environment with other kids thats the same as me” (affected female, RP).

On being the only affected person in the family, a participant spoke about feelings of added isolation from within the family. Family members are to be similar, the same, and this bonds them together but she felt different and therefore had feelings of not belonging, which led to further withdrawal.

“My parents came from big families and nothing (no family history of IRD), so that was the worst I thinkI felt isolated” (affected female, RP).

So for the most part participants felt that they wanted to become invisible. They wished it was possible to mingle in society invisibly so people would not interact with them, they spoke of wanting to avoid any form of engagement.

“Going to school I really made myself invisible, in class I would duck and pray that the teacher won’t ask me questions...or not see me” (affected female, STGD).

They said what also contributed to wanting to be alone was feelings of embarrassment and not being comfortable telling people about their eye condition.

”For so long in my life I didn’t let people know” (affected female, STGD).

One participant said that these insecurities were further fuelled by her parent’s own need for privacy and secrecy.

"I guess because my mom was like you don’t tell any body....about your condition” (affected female, STGD).

So to avoid talking about it, they reasoned that it would be better to be alone. Participants spoke about avoiding the mental trauma associated with mustering enough confidence to talk about their condition. They said the unusual manner in which the VI look at people would invariably invoke questions and stares. This situation was avoided.

“Stressful ...so I’d rather avoid situations or functions when I have to look at people directly” (affected female, STGD).

Withdrawing socially was also motivated by perceptions that sighted people would not be interested in them and that they would be a problem.

“It was difficult with friends because I didn’t have anybody ... well because you feel like people are not interested because you are going to be a burden in their lives” (affected female, STGD).

The finding of decreased socialisation within this study group, supports yet slightly contradicts the report by Ash and colleagues (1978). The report describes being young, a good financial status and a reasonably high sociocultural level to be protective factors against psychopathological reactions in partially sighted individuals because of good social relationships and the avoidance of isolation (a risk factor for depression). The current study group could also be described to be at a moderate-to-high sociocultural level. However, while many did overcome their psychopathological reactions and in the long term reported improved socialisation, early on in their disease progression social withdrawal and the sub-themes listed were prominent. In general though, social withdrawal among the visually impaired, is a prominent theme. Many studies address this reaction and intervention programmes have been established (Sacks. 1992).

Three participants reported periods of anger as a result of their vision loss. However, while others did not explicitly verbalise their feelings of anger, the theme did emerge in the discourse of interviews. The related topic of rebelliousness also emerged. Participants spoke about feelings of anger when people stared at them or commented about their eyes.

“In public if someone said anything about my eyes or how you look or something, I would become very angry, very upset” (affected female, STGD).

Participants spoke about releasing their anger in a physical way.

“I kicked alot of cupboards ...I was very angry at one stage” (affected male, STGD).

As teenagers not understanding the changes they were experiencing, feelings of anger predisposed them to rebellious behaviour. Participants related feeling that life was unfair.

“At one stage I was very rebellious because I thought it was unfair ...why do I have this?” (affected male, STGD).

Participants reported feeling angry too, that test results took so long. They felt that they needed to be updated on the progress.

“Test results took more than a year (a bit angry)” (affected female, STGD).

“I need more feedback to know if it's working ...what is the real results? you want tangible stuff” (anger at prolonged wait for results of screening tests) (affected male, STGD).

Anger is one of the reported stages of grief (Kübler-Ross. 1969), as outlined earlier and a common reaction to vision loss (Cholden. 1958).

Winton (1990), described the various responses people exhibit to a crisis or loss, as possible internal coping strategies which influences how they learn to cope. The findings in the current study which show participants displaying a range of psychopathological responses involving both adaptive and maladaptive behaviour in response to losing their vision, supports Winton's supposition. Each response to vision loss described was a means to deal with the trauma of vision loss and somehow was a stepping stone towards them learning how to navigate and survive a world with limited vision.

3.2. Learning How To Survive

Over time, many participants found that they learned a number of strategies which gave them the confidence that survival was possible. Creating a survival toolkit was important as a means to move forward in life as a person with visual impairment. Participants spoke about recognising the importance of survival strategies as a means to not getting "left behind" in the race of life.

"You just have to (adapt)..otherwise you get left behind or you just get really, really stressed out" (affected female, STGD).

The toolkit comprised several broad themes which emerged from the data, as outlined in Figure 3.3. Each of these are further unpacked.

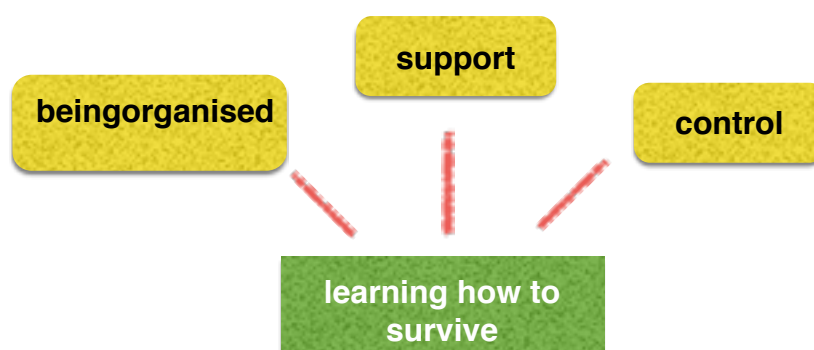


Figure 3.3.Illustration of the theme learning how to survive and its sub-themes

3.2.1. Being Organised

When participants became older and started learning how to survive the world of VI, they gave an account of how they made practical adjustments in overcoming practical challenges. They related their methods of organisation. For example the importance of

having an organised lifestyle was highlighted to hold considerable significance. Always having things in its place without clutter and chaos was related to be important in preventing accidents such as tripping and falling.

”You know you, I have a very organised house, you see there is not a lot of clutter around so I don’t fall over things” (affected female, RP).

Participants spoke particularly about the kitchen which needs to be arranged in such a manner that moving around the space and locating items could be done instinctively. This high level of organisation was important to avoid accidents with breaking glasses, crockery and avoiding hot surfaces which could cause physical harm to themselves.

“My kitchen is so organised that I can actually with closed eyes find stuff” (affected female, STGD).

Organising cupboards and wardrobes for ease of use was also highlighted.

“I pack their clothes according to items ...so shorts together etc. so its easier for them to go fetch the thing that they need” (carrier female, LCA).

Participants spoke about going on holiday and wanting to get to the destination before sunset so that they could get acquainted with the surroundings and get their orientation and organised before sundown.

“When we go away on holiday and whatever, its like I would wanna be at a place before nightfall so that I can get myself organised so that I can say to my daughter, “in that suitcase on the left hand corner there’s that, go get that for me “if I can’t get it myself. or if I have my torch I’ll know where to feel for so you know you organise yourself around your lifestyle. ja you get used to it.” (affected female, RP).

Being organised in the journey of learning to survive in the world of VI has been highlighted and discussed on several forums and blogs promoting awareness and the needs of the VI (www.Visionaware.org). The site reports that “being organised is a matter of survival”. The blog states that it is natural for the VI person to be organised and keep everything in its place as it reduces stress. Being organised helps the VI individual to function calmly and with the assurance that things have not been moved and a feeling of independence. The finding in the current study group is therefore in keeping with these views and reports by other VI individuals.

3.2.2. Support

The theme of support emerged from the data as a major contributing factor to participants' ability to survive. Most participants found that over time they were able to overcome the limitations of the psychopathological responses they experienced during the early stages of their vision loss, through various support mechanisms. These support systems helped them to deal with their condition more effectively. The related sub-themes that comprised the support mechanisms included schools, IRD clinical genetic service, online support group and relationships (Figure 3.4).

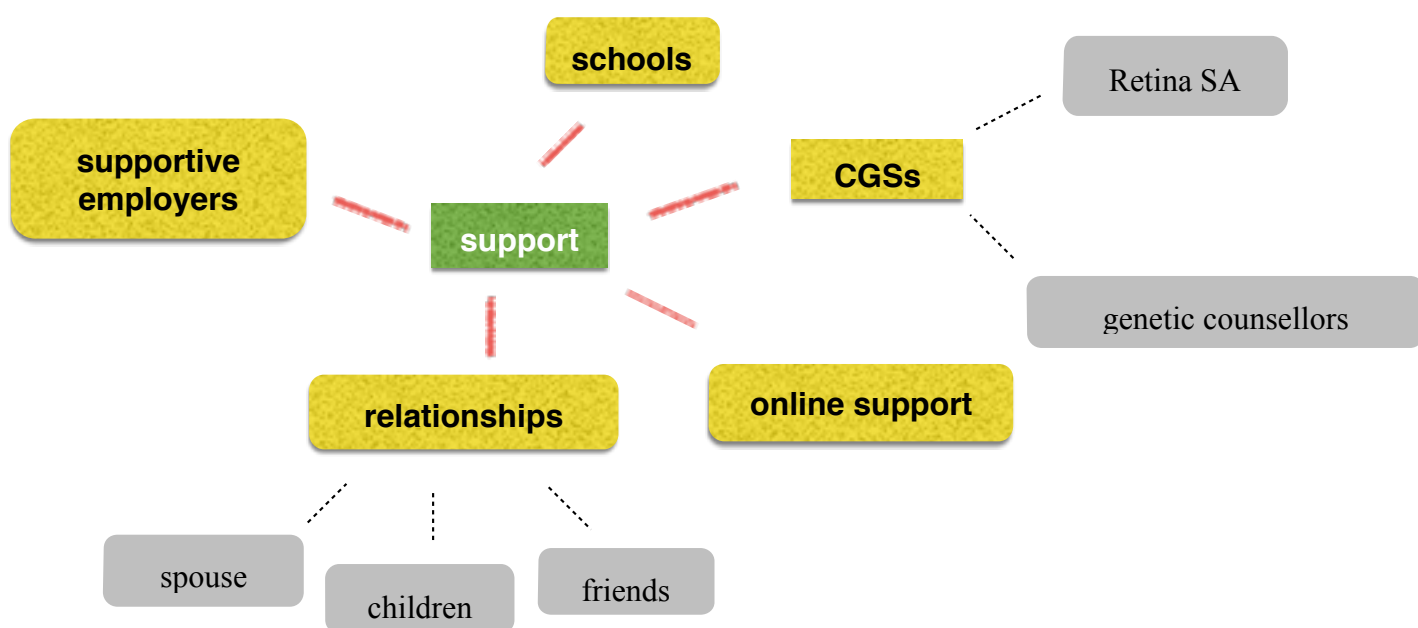


Figure 3.4. Illustration of the theme of support and sub-themes (yellow blocks)

Several participants highlighted positive views about the wonderful support they received from special schools for the blind. These participants held the view that special schools were better equipped to support the blind child with special teaching aids and assisting them with ways to deal with the disability of vision loss. Participants recounted the helpful and empowering experiences they had at special schools for the blind. One participant told how the school moulded him into the person he is.

“I’m very thankful actually, that I could go to Worcester (school for the blind)...it is a wonderful school ...all the teachers were wonderful...I think it’s strange to imagine that I would be half the person I am now if it wasn’t for them. So having being sent there was a blessing in disguise ...there is no two ways about it“ (affected male, STGD).

A participant who had attended a mainstream school reflected on how he disagreed and said that a special school would be more effective in meeting the needs of the visually impaired. He based his opinion on the difficult experience he had as a VI child, within the mainstream school system.

“I’ll do that differentlyI’ll put them in a school for the blind because they will get all the help they will need there...because my step father didn’t put me in a blind school” (affected male, STGD).

It was interesting to note, however, that the two carrier mothers with partially sighted children as well as one partially sighted participant had a different view and thought that their children and they would get better support within the mainstream school system. They were of the view that mainstream schooling would better prepare the partially sighted child to be better integrated into normal society. These participants held integration into normal society in higher regard than equipping the child with aids and meeting their specific and current practical needs. A mother said that a mainstream school was preferred because her children were not mentally challenged and because they do well at school, their physical challenges were met through other means. This mother’s view was that a mainstream school would better equip them for higher education later on in life.

“They are in a mainstream/normal school because there is nothing wrong with her brain..she does well in school, so I’m trying to keep them in mainstream schools because they need to be adaptable if they want to go for tertiary studies” (carrier female, LCA).

An affected participant with a similar opinion reported:

“For me it was a big change to go from a normal school to a special school....it was a big thing and I was never happy at that schoolI was always looking for a reason not to go to school” (affected female, STGD).

According to the American Association for the Blind, the specialised schools for the education of visually impaired students have been around since 1829 (Taylor. 2000). This report details criteria for schools to be able to provide an appropriate learning programme for the visually impaired. The report concluded that students with visual impairments are most likely to succeed in schools that provide a team approach of professionals, parents and students. Specialised schools where instruction and additional services are available to meet the unique educational needs of each visually impaired student is therefore reported to be preferable.

There are reports and views though that support the mainstream school system for the visually impaired. The view is that the mainstream school system allows for socialising with sighted people, which assists with the development of social strategies necessary for life, and that this may impact the future positively. It is noted though that within the mainstream school system that the student may feel a degree of isolation but that social skills can be developed through alternative strategies (Duquette. 2008). This study highlights though that which ever schooling system is preferred, that integration of a special curriculum aimed at the VI, is necessary and important (e.g. teaching and training in specialised aids and equipment, Braille, O&M, communication, daily living activities, etc.).

Participants also found support from the CGS for IRD in SA (Figure 3.4), useful and informative. Particularly, learning about the genetic basis of their condition through genetic testing helped them to finally gain some perspective and proved to be a turning point in their lives. Knowing the cause of their vision loss somehow brought the period of confusion and uncertainty to an end. They felt a measure of “clarity” and the means to exercise some “control” over the future.

“Diagnosed at 8 with LCA for us was a relief because now we knew what the diagnosis is and we could move on to see what we will have to deal with a relief” (carrier female, LCA).

This participant expressed some frustration though about the duration of the testing process and in fact decided to by-pass Retina SA/UCT and do their testing via a private facility. This slightly negative feedback in fact reinforced the value of the support participants receive from the various services offered by the CGS and in particular the genetic testing service.

“Retina SA said they could do the testing via Estonia but we had to wait for a bigger pool, so I took a different route, it went through the Netherlands, via the Gordon Institute and we got the results within 6 months” (carrier female, LCA).

Most participants found the exposure to global research efforts received from Retina SA, particularly the status of developing clinical trials for the various forms of IRD, to be intriguing and instilled in them a measure of “hope” for the future. So many participants responded with a positive view on the clinical trials that are underway.

“If trials come, I do want to be a part of it, I will try!! if it comes to SA” (affected female, STGD).

However, some participants expressed concern about the medium (email) used for distribution of the newsletter and being overwhelmed with the amount of information.

“I do get the Retina SA newsletters but it’s so much, I can’t read it” (affected female, STGD).

Participants, however, expressed appreciation for the general support received from Retina SA and the value of connecting with people with which they have something in common.

“I think she is the principal of Retina SA...she often gives me a lift when I need to go there and back” (affected female, STGD).

“But yes its nice to connect with other people who is are struggling with the same condition” (affected female, STGD).

A few participants, however, had an alternative view and were not comfortable meeting with the Retina SA society because they were not keen to associate with similarly affected people. They did not want to focus on various issues affecting the VI person. They just wanted to get on with life and not discuss it too much.

“I don’t like meeting people similarly affected ...I go to the AGMbut I don’t want to be bothereddon’t want to beI have no patience to listen to people nit pick on little thingsI don’t want to be part of those groups” (affected female, STGD).

Some participants highlighted the need for Retina SA to offer personal support and to create public and institutional awareness about blindness. They expressed the need for support for all types of IRDs.

“You would expect a more integrated support (from Retina SA) into the various types of disorders...I don’t want to know about RP ...because it’s not affecting me. I just don’t need the noise about other conditions” (carrier female, STGD).

“I think institutions like Retina SA should be more proactive like educating schools about how to deal with children with vision problems, and their parents and whoever else ... awareness” (carrier female, LCA).

Participants related appreciation for the support from the genetic counselling component of the CGS. Most recalled their sessions with their genetic counsellor to be informative, helpful and useful. However, their recall of the specific risks and information was limited for

some. All participants had seen a genetic counsellor twice; before and after genetic testing. They expressed that they were willing to meet with a genetic counsellor again.

“The genetic counsellor gave us more information, everything was nicely explained, this was helpful, I will feel comfortable going back to her”(the GC) (affected male, STGD).

“I was very impressed ...Jackie Jones (an alias)...I was very impressed....she clearly read up on STGD...gave various options and helped me pre- and post-testing” (affected male, STGD).

The findings of the current study pertaining to the IRD clinical genetic service in SA, reflects published reports on the challenges and guidelines for such services. The complexity of the genetic landscape of IRDs often complicates establishing a conclusive genetic diagnosis. However, technological advances have impacted gene discoveries to an extent which has increased our understanding of IRDs. The expanding knowledge of scientists in retinal research and the power of using this information to establish diagnostic interventions has crystallised the importance of providing comprehensive CGS encompassing diagnosis, therapy, rehabilitation, and counselling (Ganesh and Al-Mujaini. 2013). The report advises that new technologies and research findings be regularly integrated into CGSs. The IRD clinical genetic service in SA supports this view and have been operating on this ethos for the past 25 years (Roberts et al. 2016). Notwithstanding the approved approach to genetic service delivery, the socio-economic challenges facing the majority of the SA population, does negatively impact accessibility to these services. Retina SA has therefore implemented a strategy of part-funding genetic testing for its members.

One participant deliberated on the valued support she received from an online STGD patient support group (Figure 3.4). An online community of people who understand her specific needs and challenges made a huge difference in her life. She spoke about the personal value she derives from the interaction with people who are dealing with the same life issues. Interacting with people online who understood her needs, made her feel less isolated and this was a saving grace for her.

“I’ve also subscribed to the STGD online support group. I get alot of news and guidance from thembecause there are parents from all over the world who subscribe to that site and any question can be askedand everyone tries to help.Because I wasn’t alone, eventually I found people who I could relate to in the STGD group. They shared experiences, I think that was my saviour, that I was not alone” (carrier female, STGD).

Individuals experiencing similar disabilities, wanting to belong to a support group is commonplace. There are a number of support groups functioning around the world for various disabilities. Feelings of depression and anxiety are commonly associated with vision loss as reported in this study. Support groups provide places of safety for people to share their feelings and experiences with others facing similar challenges. Many derive benefits from interacting with people in similar situations. The peer support group exposes patients to a community of people who can fulfil these needs.

Support participants received from personal relationships (Figure 3.4), with family and friends was a major theme in this study. Each expressed their need of others and the value they receive from these individuals in learning to overcome the setbacks of their condition. They pointed out that the valued support came from special people who understood their situation and therefore were able to help in a meaningful way. Some good meaning people try to help, but not in a way that makes them (VI) feel comfortable and dignified. Participants expressed with sincere gratitude the help they received from family members.

“But as far as lifestyle goes my family is so great whenever we go anywhere, they are all so aware, they don’t make it obvious but like they know how to so that I can get..... because they know to familiarise me” (affected female, RP).

“My wife is great, I wouldn't be able to cope if it wasn't for her” (affected male, STGD).

“Family support is very important in this partially sighted world” (affected male, STGD).

Besides family members, the support of friends was highly regarded as this made them feel like a normal social being. Simple things like going to a coffee shop with a friend, was important towards promoting self esteem and confidence.

“We went for coffee, so she was a big support”(affected female, STGD).

“It’s important to have a good friend support ...people who accept you” (affected male, STGD).

The general need of support from people, was highlighted and emphasised. Participants expressed their recognition of the reality that they will always need people for help; more than sighted people do.

“you can’t ...you need people...your familyyour children your partneranything to help because you can’t” (affected female, STGD).

The findings within this study group generally reveal supportive long-standing relationships. Participants valued the support they received from these relationships and expressed their appreciation for them. Supportive relationships is a theme which has been reported among the VI (Stevelink et al. 2015). This report described VI individuals experiencing a shift in the dynamics and roles within relationships on vision loss and that only a few were reported to end.

Participants highlighted the value of having supportive employers in affording them the opportunity to earn a living and then to provide the necessary equipment and aids for the blind, to assist in performing their work responsibilities. Support and understanding from employers in this regard was invaluable in learning to survive vision loss and integrating into society as well (Figure 3.4). A participant spoke about the support he receives from understanding clients (employers) and how this made life so much easier.

“And I have good clients ...they pick me up” (affected male, STGD).

Another participant highlighted the appreciation she has for her employer who accommodates her needs around having to be available to her VI children. This helped her to face the challenge of not being able to have her children wait at school for any amount of time. She has to be on time to pick them up, this often meant leaving work early.

“I get a lot of support at work ... work must be flexible, they understand and accommodate me” (carrier female, STGD).

Participants drew attention to employers providing aids for VI people to fulfil their responsibilities effectively. They spoke about how the aids helped them overcome challenges such as not being able to read the computer screen.

“In the beginning I did have challenges at work but now I have all this equipment thats helping me” (affected female, STGD).

The benefit of supportive, understanding employers in contributing to their self-esteem was also brought into focus. A participant recounted that she was not sidelined by her employer when opportunities became available in the workplace and how much she valued this as a VI person.

“And thats what helped me (survive), is that people gave me the opportunity to do different jobs and not just sit in the same old job like your brain is dying” (affected female, STGD).

The finding of the positive impact supportive employers can have on lives of the VI supports the views of the American Foundation for the Blind. An article on the foundation website states that with modern technologies, resources, and skills, employers can provide fulfilling employment to both the blind and VI.

3.2.3. Control

Over time, as participants learnt to deal with their internal psychosocial responses to vision loss, as well as benefitting from various support mechanisms, many illustrated the importance of gaining a degree of control and “independence” in various areas of their lives. This was very important to all participants. Exercising authority over certain aspects of their lives created some balance with respect to the “dependence” (as discussed earlier), inevitable for the partially sighted person. In turn, the control gained, boosted their self-esteem, confidence and dignity. Participants said that where possible, they would do whatever it takes to achieve this some independence.

“Anything I want to do for myself that will bring me independenceI’ll do” (affected female, STGD).

The sub-themes related to control were classified into two main categories of change. These categories outlined two broad streams of change in the lives of participants. The first was through decisive assertive actions and the second stream of change was by altering their intra-personal perspectives (Figure 3.5).

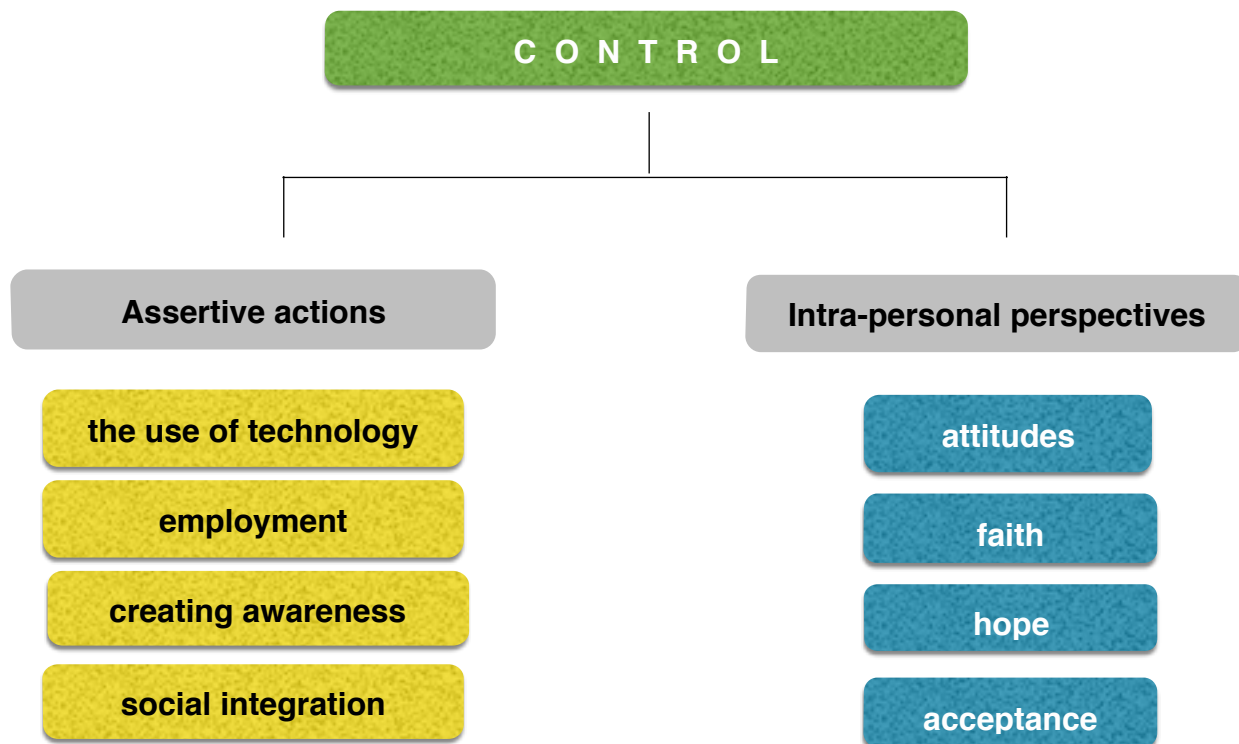


Figure 3.5. Illustration of the theme of control (green block) and its two streams of change. Assertive actions (yellow blocks), Intra-personal perspectives (blue blocks)

Assertive Actions

Participants reached a stage in their journey with VI when they had gained the confidence and self reliance to engage with a number of life altering course of actions. These actions included the use of “technology,” seeking “employment, by creating “awareness” and integrating into society,” which impacted their lives dramatically in their quest to gain some autonomy where they could (Figure 3.5).

Technological advances in modern society afforded participants the degree of independence they craved. The use of technology therefore emerged prominently with all participants and decreased their level of dependence. One participant expressed how empowering the personal computer was to her and likened it to getting her sight back.

“I use speech synthesis ...reads everything on the screen...and text recognition in a sense the computer gave my eyes back“ (affected female, STGD).

The range of Apple equipment from iPhones to cameras, had reportedly revolutionised the life of the VI. Participants explained that making use of available technologies, brought

their years of isolation to and end. They felt that they could engage with the world without feeling completely inadequate and self-conscious about what separated them.

“Now with Apple my life is much easier, with the Apple phone ... but my camera on my desk I use every single day even to chat and to use the phone. When I realised there are ways to help me...when I got my magnifying glass” (stopped isolation) (affected female, STGD).

With the ability to use computers and iPhone’s participants were able to “self educate”by researching information for themselves thereby exercising a further measure of control within their lives. Being able to research information about their disease using google, afforded participants a degree of power which was previously absent. No longer did they have to feel trapped not knowing or understanding what was happening to them.

“Researching stuff for myself, I google everything”(affected female, STGD).

These findings which highlight the value of the use of technical devices in the lives of the VI echo the views of the American Foundation for the Blind. They outline two categories of technologies for the VI: general technologies such as computers, smartphones and cell phones, GPS devices, etc. and assistive technology which includes gadgets designed to help people with vision loss such as screen readers, screen magnifiers, video magnifiers and other devices, braille watches and braille printers. The Foundation provides information necessary to learn about available technology as well as how to use the technologies to improve everyday life. Participants in this study reported the use of technologies from both categories. Furthermore, the value of technology to participants in this study in helping them to deal with various daily life situations and personal limitations as a result of poor vision is also reported by Scherer (1998). This earlier study also outlined how technologies can assist with social integrations and general quality of life, benefits which were also revealed in the current study.

Participants found that finding “employment” helped them to gain independence and to find purpose in life and therefore help in surviving a life most people associate with dependence. They realised too that they could contribute to society in some way as an employed person and this contributed significantly to their feelings of self worth. One participant told how her world perspective changed once she started working.

“It’s difficult, the only time I actually learnt to cope with it was when I started to work, because then I realised I’m in this big building and I have to share this big world with other people”(affected female, STGD).

Seeking employment also helped participants to better “integrate into society”which benefited their self-confidence and self-esteem. In talking about VI people they know who were unable to work, they tell how these individuals lead lives of hopelessness.

“Those people they just don’t have jobs now...they just give up on life”(affected female, STGD).

In contrast, participants spoke with pride about the progress and successes they’ve achieved in the workplace as a VI person.

“I’m fortunate I’m not on the switch board anymoreI’m the PA to one of the excesso I can also do a job like other people, just with the right equipment, I can do it” (affected female, STGD).

According to the American Foundation for the Blind, theVI “have proven themselves to be hard working, productive, and invaluable resources in the labor market”. Therefore in order to facilitate and promote the process of employers employing VI employees, the foundation provides a practical guide for employers. It is therefore interesting that a recent study showed that the employment rate among the VI in the United States is 37% (Bell and Mino. 2015). Not surprising when one considers that Randolph (2004) found that being disabled was a strong negative predictor of employment.

Many participants felt that “awareness” about their condition within their families and the public was important. Many felt that educational institutions and society at large exhibited a lack of awareness concerning the needs of the VI and that this ignorance negatively impacted their lives as members of society. Creating awareness gave participants some control over the extent to which the disorder was impacting their lives.

Awareness within the family circle was considered important to ensure that affected members were treated with dignity, respect and understanding, at least from loved ones. Also so that loved ones could understand their own risk status and need for possible genetic counselling. To this end, most participants discussed their condition freely with immediate and extended family members to give them a measure of control over the future.

“I have spoken to my kids, they know exactly what it's about and where it comes from” (affected female, STGD)

“We have discussed it with them like our siblings, that testing is available” (carrier female, LCA)

In a further effort to exercise control the negative effects of VI, participants included the extended family in the activities and awareness programmes run by agencies such as the League of Friends of the Blind (LOFB). These programmes gave family members a broader and more personal view of the world of the VI.

“My siblings will go with us to LOFB for a day so they can experience what Lisa is going through. It gives them an idea of what it is, even my father-in-law said when he went in there, he came out a different person, because you become more grateful for the things that you have when you see things like that.” (carrier female, LCA)

One participant took time to write a booklet detailing the genealogy of his family, describing the condition as well and its genetic basis including laboratory reports, for distribution within the family and beyond. He was passionate about the view that the condition can be better controlled through awareness. It was his view that if people know what it is, how it is caused and what testing options are available to manage the risks, that they can control how the condition affected them (affected STGD).

“As part of my legacy I wrote our family genealogy so that the message about this condition can be passed on and I distributed it among my family and to other people, because it is also a useful tool.”

Participants expressed the effects of the lack of awareness by the general public as well as educational institutions. They told how most people treat the disabled with disregard.

“Once you have a disability, people put you aside” (affected female, STGD).

They drew attention to the lack of awareness programmes to the general public but particularly educational institutions. They told how they took it upon themselves to make an effort to create awareness about the VI within the pockets of communities in which they operate. One participant spoke about her efforts in addressing the awareness issues within the school her children attended. She had also approached the Department of Education for help in taking the message of the VI to more schools, with little success.

“They’ve had incidents at the new school where children put their feet out and trip them because they can’t see. So I have addressed it with the school. Children need to be educated about people with disabilities, and it’s not even happening at schools”

“But no luck with the education department” (carrier female, LCA).

Creating public and institutional awareness was seen as a means to make a difference for the greater partially sighted community within society. A carrier mother described her efforts in creating awareness as a means to improve the lives of all VI people by collaborating with schools. She invited LOFB to mainstream schools to create awareness among the pupils (LCA carrier mother).

”Education institutions, we trying to making it better for the next person ... trying to open doors for other people, “XXX school for the blind ... not helpful ... (we) trying to make things easier for them ...like iPads etc.... verbal exams” (carrier female, LCA).

“We brought LOFB to the school to create awareness” (carrier female, LCA).

The message from participants that family, public and institutional awareness is important in addressing the inequalities faced by the VI, is outlined on the Visability website (www.visability.com.au), indicating that it is a known area of concern in society. The editors of the website indicate that awareness should correlate with effort towards providing access to facilities, services and support within the community, as well as understanding how to relate with people who are blind and VI.

Some communities have made the effort such as White Cane Awareness Days dating back to 1964 in the United States, an initiative to promote safety for the blind within the community. The Western Blind Rehabilitation Centre’s director is quoted; “By supporting an initiative that promotes safe mobility, we can help blind or VI persons gain independence.” “The use of white canes, dog eyes and public and private transportation programs ensure that travel is efficient, safe and encourages self-reliance.”

School attendance, finding employment and attending awareness programmes instilled in participants an appreciation for the value of becoming part of general society. This motivated participants to make assertive efforts to integrate into general society. So after the period of decreased socialisation and secrecy that many participants exhibited, they began to refashion their lifestyle, and set out to establish a modicum of social normalcy. Participants spoke about the stimulating feeling of meeting and interacting with people.

"Meeting people was exciting." (affected female,STGD)

They came to the realisation that they needed interaction with people and that they could benefit from other people's experiences and perspectives. A participant said simply that they needed people and that while it took some time, she found people she could connect with.

"You need people eventually I found people who I could relate to" (affected female, RP)

It also dawned on participants that they have self-worth and could make a difference in the world, that the VI themselves, could add value to the lives of other people.

"I think it was in my twenties when I came to the realisation of who I am and the value I can add to society." (affected male, STGD)

This finding of reaching a point of stepping out into society after a period of social withdrawal, seems to support the suggestion that the social development of young people with visual impairments needs to be addressed through conscious, planned intervention strategies to help them overcome common barriers such as peers' discomfort with them in social situations and difficulties with transportation (www.afb.org)

Intra-personal Perspectives

The other stream of change participants demonstrated in their pursuit of autonomy evident in areas of personal growth with respect to life perspectives and general outlook within the context of their VI. The personal growth points participants exhibited comprised adjusting "attitudes," surrendering to God in "faith," believing that there was "hope" of a cure or treatment in the pipeline and through "acceptance" of what they could not change (Figure 3.5).

Participants displayed a change in attitudes over time. At the time of the study, they generally expressed positive attitudes towards life which influenced how they managed themselves and were able to assert control. A participant spoke about the mental perspective necessary to deal with being VI (affected male, STGD).

"It's a mind thing to cope well ... you can't feel bad about things you don't have any control over."

The stage in their lives when their attitudes towards their condition changed for the better escalated their adjustment process. Participants spoke positively about the situation they

found themselves in with VI, they viewed it as a gift from God and therefore something to be thankful for.

“It’s sad but I always look at it in a positive way. I mean we are given things in life and children like that is a gift ...God won’t give you something that you can’t handle, so we always try to look at the bright side of things.” (carrier female, LCA)

Participants further related their positive attitudes with the view that being VI was not a bad disability as others were worse off. They told how VI was a “light-weight” disability and spoke about having a good life quality in spite of their VI. This change in attitude was fuelled by exposure to society through social integration and employment.

“It is only since I started doing athletics for the disabled ... showed me that this disability of mine was nothing compared to other peoples disabilities.... there is other people out there with much worse disabilities, so it’s not a big thing not to seewhile you still have a 100% life quality.” (affected male, STGD)

Positive attitudes also influenced participants disposition for the better. They spoke about being happy in spite of their VI.

“You realise that listen this is your life ...this is how it’s going to be ... so either you enjoy it and you make the best of it or you are going to have a miserable life” (affected female, STGD)

The finding in this study, that a positive attitude and approach to dealing with their disability, is beneficial to the process on exerting personal control. The concept is also reported on the websites of service agencies for the blind, which support and advocate for the VI. An article on the VisionAware website reports that a positive attitude, is important for success within the workplace. Furthermore, vision rehabilitation therapist, Linda Fugate, says that attitude, is the first step that people experiencing vision loss have to address, before they can proceed along the adaptation process. She says that without the willingness to try to overcome the challenges of vision loss, patients will not be able to find solutions to developing everyday living skills (www.visionaware.org). The participants in the current study certainly displayed positive attitudes about themselves and towards finding solutions to their challenges such as the use of different technologies, genetic testing and applied support, seeking employment, creating awareness and self-education. Results from another study support the relationship between positive attitudes and intra-personal perspectives such as hope as well as integration of the VI into broader society. The study

indicates that a positive attitude about self in the context of blindness is crucial to creating a form of normalcy (Snead and Davis. 2002).

Some participants found that their faith in God, helped them to deal with life as VI individuals. Amidst the many challenges that come with a life of low vision, they were able to be at “peace” and draw on their faith in God for courage and hope. This courage and hope helped with overcoming feelings of depression and the confidence with which to assert themselves. Participants related how their spiritual journey and faith helped them to survive the path of VI.

“I think it was a spiritual journey (to acceptance)..I am a christian and my belief system... (helped me)” (affected male, STGD)

“I have faith in God, this helps me a lot” (affected male, STGD)

“But I believe in God and I’ve prayed ...and believe that if they do get it ...at least we know how to deal with it now....” (affected female, STGD)

The theme of faith is not uncommon in general but particularly among the VI. In his book, “What I Saw When I went Blind”, John H. Erickson writes:

“When I look back over my life, or my two lives, actually - I can trace the path I followed in coming to terms with my visual impairment. I can also see the journey my spiritual life has taken me on. My developing understanding of God and His plan for my life has been another huge factor in making me the person that I am today.”

The participants who revealed their faith in God certainly displayed a peace and contentment with their circumstances beyond acceptance.

Hope was another prominent theme of intra-personal perspectives that emerged from the data. Especially in the context of the clinical trials that may soon be in SA, but also hope for a life of normalcy through genetic testing, technology, employment, awareness, acceptance and independency. Participants articulated personal hope for a seeing future.

“With the trials they will be able to correct that gene and you will be able to see again.” (affected female, RP)

Hope to participate in clinical trials when they are available was reported by several participants.

“If trials come, I do want to be a part of it, I will try!! if it comes to SA” (affected female, STGD)

Hope for a cure was often expressed but with a hint of caution.

“When I went to fetch the test results. I was excited I thought maybe they have a cure, but no they said maybe one day ...” (affected female, RP)

Hope for the invention of better enabling technologies.

“Even if in the future they have children with the conditiontechnology is always improving there will always be something...” (affected female, STGD)

Hope to be able to drive in the future once a cure is found.

“The Retina SA emails I go through them because I just want to drive” (affected female, STGD)

Hope for a better sighted future for their children.

“but by the time they have children ...there will be other options. Hopefully there would be some sort of a cure before that.” (affected female, STGD)

Hope of being something one day.

“She wants to be a physiotherapist ...so they do have vision for themselves as well...if we can make it easier for them...that’s all.” (carrier female, LCA)

Hope that they would continue to have hope.

“If they don’t have hope and if they don’t have your support ...then they have nothing.” (carrier female, LCA)

The author Helen Keller, wrote, “*Nothing can be done without hope and confidence*” and SA’s very own Desmond Tutu wrote, “*Hope is being able to see that there is light despite all of the darkness.*” The participants in this study all displayed hope for the future in terms of possibly being able to see again with the research endeavours underway around the world. Also to be able to drive again and hope that their children and grandchildren will have a better future.

Adjusting their intra-personal perspectives with respect to attitudes, faith and hope was a natural precursor to acceptance. All participants found that they eventually reached a point of personal acceptance. Self-acceptance as well as acceptance from others is what the data revealed. Self-acceptance was a process along which many psychosocial issues had

to be overcome (as discussed earlier). Acceptance from others was important to participants, however, they reached a point when this became less important and they could find “peace” and “happiness” independently. No longer did participants feel ashamed of their VI, when they reached a point of peace and acceptance.

“I realised, look I am like this and I don’t have to be ashamed, there are other disabled people in this world. So I’d say actually it’s only the past couple of years that I’ve you know, made peace with it” (affected female, RP).

Participants learnt to accept that people would always look at them strangely when out in public. They spoke about reaching a point when they were not bothered by strange stares.

“But I can’t be bothered anymore to care what they think, what they ... because it doesn’t bother me ... now I put my phone near my eyes because that’s how I see it I don’t care who looks, who gives comments, I don’t give a damn...and I live my life because if I don’t, who is going to do it” (affected female, STGD).

Acceptance was so complete in some participants that they spoke about having to carefully consider whether they would participate in any curative treatment should it become a reality. They spoke about total acceptance of self including being VI.

“But now I can’t think why I felt like that (ashamed), even if they have a cure ... I don’t think I will do it immediately because I am absolutely fine with the way that I am. I really like my life so...if there is a cure ... I will have to think about it “ (affected male, STGD).

Participants described trusting in themselves and finding peace as key factors in overcoming the fear, anxiety, feelings of being ashamed and related themes as described in this mini-dissertation.

“So it’s trusting yourself and getting over that fear, I had to make peace with it” (affected female, STGD)

They spoke about learning to accept that which they are unable to change. To have an attitude change and to manage the situation as best as they could with what is known and available.

“but then again I can’t change it, so I had to accept it and learn to live with it...and make the best of it, and that’s really all I can teach my children....work around it...and let people know” (affected female, STGD).

With respect to seeking acceptance from others, participants told how this is a natural and normal endeavour, but that VI complicates the process. They therefore told how significant

finding the right friends and life partner was (discussed earlier) and that acceptance from others influenced self-acceptance.

“At that stage in your life you seek acceptance... so on top of seeking acceptance from a personal view...you now have a condition ...so you find that friends are important and find a partner who accepts you as a person ...it's easy to accept yourself” (affected male, STGD).

Professionals have aligned the responses to a loss of vision, to the “stages of grief” as outlined by Jurecic (2015) which include, denial, anger, depression and acceptance.

The participants in the current study have indeed experienced these psychopathological responses (as described earlier). The theme of “acceptance” to emerge prominently was indicative of the journey they were on towards adjusting to life as a VI person as an almost natural next step. Furthermore the various “support” mechanisms participants could benefit from (as described earlier), the positive “attitudes”, “faith” and “hope” they disclosed during the interviews contributed to them accepting their vision loss and in turn the courage to take “control” and achieve some “independence” where they could. The life stories revealed during this study epitomises the meaning of the following quotes:

“Acceptance doesn't mean resignation; it means understanding that something is what it is and that there's got to be a way through it.”Michael J. Fox

“When we are no longer able to change a situation, we are challenged to change ourselves.”Viktor Frankle

“Being challenged in life is inevitable, being defeated is optional.”Roger Crawford

3.3. A Genetic Condition Affecting The Back Of The Eye

After a long emotional journey with VI, learning how to navigate the world of VI and exposure to various sources of information from support services, most participants related a reasonable grasp of the knowledge of their condition with respect to the medical and genetic information and associated risk perceptions (Figure 3.6.).

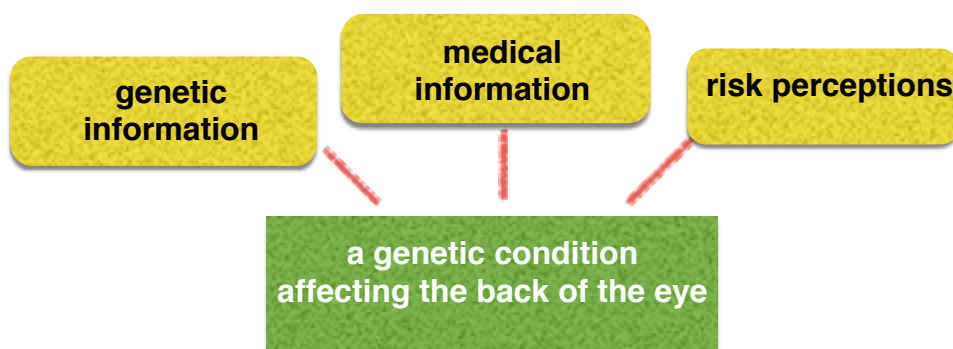


Figure 3.6. Illustration of the theme a genetic condition affecting the back of the eye, and its sub-themes.

The language used was very basic and non-scientific, however, the essence of the information was correct and accurate. They spoke about carriers and where the genes came from and who is at risk, for example as two participants stated:

“Say my son marries a woman that’s also carrying the gene they could obviously have a child with the same disease” (affected female, STGD).

“I know it is a combination of a genetic disorder from my father and my mom, non of them have it but they were carriers and they gave the error on the two chromosomes” (affected female, RP)

Participants were also very aware of the medical aspects of their condition and spoke about the retina and defects at the back of the eye.

“It is a genetic condition affecting the back of the eye, all I know its a faulty gene related to the retina”(carrier female,LCA).

They understood the implications for their offspring and what testing was necessary to get definitive answers.

“errrr my kids are carriers for sure... my husband never got tested because that would give us an accurate answer if they would be having it or not.” (affected woman with STGD)

They were confident in their knowledge and understanding to speak to family members such as unaffected siblings about their possible carrier status and what steps to take if they wanted to know.

“I did the same with my brothers daughter who got married recently I made sure she knows what to do ...gave her the report as well” (affected male, STGD).

A few participants displayed some scientific knowledge.

”After the human genome project it was possible to compare the genomes of normal and STGD people and now knowing what the defect is it is now possible to manipulate the faulty gene to try and avoid marrying somebody with the same faulty gene” (affected male, STGD).

Taken together, several participants were aware of the medical and genetic aspects of their condition and the risk implications for their offspring. They were aware that testing options were available to help manage their risks. Interestingly, even though they were perceived to grasp the essence of their condition, many were not able to speak confidently

about the information and tried to gloss over responses to probing questions and were apologetic about their inability to articulate the information well.

These findings suggest a perceived improvement in knowledge of disease in patients who receive multi-pronged input and awareness of information. This is in keeping with the findings of Bannon et al. (2014), who reported an increase in disease-knowledge among Lynch syndrome patients and family members, who were exposed to an additional educational conference designed to re-enforce and complement individual genetic counselling. The report details that the educational conference provided information on disease knowledge, supportive networking and needs assessments. The focus of the Retina SA support group of which the participants in this current study are members, echoes the input and exposure received by participants in the Lynch syndrome study.

Other participants, however, showed a very poor understanding of their disease and responded with inaccurate knowledge. Participants spoke about not knowing their carrier status, where the disease came from or how it is inherited and passed on.

“But I don’t know if I am a carrier” (affected female, STGD)

“I don’t know how its passed on” (affected female, STGD).

“I still don’t know really a lot about this problem” (affected female, STGD).

They spoke in an imprecise manner about current research trends.

“Because I don’t know if I understand but I think they said that one day they can take out that gene and replace it” (affected female, STGD).

A carrier mother explained the genetics of the disorder with a statement containing contradicting and inaccurate facts about risks to offspring and why family histories present with skipped generations.

“If only one of us (parents) were a carrier, then only one daughter would have had it (the disorder), because I know its skips generations, I know my daughter has two mutations“ (carrier female, STGD).

All participants in this study had received genetic counselling and received additional educational material through newsletters, AGMs and workshops offered by Retina SA. Therefore this finding that participants showed a poor understanding of their condition, in

spite of the level of exposure to information from varied sources, may suggest that some of the well documented barriers to effective communication may have been contributing factors. Communication is a complex process, influenced significantly by the environment. Other barriers to communication may include language differences, family interference, gender differences, and patient’s anxiety (Norouzinia et al. 2016). Furthermore, one has to consider the possibility that these participants were products of inefficient approaches to genetic counselling. However, firstly, many participants related a high regard for their genetic counsellors and secondly, the researcher had a a clear sense that the basic essence of the information was mostly correct. Any of these factors could have contributed to the limited knowledge and understanding portrayed by a few of the participants in this study. Furthermore, while some studies have reported good information recall by clients (Somers et al. 1988), others show that clients do not always accurately recall information and risks discussed during counselling sessions (Evans et al. 1994). The knowledge of participants from the current study echoes the findings from both of these studies.

3.4. Paving The Way Forward

At the time the interviews were conducted, all participants were able to make commitments for the present and future concerning among other aspects, their education, reproduction and careers within the context of their genetic diagnoses. All participants in this study had undergone genetic testing (Figure 3.7.).



Figure 3.7. Illustration of the theme paving the way forward, and its sub-themes.

In particular, they were able to use the genetic information at their disposal and knowledge about their disorder to opt for genetic testing for their children. This was an effort to gain insights into the basis of their disease and the associated risks both for themselves and

family members. The data revealed that knowing their genetic diagnosis was very important and pivotal for each participant toward finding answers, clarity and meaning to the confusion, uncertainty and despair that each faced at the outset of their vision loss. Each displayed a measure of satisfaction in knowing their genetic diagnosis given the extended period of uncertainty each faced until that point. A carrier mother described the meaning of knowing the basis of her children's condition as a defining moment.

"The gene test identified LCA, and that made all the difference. Now we just have to pave the way forward" (carrier female, LCA).

There was an understanding that the information gained from the test could be used in the future to help other family members determine their risk status.

"I've spoken to them (children) about preventing it in their children, although I don't want to mention it/say that I shouldn't have had you guys" (affected female, STGD).

"I did the same with my brothers daughter who got married recently I made sure she knows what to do ...gave her the report as well" (affected male, STGD).

Using this genetic information in reproductive choices, participants showed different views. One participant affected with STGD told how notwithstanding the implications, he and his wife decided to have children. This participant was belonged to a family with several affected members.

"Some people might not want to then (have children) because they wouldn't want to pass it on to their kids but we just thought you know what? We going to take the chance and ja we had our kids anyway" (affected male, STGD).

Another view which emerged was that caution needs to be exercised before choosing to have children.

"Well I told my children already that one day when they meet a partner and they decide to have children that its better to go get themselves tested before they make that decision" (affected female, STGD).

"I think it will be better because if they can avoid not for their children to be like this ...why not" (affected female, STGD).

Another strong motivator to do genetic testing was the knowledge that it was a deciding factor in establishing their eligibility for clinical trials (a focus area of Retina SA). Yet, while most participants spoke about wanting to participate in clinical trials when it becomes available to patients in SA, they emphasised that they would only participate if there was a

certainty that it would improve their sight. Losing the little vision they had, was not a risk they were willing take.

“I’m willing to go for trials but if there is a chance that I will lose my sightI won’t do it ... I’d rather keep what I have” (affected female, STGD).

In terms of paving the way forward with friends, several participants noted that they were particular about picking friends because a lot of people are unable to deal with a VI person. They explained that having understanding friends helped their self-esteem and dignity.

“I’m choosy with my friends, because someone who makes fun of me or puts me down is not my friend and that’s it” (affected female, STGD).

Participants were also able to overcome some of the daily challenges associated with VI and in so doing, carve out a better existence for themselves going forward. This was achieved by making necessary changes to living-spaces, country of residency and even their general approach to these challenges.

“We just busy renovating our house now so we’ll work around that to make it as mobile friendly for me as possible ja that’s as far as the future goes” (affected female, RP).

“When I went to varsity I met my husband and I left Madeira. because I thought that I would get more help in SA (I was 21 years old)” (affected female, STGD).

Although participants spoke with a sense of peace and ease about “paving the way forward”, it was interesting to note that they highlighted the difficulty and extra consideration necessary in doing so, which sighted people take for granted. They spoke of having to decide to do simple things without knowing whether they would be able to do it. But they realised that decisions have to be made.

“Its scary also to make decisions about what you can or can’t do ...will you really be able to cope with this...because it takes me longer to look at something than other people, in the workplace especially” (affected female, STGD).

“Decisions have to be made all the time”(affected female, STGD).

Participants spoke about feeling comfortable preparing for their short term future, in light of the lingering uncertainty of whether they would become blind at some point in the future.

“I don’t plan long termI plan short term and all my plans go around the benefit of my kidsall choices and decisions revolves around them” (carrier female, STGD).

Plans were therefore focused on the short term and family, particularly their children. The view was that they provide their children with as normal a life as possible while still sighted.

“I think that the impact on them at a later stage when I go completely (blind) heaven forbid” (affected female, RP).

The verbatim quotes from participants in this study (who had been exposed to genetic counselling as well as additional educational support), show a group of people with visual impairment who were able to “pave out a way forward” regarding genetic testing, reproductive choices, as well as general life choices about the future like traveling and home renovations. However, little evidence is reported that genetic counselling improves informed choice in a general context. Kaiser et al. (2002) and Hunter et al. (2005), however, do report that genetic counselling can reduce decisional conflict, it must be noted though that these studies were conducted in prenatal group settings. McAllister and Dearing (2015) therefore cautions whether this view can be applied to general clinical genetic service settings. It is therefore uncertain whether the findings in the current study, of counselees' ability to make informed choices is due to the genetic counselling and additional educational exposure and support they received, or other contributing factors.

3.5. Summary Of Findings

The findings of this study are presented in this chapter, and a discussion of the themes that emerged. The main themes along participants' journey of VI were “a roller coaster journey,” “learning how to survive,” “a genetic condition affecting the back of the eye” and “paving the way forward”.

The first reactions to vision loss participants experienced were classified as psychopathological responses and included anxiety, denial, depression, social withdrawal and anger (Figure 3.2). Learning how to survive talks to mechanisms participants sought and put into place to make dealing with the loss of vision easier. Being organised in their daily activities was one approach. Exposing themselves to various support systems proved to be another means of learning how to survive VI. Support was gained from agencies such as schools, CGSs, online support, relationships and employers. The final avenue in learning how to survive which this study exposed, involved intricate yet conscious streams of life changes. These adjustments were classified into two groups called assertive actions

and intra-personal perspectives. Assertive actions saw participants venturing out to: use technologies, finding employment, creating awareness and integrating into society. Changing their intra-personal perspectives on life as VI people saw participants altering their attitudes, relying on their faith in God, living with hope and accepting their circumstances. A genetic condition affecting the back of the eye, captures participants' grasp of their diagnosis. Most participants had a keen need to know more about the cause of their condition, how it came about and how it would affect future generations. Paving the way forward details participants efforts towards improving their immediate and short-term futures.

Chapter 4 CONCLUSIONS AND RECOMMENDATIONS

In this chapter conclusions are drawn based on the findings and literature in the field of VI, it outlines the implications of the findings, and possible recommendations for practice and research.

This study explored the experiences of individuals living with IRDs with a focus, on education (knowledge), decisions (choices) and adaptation (coping). Understanding the experiences of people with VI is invaluable in reflecting on the needs, and services available to the VI community within SA. Long standing services to the VI population in SA do exist and operate uniquely as a collaborative approach involving the CGS for IRD and the support group, Retina SA. Therefore participants within the context of the CGS, provided subjects for this study, who were accessible and willing to participate. The qualitative methodology using the semi-structured interview approach successfully drew out in-depth raw data from participants. The research highlights some important findings which may serve to inform the scope of practice of health care providers serving the VI community.

The four broad themes identified in this research study paint a picture of a journey participants travelled with VI, and include:

1. a roller coaster journey,
2. learning how to survive,
3. a genetic condition affecting the back of the eye, and
4. paving the way forward.

Theme one captured an in-depth view and understanding of the personal struggles, feelings and reactions participants experienced, particularly at the start of vision loss. Theme two describes the needs of the VI and exposes their management, support structures and coping mechanisms as they progressed along their journey. Theme three provided insights into the varying levels of comprehension of the IRD diagnosis, the genetic basis and risks for themselves and family members obtained over time particularly through various sources of support systems. Theme four described an appreciation of the mental focus, discernment and commitment necessary to craft out a comfortable existence and future for the VI individuals who participated in this study.

The roller coaster journey this study has shown participants underwent, mimics the process of dealing with grief, as those who have lost a loved one to death (Leo et al. 1999), however, along with additional psychopathological responses. The initial psychopathological responses were thus varied and prolonged and the contributors to the roller coaster journey participants experienced. The anxiety, denial, depression, social withdrawal and anger exhibited a sense of despair and hopelessness during these initial stages of disease progression confounding the struggles for survival. No differences were noted between the roller coaster experiences of the males and females within the study group within which there was an even gender distribution. A recent study by Diniz-Filho et al. (2016) also reported no significant gender bias for depression after vision loss. The responses to vision loss were a means to help them process and come to terms with the reality of losing their vision. In fact in one study, the negative effects to VI was compared to those of cancer patients about to undergo chemotherapy (Nollett et al. 2016). The observations of the current study, however, were intriguing because to some degree, it contradicts other studies on the psychology of VI. Firstly, while the profile of participants in the current study aligned with the protective factors to psychopathological responses reported by Ash and colleagues (1978), it did not seem to spare these individuals from the myriad of negative psychopathological responses reported here. Secondly, It has recently been reported that adolescents experiencing VI, statistically show a lower prevalence of negative psychological effects such as depression, when compared to individuals who were older when diagnosed (Garcia et al. 2017). The trend reported in that study was one of worsening psychological effects to VI, with increasing age. Garcia et al. (2017), suggested that individuals who become VI in their younger years adapt and cope with

greater ease than older age groups. It is thus interesting that the participants in this study all related the roller coaster experience during their adolescent years.

Given participants detailing with regret, the lack of support by low-vision specialists during those early stages, it was reasonable to conclude that this may have significantly contributed to the many psychopathological responses they exhibited. This suggestion is further supported by the proposal by the WHO that daily challenges which the VI face can be caused or exacerbated by environmental circumstances (Sihota and Tandon. 2015). While negative effects of VI on the psychological well-being of an individual cannot be refuted, the depth and spread of the responses participants related, may not have been as prolonged and extensive had they received the necessary medical support. Finally, while there is a paucity of published evidence on the significant role of low-vision specialists such as ophthalmologists in managing the emotional effects of the VI, the studies by Moschos (2014) and Garcia et al. (2017) reported that support from these specialists could have an overwhelming impact for patient outcomes. However, the mechanisms of support may vary, including active emotional counselling, active listening or through the process of providing patients with definitive answers and information about their condition. It is the last mechanism which Garcia reports, which participants in the current study expressed to be absent from their care during the early stages after onset of vision loss. This line of evidence lends further credibility to the conclusion that the limited care participants received from low-vision specialists contributed significantly to their roller coaster journey.

Participants' align with the tasks of grieving suggested to be involved and necessary for the "process of mourning to be completed" and "equilibrium to be reestablished" (Worden. 2008). The tasks include 1) to accept the reality of the loss, 2) to work through the pain and grief, 3) to adjust to a new environment, and 4) moving forward with life. The journey of dealing with loss of vision experienced by participants in this study was certainly marked by these tasks as outlined by Worden. He also proposed that people suffering a loss may find themselves revisiting certain tasks and that grief is not linear. The findings in the current study indeed confirmed this report. The participants in this study, however, seemed to present a delayed start to this necessary tasks of grieving and in turn a delayed and prolonged journey to acceptance and general well-being.

Notwithstanding the perceived profound effect of the limited support from low vision specialists, all participants in this study were partially sighted and not blind, and this may also have confounded the varied psychological responses which emerged. Leo et al. (1999) outlined that psychopathological responses are more profound among VI as opposed to blind patients. The slowly progressive loss of sight is said to be more traumatic compared to when sight is lost instantly. The immense trauma in turn leads to the negative responses described in this dissertation. The personal impact of participants' VI was therefore shown to be a life changing experience with far reaching implications. The findings of this study therefore support the thinking that hereditary conditions often redefine the nature of the person. This was evident in the impact of VI on the physical, psychological as well as the social health of participants as well as their families.

The major theme of "learning how to survive" exposed participants' varied approaches to adapting to a life of VI, which was one of the objectives outlined for this study. The study findings showed that the emphasis placed on living an ordered and organised life, significantly impacted the daily survival process. It gave participants the sense of certainty and security within their immediate environment. Creating this safety and comfort zone for themselves gave them a sense of power with which to tackle bigger challenges in the world beyond their homes. A literature search for published reports on the ordered lifestyle of the VI, was not fruitful, however, agencies' websites which advocate for the VI place considerable emphasis on various approaches which the VI person could adopt to improve their daily lives (visionaware.org). These websites, however, report feedback on the experiences of VI individuals which are in keeping with the messages communicated by participants during the interviews of this study.

Mechanisms of support which participants did eventually receive, were found to make significant inroads into participants finding a way through the difficulties of losing their vision (and learning how to survive). In this study the availability of support as well as resources influenced how participants coped with their loss. This finding which highlights the significant positive impact of support on the survival journey of the VI, confirms other reports (Stevellink. 2015). However the findings attest to and are consistent with the report that support varies widely among individuals, (Antonucci and Johnson 1994). The varied support mechanisms highlighted in this study, included schools, the range of services included in the IRD CGS in SA, online support, relationships, and employers. These findings reveal participants found various forms of support beneficial, depending on

individual circumstances and personalities. Choosing a school which meets the special needs of the VI child held significant sway in the ability of participants to adjust to the process of vision loss. The support received at special schools in terms of learning how to read braille, learning how to overcome daily practical challenges, having access to special teaching aids for the VI and the much needed emotional support impacted the lives of participants in this study significantly. On the other hand some participants did not see the need or value of the support of special schools for the blind. CGSs support represented a meaningful interaction with participants as it afforded them the clarity and direction which they felt had been absent. The multidisciplinary service approach of the CGS for IRD in SA including patient support services such as genetic counselling and testing, psychologists, social workers and the patient support group Retina SA contributed significantly to addressing some of the issues participants faced. Participants' felt a sense of relief and clarity on obtaining a definitive molecular genetic diagnosis which was facilitated through the CGS. This finding is in keeping with the report by Berger et al. (2010) that a confirmed genetic diagnosis is a crucial step in the genetic management of patients and families as it provides a sense of certainty and direction. This was seen in the participants reaching a turning point in their lives once they were exposed to these services. The genetic factors, however, which confound reaching a definitive molecular diagnosis for an IRD (Berger et al. 2010), has and will continue to pose a challenge in providing patients with the timeous conclusions they seek and emphasises the need for CGSs to be on the cutting edge of scientific discoveries and developments. Online peer support was found to be a powerful source of comfort for those participants not comfortable with exposure to direct group meetings. In contrast, other participants enjoyed the direct interaction. Interestingly though, not all participants expressed the absolute need for comfort and support from similarly affected people, whether online or directly. This finding further supports the suggestion made in this study that it is a matter of personal opinion, need and personality, whether peer support would benefit the VI person. On the other hand, close family members and friends were found to be the best and most valued support system for all the VI participants in this study. The support network of personal relationships were found to help the VI to take full advantage of the additional support systems highlighted in this study. The support of employers was found to provide a sense of self-worth, self-confidence and independence. The pivotal role of these various sources of support in their survival as partially sighted individuals cannot be underestimated. Support for the VI, in whichever form, was shown to be crucial in the development of survival strategies. As found by Garcia. Similarly exposure to the support systems which participants in this

study spoke of, therefore did much to ameliorate the limits imposed by VI on the capacity to perform essential activities of daily life. The value of various support mechanisms for the psychological well-being of the VI, has been reported (Garcia et al. 2017).

Notwithstanding the value of good support mechanisms and networks, where possible, the ability to exercise autonomy and independence, was found to empower participants to navigate the survival process and provided a sense of dignity and self-respect. Control over their daily actions, making financial contributions to their cost of living as well as how their disorder impacted their lives, was found to be achieved through two streams of change or adjustments they made in their lives through a series of conscious decisions. Firstly, they took assertive steps to change their situation for the better through the use of technology, employment, by creating awareness and through social integration. The development of these independent living skills afforded participants control and hence a more positive outlook on life as a VI person.

Mechanisms of support, aids, and attitudes of the sighted towards the VI, or lack thereof, were found to all contribute profoundly to the general feeling of either being in control or hopelessness. The VI in this study revealed a general courageous and determined approach to life with the aim to succeed. They were confident and able to explore whatever options were available to them in order to achieve this goal of “accomplishment”. These observations among the VI are consistent with other studies. The study by Garcia et al. (2017) highlighted the association of technology with positive effects on the psychological well-being of the VI. More specifically, participants in this study spoke about how the iPad and iPhone had impacted their lives for the better. A study by Haji et al. (2015), reported improved reading ability on an iPad among VI individuals. The current study highlighted that the ability to read independently without assistance contributed significantly to participants sense of independence and control. Studies on the usefulness of technology and its impact in the lives of the VI is few, however, its benefits highlighted in this dissertation coupled with preliminary reports could suggest that low-vision specialists could more consciously promote their use to patients (Robinson et al. 2017). Furthermore, as this study revealed the reticence of participants to draw attention to themselves in public with the use of a magnifying glass, the popularity of electronic devices could underpin the benefit of destigmatisation.

The impact of VI on career and occupational goals emerged in this study to be an ongoing challenge and is a relevant consideration, however, not many studies have investigated

these effects. One study showed though, that VI was associated with unemployment and a decreased socio-economic status (Rahi et al. 2009). Interestingly though, the profile of most participants in the current study, defied these findings as most were employed and of a middle to upper socioeconomic status. It may be that the protective factors against the negative effects of VI reported by Ash and colleagues (1978), indeed played a role in their overall process of adaptation, but at a later stage in the lives of these VI participants. All the male participants were employed and in satisfying careers, one was retired, and only two females were unemployed. The unemployed females could fit with some of the findings of Rahi et al. (2009) as they did complain of the challenges of finding employment. The challenges they highlighted were a lack of awareness of employers and an unwillingness to accommodate their disability. However, it's believed that these two participants exhibited other contributing personal factors which considerably influenced their views on employment. Both wanted to be able to find employment, but were not entirely confident in uncontrolled public spaces. They showed a preference to only operate within their comfort zones of the home and other spaces only when completely necessary. VI has been shown to affect the social behaviour of individuals due to the physical challenges of maintaining eye contact as well as the inability to recognise facial expressions and other nonverbal communication cues, important aspects of effective communication (Frith. 2009). The effect of VI on the social behaviour of participants in the current study was observed, however they did reach a point of acceptance of self and self-confidence which facilitated the movement from isolation to social integration. The many factors outlined in this dissertation from personal progression to support and exercising autonomy, is believed to have had a profound positive effect on the social behaviour of study participants.

Secondly they adjusted their personal perspectives on attitudes, faith in God, hope and acceptance. These personal adjustments contributed significantly to the adaptation process of the group of individuals who participated in this study. The streams of change was found to be highly empowering and gave participants a sense of the value they could add to the world around them. Other studies also pinpointed the value of exerting autonomy in the lives of the VI with respect to their confidence, feelings of self-worth and independence (Stevellink. 2015).

The study found that the study approach which facilitated participants relating their experiences allowed for exploration of the genetic knowledge of participants, another specific objective of this study. Participants' stories exhibited a reasonable yet basic level

of knowledge and understanding of their diagnosis, the genetic basis and risks involved. Notwithstanding the perception of a basic level of comprehension, the essence of what they perceived to be important for further management of their disorder for themselves as well as family members, was deemed sufficient. The research did, however, find that there was a lack of understanding in some participants. They were unable to articulate any information regarding their condition and confessed that they didn't understand. This finding was interesting as all participants had received genetic counselling as well as additional educational exposure from Retina SA through workshops, AGMs and regular newsletters. Input from Retina SA complements individual genetic counselling for updating disease knowledge, supportive networking and needs assessment. Furthermore participants in the current study identified genetic counsellors, as an important source of their disease knowledge even though exposure to a genetic counsellor comprised at most 2 sessions.

The combination of a basic and lack of understanding found in the study participants were initially taken to be either due to poor information recall or to some of the known barriers to effective communication between health care providers and patients (Norouzinia et al. 2016). One of the main objectives of genetic counselling is to give medical genetic information to patients in a manner that they can understand and recall. The level of recall of information is reported to be a function of, 1. the type of information communicated, 2. when during the process of communication it is presented, and 3. location (Giles et al. 1982). The researchers considered that scientific and medical information communicated in a complicated manner can be daunting for lay people to grasp and put into perspective. However, Michie et al. (1997) suggested that patient knowledge after genetic counselling was not associated with the quality of information received. Communication is a dynamic and complex process influenced by many factors, and for it to be effective, is reported to be an important aspect of patient care (Norouzinia et al. 2016). However, all participants' receptivity to management options and prospective treatments in spite of the basic and some lack of knowledge shown, indicated to the researcher a reasonable underlying genetic knowledge but possibly some insecurity in articulating the information well. The strength of this conclusion is supported by the Norouzinia et al. (2016) report which suggested that effective communication had a profound effect on patient's perceptions pertaining to management and outcomes. Furthermore, Rutherford et al. (2014) reported a positive association between knowledge and uptake of treatment as well as patients' knowledge.

Finally, participants' learning about their personal genetic information and more importantly grasping the implications there of, was revealed in this study to represent a crossroads in their lives towards finding renewed meaning and direction. Particularly after the period of confusion during which they exhibited psychopathological responses to their vision loss. The new found perspicuity concerning their condition, gave participants the direction, energy and drive to firstly, discern a future for themselves, and secondly, to make the necessary choices to be able to realise their vision for the future. This is in keeping with Wordens (2008) fourth task in the grieving process. The theme "paving the way forward" captures the study objective to explore participants decision-making abilities. The study found that participants were able to carve out a system of existence for themselves both for the present and the future, particularly the short-term future within the context of their VI. The raw data revealed many experiences depicting a group of VI people making life choices ranging from genetic testing, home renovations, personal relationship choices, career, educational decisions, creating awareness to traveling abroad. It was found that while everyday choices for the VI are not easy, that this group over time had learnt to transcend the challenges associated with their condition and find purpose and meaning to life. Specifically, discerning the value of genetic testing for risk management for the future and most importantly for establishing their eligibility for clinical trials and treatments currently being investigated and developed for the IRD group of disorders (<http://www.clinicaltrials.gov/>). The view and hope of a future where treatment for VI would be possible, was found to be a motivator to take conscious steps towards paving the way towards that reality. They used the genetic information also to make reproductive choices within the context of their beliefs and values. They were found to have enough confidence to be choosy with respect to friendships. They chose to only draw those people close to them, who would help them to grow and develop into more well-rounded members of society. They chose friends who respected them. Participants showed that they were able to craft out a life for themselves but recognised that considerable more effort and consideration was necessary in paving the way forward, things most sighted people take for granted. The findings therefore describe a group of individuals who exhibited resilience in their ability to rise above their shame and fear to a point where they were able to accept who they were as VI people, and to forge a way forward for themselves with courage. Participants in this study therefore demonstrated the findings of Brown (2010), a leading expert on shame, authenticity and belonging by engaging with the world as worthy members of society, notwithstanding their VI.

4.1. Summary

VI patients within the CGS for IRD in SA who constituted the target group for this study portrayed a journey with VI marked by similar experiences. Their experiences were almost reminiscent of a chronological timeline since the onset of VI to the present. The initial psychopathological responses to vision loss (which presented in no particular order), marked the first stage followed by a stage when they applied themselves to learning how to adjust to their new reality of VI. The third stage along their journey, saw them coming to terms with the facts about their condition and the implications thereof. Having developed mechanisms of survival (adaptation) and with a better understanding of the gravity of their VI, the study found the final stage of the journey saw them using the skills they developed over time, to manage their condition to ensure a better future for themselves and family members. The participants therefore adapted to their VI over the time of their journey explored in this study. During this time they gained a reasonable level of understanding and a knowledge base with respect to their condition, and they displayed the confidence and self-esteem necessary to make life choices for the present and future within the context of their VI. The aim and objectives of the study to investigate a group of IRD patients' perspectives on their journey with genetic counselling and Retina SA support, through the exploration of:

1. participants' understanding of the medical and genetic aspects of their diagnosis
2. how their counselling and support group experiences have influenced reproductive and life choices (decision-making)
3. how their counselling and support group experiences have influenced how they have adapted to their diagnosis,

were therefore realised as well as additional interesting findings which marked participants' journey with VI.

4.2. Strengths And Limitations Of The Study

- The raw data were obtained from several participants and the similarities of emerging themes therefore accurately represented and established the findings. The experiences and perspectives from each participant were integrated and assimilated to structure the findings with which to address the research question.
- The language used during interviews, the open-ended questioning style and participant chosen private locations, allowed participants the comfort, assurance of confidentiality

and above all the willingness to express themselves freely which facilitated the collection of rich in-depth raw data.

- Audio recordings and field-notes provided the raw data and valuable sources for analyses in structuring themes, meaning and viewpoints from participants towards drawing final conclusions.
- A limitation of the study is that inherent in qualitative research which involves collating peoples' perspectives and views, the researcher could only trust that participants were honest and that they gave a truthful account of their experiences, perspectives, feelings and actions. Dishonesty could have influenced the authenticity of the research findings.
- All participants were members of the organisation Retina SA. Whether these findings can be extrapolated to different VI populations who are not Retina SA members remains uncertain.
- While research bias was consciously avoided, often this can be unavoidable due to the human nature's built in bias to ones own views and beliefs.
- The study set out to gather comprehensive, in-depth raw data and the intensive nature of this research approach could have had a limiting effect on the scope of the study.
- The middled to high socio-economic status of participants may have skewed the collective views and experiences expressed.
- The willingness of participants may have biased the findings positively, as unwilling subjects' experiences have provided a different view.
- The retrospective nature of the study, relied on the ability of participants to clearly recall their earlier lived experiences.

4.3. Recommendations

In view of the current research findings, the following recommendations for practice and future research could be made:

- Specialists within the low vision field of medicine, become better equipped to understand and manage the spectrum of IRDs, as this group of conditions do not have a favourable outcome. This can be arranged through the establishment of continued medical education programmes with the Health Professional Council of South Africa.
- Networks be established with the clinical genetic services team and low vision specialists to effect better management and care of patients with IRDs. A collaborative multidisciplinary approach will be valuable towards shortening the time between clinical

and genetic diagnosis as well as crucial counselling services. This may facilitate a shorter period of adaptation and adjustment.

- Guidelines and policies for the practice of low vision specialists be established.
- Patients receive ongoing educational exposure over and above the counselling sessions usually performed pre- and post-DNA testing in order to facilitate continued and updated knowledge to patients and families.
- It would be valuable to explore the experiences of an expanded study group of VI people and to including individuals with different socioeconomic backgrounds.
- A lot of research has been undertaken on the psychopathological responses to vision loss but little research has investigated possible approaches to minimise any negative effects. It would therefore be useful to investigate approaches to the management of VI with the view to maximise the ease with which patients adjust in their journey to acceptance and well-being.

This study has shown that the VI are prone to many repercussions. An in-depth understanding of the psychological consequences of VI, the resources and approach focussed on minimising potential negative effects and maximising the ease with which patients move to a stage of general well-being, is pivotal in the holistic care and management of VI patients.

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Appendix A

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INFORMATION AND CONSENT FORM

Brief Overview Of The Study

Exploration of the impact of genetic counselling and patient support group involvement on retinal degenerative disorders (RDD) patients: a qualitative study

The research study is being conducted within the Division of Human Genetics at the University of Cape Town for a minor dissertation towards a Masters degree in Genetic Counselling. The study will investigate the experiences of Retina SA members who have undergone formal DNA testing and counselling and explore whether it (their experiences) have had an influence on their basic medical genetic knowledge about their diagnosis, decision making and adaptation to living with a retinal disorder.

Emerging findings will hopefully highlight the benefits of offering a clinical genetic service to patient groups in collaboration with support groups and which aspects of service delivery should be reviewed in order to provide improved support and care. The study is therefore aimed at the RDD patient group as it is the only genetic service in SA which is offered in close collaboration with a support group. Your participation is appreciated provided:

- you are 18 years and older,
- you have received genetic counselling,
- you are a member of Retina SA,
- you are fluent in English/Afrikaans.

A personal questionnaire based interview will be conducted to explore your experiences. The duration of the interview will be approximately 1 hour long and will be audio recorded to ensure that the information is captured accurately. The recordings as well as any other hand written notes will be kept confidential and anonymised.

The study will not provide any medical or financial assistance for you. Your participation is voluntary and thus you are able to withdraw from the study at any time, if you feel the need.

If you remain interested in participating 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, you can give verbal consent and have a witness sign on your behalf.

Participant Information

1. Your participation will remain anonymous and privacy will be ensured by omitting names so that confidentiality will be maintained and information gathered will only be used for the research project.
2. Your name and information discussed in the interview will not be given or discussed with other participants or anyone else not associated with the study.
3. The interview will take place at a private location of your choosing, at a suitable time and should not take longer than 60 minutes.
4. This is a once off session.
5. Some questions may be sensitive and may cause an emotional reaction, but the risk of harm to you is small. Should you wish to see a counsellor, arrangements will be made by the researcher via Retina SA.
6. Involvement in the study may contribute towards new insights into genetic service delivery and management of RDD patients and families.
7. The interview will be recorded for research purposes and information discussed will be accurately transcribed. These recordings will be locked away and saved onto a password-protected computer. Only the researcher and her supervisors will have access to this raw data. The recordings will be destroyed after the research has been finished. No record of your name will appear anywhere in the mini-dissertation write-up or ensuing publications.
8. The researcher will attend and conduct the interview.
9. If at any stage you no longer want to participate, your decisions will not have any negative effect on your ability to access medical services or those offered by Retina SA or the UCT RDD clinical genetic service.
10. This study has been approved by the Human Research Ethics Committee at the Faculty of Health Sciences of the University of Cape Town (REF: 422/2016).

Consent For Study Participation

To Whom It May Concern:

I, the undersigned..... confirm my consent to participate in the research study, *Exploration of the impact of genetic counselling and patient support group involvement on retinal degenerative disorders (RDD) patients: a qualitative study.*

1. I understand that my participation is voluntary and will involve me being interviewed
2. I understand the objectives of the study as outlined above
3. I understand that the interview will be audio recorded
4. I understand that my identity will be kept confidential
5. I understand that the information learnt will not affect me detrimentally in any way
6. I understand that the information will be anonymised for analysis and publication purposes
7. I understand that I can withdraw from the study at any stage
8. I understand that my participation poses minimal risk of harm and arrangements for counselling will be made if I require it.
9. I've read the Brief overview of the project and Participant Information provided
10. _____ has explained the information of the study to me in English and I clearly understand the information

Place :Date :Participant's Signature :

InterviewerName.....

Date: Place:Signature.....

IMPORTANT INFORMATION

Dear Participant

Thank you for your involvement and participation in this study. If you have any questions about the research project or if you want to withdraw from the study, you can inform the researcher or the supervisors, whose contact details are below:

Rene Goliath(researcher) cell: 082 909 6772 email:mntren001@myuct.ac.za

Tina Wessels (first supervisor) email: tina.wessels@uct.ac.za

Jacquie Greenberg (co-supervisor) ...email: jacquie.greenberg@uct.ac.za

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

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

Appendix B

Question guides

1. Lets start by Telling me about your experience or journey with your diagnosis of

Knowledge/Education

2. Tell me about your condition, clinically (RP/STGD) and what it means (see #3 & 4)
3. What is yr understanding about how RP is passed on?: Tell me how you would explain your condition to people in your family who may need to know
4. Tell me a bit about how this condition could affect other relatives? (brothers, sisters, aunts, uncles, cousins)? (see #7)

Decisions/Choices

5. How would/did your experience influence how you make decisions that will change the future for your children/any children you might have? (see #7)
6. Tell me how do you get the non-medical help you/your family need(s) (e.g. educational, financial, social support)?
7. How would you control/manage how this condition affects your family? (see #5)

Adaptation/Coping

8. Tell me how do you feel about the impact of the condition on your child(ren)/any child you may have? (see #13)
9. How does thinking about the condition in your family make you feel?
10. Tell me about anything good that has come from having this condition in your family
11. How was your experience in learning to **cope** with having this condition in your family?
12. How do you feel about making plans for the future?
13. How do you feel about having (might have) passed this condition on to your children (see #8)

Appendix C

Institutional Review Board (IRB) number: IRB00001938

This serves to confirm that the University of Cape Town Human Research Ethics Committee complies to the Ethics Standards for Clinical Research with a new drug in patients, based on the Medical Research Council (MRC-SA), Food and Drug Administration (FDA-USA), International Convention on Harmonisation Good Clinical Practice (ICH GCP), South African Good Clinical Practice Guidelines (DoH 2006), based on the Association of the British Pharmaceutical Industry Guidelines (ABPI), and Declaration of Helsinki (2013) guidelines.

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

