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The Impact of Genetic Disease on the Family: Examining the Relationship Between Psychological Well-Being, Social Support, and Spirituality in Unaffected Carriers of Leber's Hereditary Optic Neuropathy

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THE IMPACT OF GENETIC DISEASE ON THE FAMILY: EXAMINING THE
RELATIONSHIP BETWEEN PSYCHOLOGICAL WELL BEING, SOCIAL SUPPORT, AND
SPIRITUALITY IN UNAFFECTED CARRIERS OF LEBER'S HEREDITARY OPTIC
NEUROPATHY

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Submitted in Partial Fulfillment
of the Requirements for the Degree
of Ph.D. in Counseling Psychology

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Abstract

The current research was undertaken to explore the psychological well-being of mothers and siblings of an individual with vision loss symptoms due to Leber's Hereditary Optic Neuropathy and whether perceived social support and spiritual involvement and beliefs served as protective factors as these family members coped with their child or sibling acquiring a disability. One hundred seventeen participants, 65 mothers and 52 siblings completed a Demographic Questionnaire, the Outcome Questionnaire-45, the Impact of Event Scale, the Spiritual Involvement and Beliefs Scale, and the Multidimensional Scale of Perceived Social Support via an online internet survey. The data revealed that both mothers and siblings experienced higher levels of psychological stress than a normative sample and siblings experienced significantly higher levels of interpersonal problems and social role dysfunction than a normative sample. The results also showed that mothers of children with vision loss due to LHON showed significantly lower levels of psychological stress as levels of perceived social support from family and a significant other increased and siblings of a person with vision loss due to LHON showed significantly lower levels of psychological stress, interpersonal problems, and social role dysfunction with higher levels of perceived social support from family. Implications and suggestions for future research are presented.

Chapter I: Introduction

Currently, there are about 4,000 known genetic diseases, with new ones being discovered every year. The vast majority of such diseases are extremely rare, only affecting one in several thousand or million people worldwide per year. A high number of these individuals are young adults who are faced with, and diagnosed with, a range of genetically transmitted medical conditions that are associated with progressive and often severe acquired disability. Research that informs medical diagnostics, management, and treatment options is obviously essential, but not sufficient for understanding the factors related to optimal treatment response. This study explored the relationship between a number of psychosocial factors and optimal treatment outcomes in adults whose family members are affected with the rare genetic disease known as Leber's Hereditary Optic Neuropathy. The overarching goal of this research was to unveil relationships between the overall well-being of family members that remain unaffected carriers of the condition and protective factors that have been identified in similar research that may help them cope and adjust to the disability that has entered the lives, not just of the individual affected, but the entire family and how such an acquired disability impacts or changes the family dynamic and family system.

A Brief History of LHON

Leber's Hereditary Optic Neuropathy, LHON, was first discovered in 1871 by German ophthalmologist Theodore Leber as a disease that caused central vision loss in young adult males. Leber was unable to confirm the exact cause, but assumed the condition was an X-linked hereditary disease after studying four different pedigrees (Sadunet al., 2002). It is characterized by bilateral, painless, sub-acute visual failure that develops during young adult life (Makey, Oostra, & Rosenberg, 1996; Newman, 2005; Sadunet al., 2002). Males are four times more likely

(50% to 10%) than females to be afflicted (Newman & Biousse, 2004; Shaefer et al., 2006).

Affected individuals usually experience typical 20/20 vision throughout life until blurring begins to occur in the central visual field in one eye. The course continues with a bilateral loss of vision four to eight weeks following onset (Sadun et al., 2002). While these are the symptoms of the classic LHON case, exceptions do exist. For example, in 25% of affected individuals, vision loss is bilateral from the onset of symptoms (Newman, 1997). This optic neuropathy has severe implications for diagnosed patients as many are left only able to count fingers or see shadows following the termination of the acute phase.

After the acute phase is over, the optic disc becomes atrophic. Noteworthy improvements in vision are rare after this point with disease progression and most individuals qualify for registration as legally-blind (20/200 vision or less in both eyes). It is essential to note that the acute phase only lasts 6-12 months, making the painless progression of vision loss extremely rapid. This is a unique feature of LHON, as most vision diseases (e.g. Retinitis pigmentosa, Macular Degeneration, and Glaucoma) are slow moving and cause decreases in vision over a long period of time, sometimes many years. Some individuals with LHON also have been observed to develop a condition known as "LHON-Plus." It affects more than just the optic nerve and can lead to symptoms similar to multiple sclerosis (Newman & Biousse, 2004).

Leber's Hereditary Optic Neuropathy is an extremely rare genetic disease. While clinical impressions can lead to a preliminary diagnosis, a DNA test is usually required for confirmation. LHON is caused by mutations in the mitochondrial DNA (mtDNA). Specifically, three primary point mutations in the mitochondria are responsible for 95% of cases. These three mutations in the mitochondrial genome are: m.3460G>A, m.11778G>A, and m.14484T>C (Wallace, 1994; Harding, Sweeney, & Gowan, 1995; Sadun et al., 2002). Currently, there is no cure or effective

evidence-based treatment for LHON other than experimental clinical trials. Therefore, many individuals and their families are left to manage the disease and the associated clinical, social, and humanistic manifestations (e.g., loss of function, motor vehicle driving, capability to read normal sized print, etc.) independently.

There are a number of societal manifestations for individuals considered legally-blind. Currently, 75% of individuals living in the United States that are registered as legally-blind are unemployed (Scott et al., 1999). If such issues are similar for individuals affected with vision loss symptoms due to LHON, this can put a major financial strain on the family, most especially with the current hard economic times facing most individuals and families.

Statement of the Problem

Dominating the social science literature is a solid amount of empirical research examining the psychological well being of individuals affected with chronic illness and acquired disability, including a small subset on visual impairment. However, articles in peer reviewed journals on the impact that disability, and more specifically, genetic disease, has on the family dynamic is extremely sparse. Leber's Hereditary Optic Neuropathy (LHON), while an extremely rare genetic disease, is one of the most common hereditary optic neuropathies and one of the most common mitochondrial diseases. Many neuro-ophthalmologists that diagnose patients with LHON have noted their observations of the psychological stress present in both mothers and siblings of the affected LHON patient, but no empirical data has been collected on this likely reality for such family members.

Background

A variety of research studies have utilized validated and reliable instruments to measure the psychological well being of mothers of children with disabilities over the last forty years.

Within the last twenty years, this avenue of research has expanded its lens and focused on sibling relationships and nondisabled siblings. Such work is especially important when assessing mothers and siblings of a family member with a maternally-inherited genetic disease. Mothers that have knowledge that they have transmitted genetic disease to their children experience heightened levels of depression, guilt, and blame. Such guilt can have a severe and significant impact on the overall psychological well being of mothers, impacting the overall family dynamic as well as the marital relationship as blame commonly occurs. While siblings of children with disabilities have shown both positive and negative reactions, no current studies exist that have taken heritability into account. Siblings with prior knowledge or newfound knowledge now that their sibling has been diagnosed with a genetic disease such as LHON, could experience a great deal of psychological stress with the awareness that they could go through a similar process at any moment as well. A thorough literature review has revealed two overwhelming protective factors in family disability research: spirituality and perceived social support. These factors have a moderating effect on how mothers and siblings of disabled family members have coped in regard to other genetic diseases. This current study sought to determine if such protective factors also exist for mothers and siblings of an individual diagnosed with Leber's Hereditary Optic Neuropathy.

Purpose

The purpose of this exploratory study was to attempt to uncover the psychological well being of mothers and siblings of an individual diagnosed with and affected by symptoms of LHON. Additionally, this study determined the impact of possible protective factors (spirituality and perceived social support) on well being that, ultimately, may have major implications for clinical practice and the need for integrative care among professionals of various disciplines. If

such data can shed light on the need for counseling and psychological services among this small community of family members affected by LHON throughout the world, it will enable enhanced coordinated care among neuro-ophthalmologists and mental health practitioners. This study also provided information on the unique experience mothers and siblings go through during this very difficult and challenging time of life adjustment.

Limitations of Existing Studies

Perhaps the strongest limitation of existing research is the very paucity of studies addressing LHON, associated factors, and potential psychological impact. While the field of psychology, both in research and practice, has been a pioneer in the arenas of social justice and multicultural competence, disability research is often not as well addressed. While the research that has been published is quite rich, it is quantitatively less voluminous than other specialties within the field.

Regarding limitations, it is crucial to note that Leber's Hereditary Optic Neuropathy is something of a mystery in and of itself (Sadun et al., 2002). While initially discovered in 1871, an additional 117 years were required before researchers uncovered that it was a disease caused by specific mutations in the mitochondrial genome (Yu-Wai-Man et al., 2008; Sadun et al., 2002; Newman & Bioussé, 2004). Since the discovery by Wallace (1988) and colleagues, the study of mitochondrial disease and its causes and symptoms has picked up rapidly and furiously (Newman, 2009). That being said, general knowledge of the disease itself remains quite limited. It is not understood, for example, why some carriers become affected and some do not, why more men become affected than women, why some people spontaneously recover their sight but most do not, and what risk factors are implicated (Newman, 2009). Among the research that is available, much is contradictory and potentially confounding due to gender stereotypes (Sadun,

Morgia, & Carelli, 2010). From a psychological perspective, this alone can have significant implications, where those with the genetic mutation, unaware of risk factors or protective factors, could experience psychological stress. Additionally, while mothers may feel guilty for transmitting this genetic disease to their offspring, they may also feel equally guilty about not knowing whether they unintentionally exposed that child to one of the hypothesized risk factors at some point in their life (e.g. smoke, certain medications, toxins). Surprisingly, only one article (Newman, 2009) has ever mentioned psychological stress as being a potential risk factor in the onset of LHON vision loss symptoms. Other than one published study (Kirkman et al., 2009), the psychological well being in LHON patients and their families has never been explored.

The psychological research also has a number of distinct limitations. For example, while there is a rich literature studying the impact of genetic disease, the majority place emphasis on the affected patient and neglect to explore curiosities about the experiences of other immediate family members (Lillie, Clifford, & Metcalfe, 2010). As stated by Stoneman (2005), historically such research has always focused on the reaction by the mother to disability. Fathers have almost been completely disregarded and research into the sibling relationship has only been addressed in the last two decades (Stoneman, 2005). Hodapp, Glidden, & Kaiser (2005) question the validity of sibling disability studies, considering whether positive or negative results in the quality of the relationship are due to the disability in the family or simply typical for development based on the age of the siblings. Leonard (2008), stated in a very critical review of the impact of disability on the family research that almost every study has focused on whether different stressors exist, yet no progress is being made on why they occur or how to alleviate them once they are present in the home.

In summary, the most prevalent protective factors in the literature were perceived social support and levels of spirituality. The data on perceived social support within the populations being studied in this present research is quite extensive, validated, and strong. Various methodologies have been used and studies on visual impairments and social support have been published (Cimarolli&Boerner, 2005). Guerette& Miller-Smedema (2011) explained that perceived social support has been a cornerstone of inquiry in the disciplines of rehabilitation psychology and rehabilitation counseling. Conversely, spirituality research has a unique set of challenges. Considerable research on spirituality within the community of persons with disabilities has made an intentional effort to be inclusive of individuals from diverse cultures and backgrounds, both racially and ethnically as well as with regard to socioeconomic status (Kimet al., 2011; Borum, 2008). The one downside to the current studies on spirituality as a protective factor with populations of individuals with disabilities and their families is that it is almost entirely qualitative research. Qualitative research is highly effective, informative, and useful both in the field of science and to consumers of research. Qualitative research would be enhanced, however, with additional quantitative studies.

Research Questions

There were six research questions that were the focus of this study. The first research question asked, “What is the overall psychological well being of mothers of children affected by vision loss symptoms secondary to Leber’s Hereditary Optic Neuropathy?” This assessed the overall psychological well being of mothers that transmitted this genetic disease to their affected children. The second research question was, “Does spirituality impact psychological well being among mothers of children with LHON related vision loss?” The third research question asked, “Does Perceived Social Support impact psychological well being among mothers of children

with LHON related vision loss?” Questions two and three sought to explore potential protective factors that may help mothers of LHON affected children cope with the adjustment to the diagnosis. Questions four through six shifted from the psychological well being of the mother to that of the siblings. The fourth research question asked, “What is the overall psychological well being of the siblings of people affected by vision loss symptoms secondary to LHON?” while question five was, “Does spirituality impact psychological well being among siblings of people with LHON related vision loss?” and question six asked, “Does Perceived Social Support impact psychological well being among siblings of people with LHON related vision loss?” As discussed in this review, siblings of individuals diagnosed with LHON were assessed for the same psychological response as well as the same potential protective factors as mothers.

Statement of Hypotheses

There were six hypotheses for this study, based on the six research questions listed in the previous section of this chapter. The first hypothesis in this study stated that, “Mothers of a child affected by vision loss symptoms due to Leber’s Hereditary Optic Neuropathy (LHON) will have significantly higher levels of psychological distress, interpersonal problems, and social role dysfunction than a normative sample.” The second hypothesis in the study stated, “There will be a significant, positive relationship between spirituality and psychological well being among mothers of a child with LHON related vision loss” and the third hypothesis stated that “There will be a significant, positive relationship between perceived social support and psychological well being among mothers of a child with LHON related vision loss.” The fourth hypothesis in the study was that “Siblings of a person affected by vision loss symptoms due to LHON will have significantly higher levels of psychological distress, interpersonal problems, and social role dysfunction than a normative sample.” The fifth hypothesis in the study stated, “There will be a

significant, positive relationship between spirituality and psychological well being among siblings of a person with LHON related vision loss” and the sixth hypothesis stated, “There will be a significant, positive relationship between perceived social support and psychological well being among siblings of a person with LHON related vision loss.” As was the case with the six research questions, the final three questions in regard to siblings are replicated hypotheses to the researcher’s expectations about mothers.

Operational Definitions and Terms

The following terms have been defined for preparation and clarification, as they are mentioned throughout the literature review of the dissertation.

Mitochondrial DNA. Mitochondrial DNA is separate from DNA inherited from the nuclear genome. It has been determined that mitochondrial DNA is exclusively maternally-inherited. The main purpose of mitochondria is to produce ATP, the energy of the cell. It is for this reason that mitochondria are labeled, “the powerhouse of the cell” by researchers and geneticists. The mitochondria oxidize glucose, and the energy given off from this process leads to cellular respiration. Therefore, the role of mitochondria, and mitochondrial DNA, is extremely important for various systems throughout the body. Due to its reputation as a provider of energy to the cells, it has been theorized that mitochondria play a key role in the human aging process and perhaps even in the onset of age-related illnesses and health conditions.

Mitochondrial Disease. Dysfunction in mitochondrial DNA leaves an individual susceptible to various mitochondrial diseases. These diseases range from diabetes, optic neuropathy, to various other neuropathies affecting the muscular system. Mutations in particular genes of the mitochondrial DNA are responsible for this vulnerability. Since there

are mitochondria in every cell of the body, except red blood cells, mutations will express themselves in unique and specific ways.

Genetic Point Mutations. Scientific advancements and technology have allowed for genetic testing that can now determine the exact gene, in the case of LHON in the mitochondrial DNA, that is mutated and causing symptoms to affected carriers of a particular disease. Genetic point mutations m.3460G>A, m.14484T>C, and m.11778G>A are responsible for 95% of all cases of LHON. While they cause the same symptoms, these genes are quite different as the 11778, although the most common, also holds the least likelihood for spontaneous visual recovery (4%), whereas 14484 (70%) and 3460 (40%) have better odds of visual recovery.

Optic Neuropathy. An optic neuropathy is damage or death of optic nerve cells, fibers, or neurons. Damage or optic nerve death has a direct physical result: loss of vision. This loss of vision usually starts with loss of color vision and leads to complete or partial loss of overall sight. In the case of Leber's Hereditary Optic Neuropathy, there are "classic" cases in which peripheral vision remains intact while all central vision is lost. However, no two cases of LHON are exactly alike.

Retinal Ganglion Cells. Retinal Ganglion Cells (RGC's) are neurons of the retina of the eye that receive and process visual information from photoreceptors

Bilateral Vision Loss. Bilateral Vision Loss is what classically occurs in LHON. It is the process in which vision deteriorates in one eye, and then the process is repeated in the other eye until both eyes have nearly equal loss of vision. Almost all LHON cases take on this feature, although extremely rare cases have been documented in which only one eye was affected with vision loss. Numerous research studies have attempted to understand how to keep vision loss concentrated to only one eye, but without success thus far.

Acute & Chronic Phases.The Acute phase of LHON typically lasts 6 to 12 months. It is the process described above, in which bilateral central vision loss occurs over a 4 to 8 week span in one eye, followed by the same process in the other eye. Since there is currently no cure for LHON, once vision loss has leveled off and remained at relatively the same level for an extended period of time, the acute phase has ended and an individual has advanced into the chronic stage of the disease.

Spirituality.Spirituality does not have one universal definition, but is more appropriately defined by individuals who practice. Many in the religious community consider spirituality part of their particular religious sect or organization. In contrast, people that may not label themselves as being a member of one particular religion can still be spiritual and engage in practices that include prayer, contemplation, and meditation. Spirituality is also considered to take on humanistic components that include the desire to find meaning. The connection between spirituality and psychology has gained significant momentum in recent decades with numerous validated psychological instruments measuring levels of spirituality having been created. As well, one of the core components of numerous treatment modalities has been the use of spirituality in different forms. For example, spirituality is stressed regularly as a protective factor in addictions treatment of various kinds and has become used in both research and practice by clinical health psychologists that engage in Mind/Body interventions (e.g., Mindfulness, Acceptance and Commitment Therapy, Meditation).

Perceived Social Support.Social Support is one's own perception that they have others (friends, family, or a significant other) that care for their overall well being and happiness. This term can also take on global ramifications as people perceive themselves as being part of something greater than themselves and their immediate social network, namely as members of

society as a whole. Levels of perceived social support have been measured in a variety of empirical research studies and this variable has been found to be a significant protective factor for various populations. It is essential to note that such a protective factor for majority populations is likely to be a crucial piece when examining how individuals from underserved and underrepresented groups, such as individuals with disabilities and their families, cope and adjust to various life circumstances and transitions.

Chapter II: Review of the Literature

Background of LHON

Leber's Hereditary Optic Neuropathy, LHON, was first described as a distinct clinical entity in 1871 by German ophthalmologist Theodore Leber (1840-1917). He first reported a characteristic pattern of visual loss among members of four families. These observations were later confirmed in pedigrees from different populations (Yu-Wai-Manet al., 2008; Sadun et al., 2002). While Dr. Theodore Leber did not have the luxury of access to advanced technology in 1871, he remained extremely effective in observing some of the most salient features of the disease. For instance, Leber noted that the condition directly impacted the optic nerve rather than the eye, that it was significantly more common in young adult males, and that maternal inheritance was the cause (Yu-Wai-Manet al., 2008). Very little was known or understood about mitochondrial DNA transmission 140 years ago, thus causing Leber to make the assumption that LHON was an X-linked disorder rather than a disorder caused by genetic point mutations in the mitochondrial DNA (Sadun et al., 2002).

In 1988, LHON became the first human disease proven to be caused by a point mutation (m.11778G>A) within the mitochondrial genome. This was uncovered by Wallace and colleagues (1988) at a research lab at Emory University in Atlanta, Georgia. Ninety-five percent of all LHON pedigrees are now known to harbor one of three mitochondrial point mutations: m.3460G>A, m.11778G>A, m.14484T>C (Yu-Wai-Manet al., 2008; Newman, 2005; Sadun et al., 2002). While LHON remains an extremely rare condition, it is one of the most common forms of hereditary optic neuropathy (Yu-Wai-Manet al., 2008). It results in visual morbidity among young adults. The disease is the result of mitochondrial dysfunction, from primary mitochondrial DNA (mtDNA) mutations. Additional genetic and environmental factors modulate whether the

disease expresses itself or never affects someone with a genetic mutation (Newman, 2009; Yu-Wai-Man et al., 2008; Sadun et al., 2002).

Mitochondrial disorders are a major cause of chronic illness in humans (Wallace, 1994). As stated, LHON may be the most common form of hereditary optic neuropathy seen in clinical practice, but it is often misdiagnosed as many practitioners in the field of ophthalmology remain unaware of it, most likely due to a lack of training in neuro-ophthalmology and neuro-ophthalmological diseases. The most common presenting problem in patients with LHON who seek treatment is dysfunction or deterioration of the optic nerve, a process that involves symptoms that can be commonly seen in other vision diseases as well (e.g. optic neuritis). The specific pathology in LHON is limited to a highly specialized group of cells within the eye known as the Retinal Ganglion Cells (RGC's). The degeneration of RGC's and their axons causes severe physical disability for patients that are affected, as they lose the majority, if not all, of their central vision as a direct result.

Most of the specialists that dedicate their life's work to the research and practice of LHON tend to agree that it is not a disease that discriminates. In other words, while it is an inherited condition, it also is seen worldwide, in practically every nation and racial and ethnic group. That being said, Yu-Wai-Man et al. (2008) discussed some noteworthy geographical differences in a meta-analysis of 159 families in Europe and Australia. The data revealed that mutation m.11778G>A was the most prevalent in these continents (Yu-Wai-Man et al., 2008). It is also the most prevalent in the Far East. While mutation 14484 is relatively rare, it is the most pronounced among French Canadians (Yu-Wai-Man et al., 2008). The disease, and mitochondrial DNA itself, provides an in-depth look into the ancestral roots and genetic origins of those affected via the letter of their haplotype.

With all of the advancements in genetic testing and an increased knowledge base about mitochondrial DNA, Theodore Leber still hypothesized incredibly accurately that LHON is a young adult male dominated disease. It is accepted that the typical age of onset is between 15-30 years of age, with 95% of carriers being affected prior to the age of 50. Yet, it remains very possible to lose sight at any point during the first seven decades of life (Sadun, Morgia, & Carelli, 2010). The alternative to such information is that many people go their entire lifetime with the genetic mutation, but without losing their sight. Perhaps due to past generations of misdiagnosis, or simply no prevalence in past generations, 40% of diagnosed patients have no knowledge of prior family members having Leber's Hereditary Optic Neuropathy (Yu-Wai-Manet al., 2008).

Vision loss symptoms related to LHON are a significant life adjustment. LHON is an extraordinarily rare disease, even by vision disease standards, in that vision loss occurs in young adulthood for the most part and also in that symptoms occur rapidly, painlessly, and level off within a matter of months. There are three phases of LHON.

The first phase is the pre-symptomatic phase. Most individuals are not observed during this phase, unless there is a known family history and the individual is enrolled in a clinical trial (Sadun, Morgia, & Carelli, 2010). However, subtle impairment of optic nerve function is possible during this phase (e.g. the loss of color vision).

The second phase of LHON is often extremely traumatic for individuals affected, and at the same time shocking and confusing. This phase is known as the acute phase. Carriers remain asymptomatic until they experience blurring or clouding of vision in one eye. In classic cases of LHON, visual deterioration is bilateral, meaning that the other eye becomes affected either simultaneously or sequentially after initial blurring in the first eye (Yu-Wai-Manet al., 2009;

Newman, 2005, Sadunet al., 2002). This process takes between 1-2 months. While it is extremely rare, there have been reported cases of unilateral vision loss in LHON patients (Yu-Wai-Manet al., 2008). Within 4-6 weeks after the initial onset of symptoms, visual impairment becomes quite severe. This typical characteristic of LHON vision loss causes significant, if not full, deterioration of the central vision. Other features include the initial loss of color vision and a painless experience for the eye throughout the process (Yu-Wai-Manet al., 2008). A blood test that can examine mitochondrial mutations is absolutely necessary. While in some cases swelling of the optic nerve fiber layer may signal an LHON diagnosis, it is essential to note that in 20% of cases the optic disc can look entirely normal during the acute phase (Yu-Wai-Manet al., 2008).

The third and final phase of LHON is the chronic phase. During this phase the retinal nerve fiber layer gradually degenerates and after six months, optic atrophy is a universal feature (Sadunet al., 2002). If patients are seen for treatment at this stage of the disease, it often becomes difficult to rule out other causes. Therefore, many individuals have gone through months and sometimes years of misdiagnosis. Molecular genetic testing is an absolute must to guarantee that symptoms are directly due to LHON.

The chronic phase of LHON has been noted above as the third and final stage of disease progression. That being said, it is important to note that this is not so for everyone. Important to note is visual recovery: Even several years after the initial onset of symptoms, visual recovery is observed in some patients. The mutational status of the patient has a direct relationship with whether or not one is likely to recover vision. The odds of visual recovery are smallest with the m.1778G>A mutation, the highest with the m.14484T>C mutation, and intermediate for m.3460G>A mutation (Yu-Wai-Manet al., 2008; Newman, 2005). While it remains a mystery as to why certain individuals recover vision and others never do, a large optic nerve head, subacute

features, and onset of symptoms prior to the age of 20 seem to be patterns in those that have recovered significant amounts of sight (Sadunet al., 2002).

Visual impairment and deterioration of sight in the center of the visual field is the dominant feature of LHON. However, there are additional features that professionals in the fields of medicine and mental health alike should know. Cardiac arrhythmias and neurological abnormalities such as postural tremors, peripheral neuropathy, and movement disorders have been seen more often in LHON than in control groups (Yu-Wai-Manet al., 2008). While even rarer, there are some pedigrees of LHON that have been shown to have severe neurological deficits such as spastic dystonia, ataxia, and juvenile onset encephalopathy, in addition to the optic neuropathy. The symptoms described above are now known as LHON Plus. LHON Plus has been seen in mitochondrial mutations in the United States, Holland, and Australia. Clinical and neuro-imaging features indistinguishable from multiple sclerosis (MS) are seen in a very small minority of Caucasian females with LHON mutations. Because this is such a rare minority of the LHON community, it is still not known whether this is just a chance coincidence that a certain population of patients has MS-like symptoms or if there is a direct connection between the two disorders.

When it comes to diagnosis, molecular genetic testing is the best avenue to determine whether someone has one of the three primary mtDNA LHON mutations. This testing is crucial if there is no known family history and it can also be useful as future genetic counseling can be used to determine what future generations would and would not be at risk for developing symptoms of LHON.

Only 50% of males and 10% of females with one of the three primary mutations actually develop the optic neuropathy (Kirkman et al., 2009; Newman, 2005; Sadun et al., 2002). If not everyone with the mutation is actually losing their sight, then this implies that in addition to the mutation, there are other genetic or environmental factors that cause onset of LHON (Newman, 2009). It remains a medical mystery as to why only 50% of males develop symptoms of LHON, and while the gender bias is also not completely understood, chromosomal and hormonal differences between males and females has been theorized as a potential explanation (Sadun, Morgia, Carelli, 2010).

Mitochondrial DNA has a very high mutation rate, at least 10 times that of nuclear genes (Wallace, 1994). Because all human cells contain mitochondrial DNA, these mutations can occur in all human tissues. Mutations that arise in the somatic tissues degrade cellular energy production (Wallace, 1994). Mutations which arise in the mitochondria are transmitted by females to the next generation where they can become devastating mitochondrial diseases and while the tail of the sperm does carry mitochondria, none of it ever enters the embryo (Wallace, 1994).

One of the largest longitudinal studies of an LHON pedigree occurred during the early portion of this century. During the early 2000's, a newly discovered and very large pedigree in Brazil with the primary m.11778G>A mutation was studied. This extensive family all originated from and lived in a rural region of the country. A team of international experts, including a team from the Federal University of Sao Paulo (Brazil) were part of this research. Two hundred seventy-three of the 295 family members that were still alive were examined. Epidemiological interviews emphasized possible environmental risk factors. It was uncovered that the very first family member from this pedigree had immigrated to Brazil from Verona, Italy (Sadun et al.,

2002). The overarching goal of this research was to better uncover why only some of those with the mutation develop symptoms of LHON. Some results from the research showed that consuming fruit could be a protective factor, smoking and drinking heavily seemed to increase the risk of onset, and a combination of both of those things was even more significant (Sadun et al., 2002).

Follow up data by Sadun, Morgia, & Carelli (2010) in Brazil revealed that despite various clinical trials, patients are unlikely to benefit after optic atrophy has occurred (which happens during the acute phase). Trials featuring idebenone, a combination of vitamins B12 and CoQ10 hold promise in reducing degeneration of the optic nerve and improving final outcomes. That said, this is not considered a treatment to improve or fully restore sight (Sadun, Morgia, & Carelli, 2010). Further exploration into risk factors revealed that both in a large Brazilian family with LHON as well as another study examining 402 LHON carriers with all three mitochondrial point mutations, indicated that smoking and high consumption of alcohol could trigger symptoms of vision loss (Sadun, Morgia, & Carelli, 2010; Kirkman et al., 2009). Smoke in general, not just tobacco smoking, may also trigger LHON as some reported cases have been associated with exposure to tire fires or malfunctioning stoves in Brazil (Sadun, Morgia, & Carelli, 2010). Connections have also been made with potential risk factors that include certain antibiotics and antiviral drugs (for HIV) as these are known for interfering with mitochondrial respiratory function. In regard to pharmacological interventions, vitamin supplements (e.g. vitamins B12 and CoQ10) did not prove their efficacy in LHON (Sadun, Morgia, & Carelli, 2010). Topical bimonodine was also unable to prevent involvement of the fellow eye during the initial acute phase of LHON.

In regard to additional research exploring risk factors, Kirkman et al., (2009) interviewed 402 individuals to assess for harm surrounding factors such as smoking, consumption of alcohol, trauma history, exposure to toxins, and use of other illicit drugs. The results showed that two-thirds of those affected with LHON were smokers, whereas only half of those unaffected were smokers and those affected also smoked more frequently than the unaffected half (Kirkman et al., 2009). There were no significant differences among those that drank alcohol, although the intensity was significant, as those that were affected consumed more than double the amount of drinks than the unaffected group. Most LHON research on environmental factors has been based on anecdotal reports and small case studies. Prior large scale studies also produced conflicting results (Kirkman et al., 2009). While this research is a key addition to the field of LHON research, previous warnings by Sadun et al. (2002) should be considered. Sadun et al. (2010) theorized that while significance may be given to smoking and heavy drinking, the reality that more men are affected with LHON vision loss, and more men typically smoke and drink heavily than women, could make the significance of such results confounding, or a bit questionable at the very least. The majority of LHON research does not seem to take potential confounding issues such as this, or such as cultural contexts into consideration very often when such factors could play pivotal roles in the results.

Maciel-Guerra et al. (2010) conducted quite extensive research in Brazil as well. They examined 13 unrelated patients in Brazil and found that 9 held the 11778 mutation, 2 held the 14484 mutation, and 1 held the 3460 mutation. Based on prior studies that attempted to extract geographical differences based on genetic point mutation, the diversity of these numbers speaks to the overall diversity of the Brazilian people (Maciel-Guerra et al., 2010). Unlike many other regions of Latin America, Brazil is incredibly heterogeneous in regard to race and ethnicity.

Participants in this study ranged in descent from European, African, Amerindian, Biracial, and Mesitzo (Maciel-Guerra et al., 2010).

In closing, neuro-ophthalmological research is continuing to attack the mysteries of LHON, despite conflicts between what is seen in clinical practice and what some of the general data about the disease suggests (Newman & Biousse, 2004). The more that has been learned and understood about the disease seems to have researchers seeking to finally have answers to questions that have yet to be solved (Newman & Biousse, 2004). That being said, the future is brighter than it may seem for individuals affected by LHON and their families. Research can typically move quite slowly, but that is not the case over the last 25 years with LHON. Since the discovery that LHON was a mitochondrial hereditary optic neuropathy, and one of the most common ones at that (Newman & Biousse, 2004), the research has surged forward both for LHON and for mitochondrial diseases caused by mutations. There remains a lot of knowledge to gain and much to understand further, but studies of large pedigrees and unique cases are helping the field get to that point. Families can also harbor hope in future clinical trials that focus their efforts on experimental dosages of idebenone, gene therapy trials, and potential stem cell remedies. Outside of medical cures, governmental support in terms of finances for families and scientific advancements in assistive technology are likely to make the overall quality of life and quality of relationship between individuals affected by LHON vision loss and their families significantly more manageable than in previous generations.

Mental Health of LHON-affected Individuals

To date, there has only been one empirical research study that examined the overall quality of life of individuals diagnosed with LHON that has been approved for publication

(Kirkman et al., 2009). This first formal assessment of visual disability in affected and unaffected individuals with LHON featured 402 LHON carriers – 196 affected and 206 unaffected – from 125 families that were given the Visual Function Index (VF-14). Seventy-one had mutation 3460, 270 had mutation 11778, and 61 had mutation 14484. These disparities in regard to the mutations may seem quite significant, but they are actually typical and can be generalized to the overall population of the LHON community. The mean age of onset was 27.9 years old, mean disease duration was 15.5 years with 74.5% of affected subjects being men. The results showed that the mean score on the WF-14 was 25.1 in affected patients and 97.3 in unaffected patients. For the affected, scores did not worsen based on disease duration (Kirkman, et al., 2009). Those affected with the 14484 mutation had higher VF scores than the other two mutations (Kirkman et al., 2009). This could have been a complete coincidence or it could indicate that individuals with the 14484 may be coping better, or show higher levels of confidence in their abilities because they have knowledge that the 14484 mutation has a much higher rate of potential visual recovery than the other two mutations. As well, these numbers may simply reflect the reality of this study, which was that those with the 14484 mutation were dramatically outnumbered in the overall sample of this study, by nearly five times.

Based on these results, it would appear that LHON has a severe negative impact on quality of life (Kirkman et al., 2009). The problem with this particular study is that it only looked at visual functioning between two groups, using a t-test. When comparing one group, with another group that is unaffected with the disease, quality of life scores are almost guaranteed to be higher. Therefore, this particular study, the only one published that looks at quality of life among LHON patients, may not necessarily portray the true experience of those both affected

and unaffected with the condition, as it may have been more effective to use other scales or techniques.

Reaction of Unaffected Carriers

The LHON research discussed throughout this chapter has almost exclusively focused on the experience of the diagnosed LHON patient. This makes sense as this rare disease is a unique one and needs to be understood more by those in the field that do such work. Yet, little to no mention is made of mothers, other than that the mutation in their mitochondrial DNA is what is eventually responsible for the blindness of their offspring and that they have passed this condition on to future generations of their family. There is no mention of fathers of LHON individuals whatsoever and siblings are only mentioned in regard to the necessity that they be tested and monitored closely in the event that they one day lose their sight.

Mothers. The experience of mothers has been studied quite extensively in the disability literature, both in terms of genetic transmission and in terms of parenting a child with a non-genetic disability. Yet, mothers of children affected with LHON have yet to be studied. Studying such a population could lead to an extremely diverse sample in regard to age. Since most men become affected in young adulthood, the average age of unaffected mothers at the time that this is discovered would be during mid-life, already a stressful time in regard to marital relationships, vocational choice, and comfort level financially. When individuals are diagnosed with LHON, they learn that their mother transmitted this condition to them, and that a mutated mitochondrial gene from their mother is the direct cause of their vision loss. This can put an incredible amount of psychological stress on the mother as they deal with feelings of guilt, blame, frustration, and perhaps even severe anxiety and depression.

Siblings. Sibling relationship in regard to disability research has really only picked up steam in the last few decades. Similar to research looking at the psychological well being of mothers, there is no data on the psychological well being of siblings of individuals affected with vision loss symptoms due to LHON. This is a very rare and intriguing population, as siblings become aware of the heritability of this disease, they also become aware that they, too, could convert from someone unaffected to affected at any time due to the transmission of the mutated mitochondrial DNA from their shared mother. This is likely to cause a great deal of anxiety among siblings of LHON affected individuals. There could also be some challenging family role dynamics, as siblings may find themselves taking a backseat to the overwhelming attention their affected brother or sister is getting during the very traumatic acute stage of LHON. Overall, siblings of children with disabilities have displayed both positive and negative reactions in the literature to their situation, and this may very well be a similar case for unaffected siblings within the LHON community.

Psychological Impact of:

Genetic diseases on the family. While genetic testing of mitochondrial DNA is highly important, especially in LHON, this is a disease in particular that presents many difficulties in genetic counseling (Huoponen et al., 2002). One particular ophthalmological and molecular genetics study examined one affected and three unaffected members from a family with the primary point mutation 3460. The results of this study found that the unaffected mother of the individual diagnosed with LHON had minimal mitochondrial DNA mutation in her tissues, and the expression was even more minimal in the affected individual's sister and maternal aunt, as it was nearly nonexistent (Huoponen et al., 2002). This makes genetic counseling for LHON an extremely difficult process, and at times hard to understand for patients themselves. Even with

minimal mutant mitochondrial DNA, this mother transmitted a significant amount of it to her son, who according to the study, was severely affected with vision loss (Huoponen et al., 2002). Therefore, while low levels of mutant mitochondrial DNA in the tissues may be a sign of optimism within females that want to have children, it is still the duty of the practitioner to inform patients that they will still pass the mutated gene on to their offspring, putting them at risk for severe vision loss at some point in their lives. For patients with LHON, this can be a major dilemma. Because vision loss is not a guarantee, and many carriers go their entire lifetime without being affected, women may have offspring that develop symptoms of vision loss or they may have offspring that remain unaffected (Huoponen et al., 2002). The choice to have children is one that women have to consider due to the risks involved, and unfortunately genetic counseling can inform them wisely, but does not provide them with any solid guarantees due to the unpredictability of onset associated with LHON. Sporadic cases of LHON are not uncommon at all, therefore making it very common for there to be no prior known family history when one member of the family is affected. Situations such as this can make the mother feel guilt, knowing she harbors the mutated gene in the mitochondrial DNA that passed the disease to her child (Huoponen et al., 2002).

The mode of inheritance of a genetic condition has long been considered to have both medical and psychosocial consequences for families (James et al., 2006). Surprisingly, the pool of literature on such a topic is extremely shallow, with very few empirical studies examining this important subject. In a study by James et al. (2006), 112 members of 51 families with chronic granulomatous disease were surveyed to explore the influence of mode of inheritance the disease had on parents, siblings, and the patients themselves. This condition is caused by a variety of hereditary factors and it causes a weakened immune system, making those affected very prone to

infections and conditions such as pneumonia. The survey analyzed the knowledge of inheritance and risk of transmitting it to future generations, the level of concern in regard to transmitting it to future family members, and finally feelings of guilt, blame, and stigmatization. Ninety-six members of 51 families with Duchenne muscular dystrophy and spinal muscular atrophy were also studied. Duchenne muscular dystrophy is a genetic, X-linked recessive disorder that leads to difficulty walking, breathing, poor muscle coordination, and even death. The onset for this condition is typically in boys prior to the age of five. Spinal muscular atrophy, while not X-linked, is a genetic disease due to a mutation in the nuclear genome. This condition causes neurons in the spinal cord to die, leading to mobility impairment and respiratory problems. Most children diagnosed do not survive beyond the age of 10 and the condition gets progressively worse. The overall results found that families of X-linked (maternal) diseases had a high awareness of reproductive risk. Mothers worried more about risk of transmission to future generations than did any other family members (James et al., 2006).

Interestingly, the results also uncovered that mothers of patients with X-linked diseases felt a significant amount of guilt and blamed themselves ($p < .001$). Fathers of children with X-linked disease blamed their child's mother ($p < .05$), and mothers also reported feeling more blamed by the father ($p < .001$). All family members that were carriers considered the feeling and label to be stigmatizing ($p < .05$). These results supply overwhelming evidence that genetic counselors and mental health professionals alike must give attention to the blame that occurs within the family dynamic, while still informing these families of the reproductive risks involved in transmitting inherited conditions to future generations of family members.

Cancer research also has moved into the venue of genetic diseases and genetic testing due to the reality that some forms of cancer can be predicted based on patterns and trends within

family history. While care for the patient is of the utmost importance in the medical community, little attention has been paid to the needs of families who are concerned about whether their family history of cancer is associated with an inherited genetic predisposition (Lillie, Clifford, & Metcalfe, 2010). Little to no cancer research has ever focused on such a topic, and it often does not come up in clinical practice despite the general concern from fellow family members of a diagnosed patient. Via semi-structured interviews, data was recorded in a study with 10 nurses who had worked in specialist palliative care (Lillie et al., 2010). The results of this research found that there are legitimate arguments and concerns about raising the issue of an inherited genetic predisposition if nurses are aware of a family history of cancer (Lillie et al., 2010). This suggests that nurses need to have a good deal of knowledge about genetic risk factors of cancer, so that they can relay such information to their patients, but from a mental health perspective it is also important that nurses be aware of how to have such a discussion with patients and how they can best be a support for them. This is pivotal, as nurses quite often have the most face to face contact with patients at medical centers and hospitals. The results speak to the hesitancy, and perhaps fear, that many practitioners have about opening up the discussion about the potential to inherit diseases that run in the family.

Familial Dilated Cardiomyopathy (FDC) is an example of a disease that for much of its history, similar to LHON, could not be defined as specifically genetic (Burkett & Hershberger, 2005). As with LHON, where it was initially characterized as X-linked, FDC was theorized to be genetically based, but there was never direct evidence to support such a theory. Once it was uncovered in the 1980's that this condition had a genetic basis to its symptoms and causes, it changed the viewpoint of many that had the disease (Burkett & Hershberger, 2005). There began to be increased concern and worry over both transmission and onset of symptoms that were

never considered or taken into account previously. This is pretty significant, because it brings up the great old debate about how efficacious it is to inform, when informing could cause more psychological stress than the onset of symptoms, especially in diseases such as FDC and LHON in which not every carrier of the disease actually develops the symptoms of that particular disease at any point during their lifetime.

Perhaps the most studied of all genetic conditions, from a psychological perspective, is Huntington's disease. Huntington's disease is a neurodegenerative disorder that leads to difficulties with muscle coordination, cognitive decline, and at times psychological distress (Meiser& Dunn, 2000). This disorder typically has an adult onset, and life expectancy is only about 20 years after initial onset. Any child of an affected carrier has a 50% chance of becoming affected themselves. Huntington's disease had a great deal of historical significance in the field of genetic testing. It was the very first adult onset genetic disease to offer genetic testing to its carriers. However, while this may have been seen as a medical breakthrough and a positive, a great deal of concern has been raised that predictive testing may lead to an increase in deaths by suicide among identified carriers of Huntington's disease (Meiser& Dunn, 2000). Worries such as this make research into the psychological impact of predictive testing for Huntington's disease, as well as other genetic diseases, all the more important. A review of the literature found that in the case of Huntington's disease, only 10%-20% of people at risk request testing when approached by registries of testing centers (Meiser& Dunn, 2000). Adjustment to the results of testing for Huntington's disease was found to depend more on psychological adjustment before testing than the testing result itself. The psychological impact of such genetic testing may also depend on linkage and analysis of mutation detection. Cohorts that were enrolled in mutation detection programs had higher levels of depression before and after testing, compared with

people who sought genetic testing when linkage analysis was available (Meiser& Dunn, 2000). Something that may be confounding in such research is the possibility that individuals who choose to be tested for genetic diseases such as Huntington's disease may be psychologically selected for a favorable response to testing. A comprehensive, integrative care model is essential with testing for Huntington's disease, as it would not be ethical to leave a patient with the feelings associated with their results and not provide counseling from a more therapeutic model than genetic counseling can offer. That being said, such integration is necessary for all genetic diseases, as psychological impacts can range from depression about results to anxiety about the possibility of eventually becoming affected with whatever disease the individual was tested for, if their results show that they are a carrier of a specific genetic mutation that would make them predisposed.

Parents of children with disabilities. Individuals with disabilities make up the largest minority group in the United States, representing 16% of Americans (Noonan, 2007). More and more families now have a child with a disability due to incredible advances in neonatal care, which has led to the survival of both low birth weight babies and babies born with major birth defects (Noonan, 2007). These children often develop physical or cognitive deficits as children or later on in their development. This all leads up to the reality that more attention – in clinical practice, in research, and in the policy arena – is needed for parents of children with disabilities so that they know what their resources are and so that they can advocate for necessary resources that they do not presently have. There are specific suggestions in the literature for how to deliver the diagnoses of various conditions (e.g. Down syndrome) in both a sensitive and balanced manner to the parents (Noonan, 2007).

The social science literature in regard to raising a child with a disability being stressful is quite pronounced. The specific factors that impact the family dynamic have not been explored deeply enough, however. A great deal of disability research has either focused on life satisfaction of the individual or the entire family unit (Eddy & Engel, 2008). Numerous relationship studies have found that marital difficulties are heightened if there is a child in the home with a disability (Eddy & Engel, 2008). However, some large sample studies have found both positive and negative marital relationships, if a child with a disability is in the home. Interestingly, quality of life and life satisfaction scores in families with a child that has cerebral palsy are similar to comparative groups that do not have a child with a disability in the home (Eddy & Engel, 2008). Numerous studies have been quite contradictory, as some have stated that the nature and exact type of disability impacts the mother's adjustment, whereas others have found no significant difference when examining such variables. Eddy & Engel (2008) studied a sample of 205 parents with children suffering from disabilities that included spinal cord injury, amputation, congenital limb deficiency, cerebral palsy, spina bifida, or neuromuscular disease who were receiving health care services in Western Washingtonstate. The results showed that parents of children with medical conditions that were less stable such as neuromuscular disease, spina bifida, or cerebral palsy, experienced more worry and concern than parents of children with more stable conditions, such as congenital limb deficiency or amputation. Families whose children had more "stable" conditions were more likely to have family activities be interrupted or limited (Eddy & Engel, 2008). The results significantly differed based on the type of disability.

The results of studies such as the one conducted by Eddy & Engel (2008) indicate that practitioners must create individualized treatment plans that best serve the individual and their parents as opposed to using approaches for the broad disability community.

Gupta &Singhal (2004) made the observation that the majority of research focused on the negative attitudes and adverse consequences of having a child with disabilities. There is a cognitive process that occurs when experiencing a traumatic event such as the onset of disability within the family. Positive perceptions, both about their child and about the disability their child has been diagnosed with, can help parents develop more effective coping strategies (Gupta &Singhal, 2004). It is very possible for families to feel as though life has improved following acquired disability in their child. Successful adjustment in families should not always be viewed as, or assumed to be, denial or naïve (Gupta &Singhal, 2004). Parents of children with disabilities, as other studies have shown, do go through long periods of stress, more so than parents of children without disabilities. High levels of social support from the family and community have been shown to help in coping (Gupta &Singhal, 2004). As counseling psychologists, a strengths based approach is embraced, and researchers are hoping that disability related research in regard to the family can begin to move in a similar direction.

Gupta &Singhal (2005) followed up with their desire for a more positive psychological approach by examining parents of children diagnosed with autism. They found that pre-intervention parental stress levels predict the success of early intervention programs and determine prognosis for autism. They also found that psychosocial support is highly critical to these parents (Gupta &Singhal, 2005). These services are severely lacking as most programs for children with autism are child-centered and lack a family-centered approach. This can increase stress levels on the family, as they are trying to do everything they can for their child while perhaps letting their own mental health needs go unattended. Gupta &Singhal (2005) also stressed that services in less industrialized nations are especially lacking in comparison with those available in the United States.

While much research about parents of children with disability charts out “stages of adjustment” this implies progression, something that does not necessarily apply to parents of visually impaired children, most especially in the case of parents of children with LHON. According to an article published by the Texas School for the Blind and Visually Impaired (2012), there is not necessarily a sense of “shock to acceptance” in the visually impaired community, but rather a process that is on more of a continuum. Recurring bargaining (e.g., “If I just do ___ things will be alright”) and depression are highly common. While parents typically spend more time and provide more attention to the child with the visual impairment, normalization in spite of ability status is crucial to the child with a visual impairment. The Texas School for the Blind and Visually Impaired (2012) also stressed that children that are born blind, however, should be given additional care and treatment to avoid any complications during the bonding process (e.g. not being able to make eye contact with the mother). If parents fall into this trap, it could impact development and attachment styles for the child. This makes parenting a child with a visual impairment a fine line, and one that parents cannot navigate on their own without a solid sense of community and support. Social support has been seen as a positive factor in how parents both cope personally, and how they adjust their parenting styles (Texas School for the Blind and Visually Impaired, 2012). Just as the adjustment process is on a continuum for this population, so is parenting style.

Siblings of a disabled family member. Very little literature has studied the sibling relationship in terms of disability within the family. A family systems approach has been absent in the research until recently. Unfortunately, most conceptualizations of an ideal sibling relationship involving a child with a disability are ones that do not harm the typically developing sibling (Stoneman, 2005). Despite various studies, no theoretical framework has evolved from

such a conceptualization. Another way to term the ideal sibling relationship has taken on a very European American context, stating that sibling relationships should be “warm, supportive, and free of conflict” (Stoneman, 2005). The research has shown that most siblings fail to meet these idealized standards, and the literature often focuses the reasons why on the child with the disability. This seems unfair, as even sibling relationships featuring two or more typically developing children would not achieve such conflict-free relationships. A lot of research also focuses on the negative, making the assumption that having a sibling with a disability must be a bad thing for the typically developing sibling (Stoneman, 2005). This conclusion is made on the basis that siblings of children with disabilities would have low self-concept and exhibit increased behavior problems and psychological stress. There has also been conflicting research in regard to depression and loneliness, with the limited studies finding no differences between depression in children that have siblings with disabilities and children that do not have siblings with disabilities, whereas research without a comparison group has found high levels of depression and loneliness among individuals that have a sibling with a disability. However, when looking at quality of the relationship, siblings of children with disabilities most often report positive, satisfying, nurturing, and warm relationships with their siblings. Siblings of children with disabilities often take on the dominant role, serving as teacher, manager, and helper. Such a dominant role is taken on regardless of whether the sibling is older or younger (Stoneman, 2005).

Parents play an essential role in the sibling relationship as well, as even when parents try hard not to, they often show more attention to the sibling with the disability than they do to the typically developing child (Stoneman, 2005). Differential parenting within families is hard to avoid but is something practitioners can keep parents aware of during the development of their children. Low family conflict, cohesive families, and strong marriages also improve the sibling

relationship, regardless as to whether one sibling has a disability or not. An issue with sibling research is that while it is expanding significantly, conceptual frameworks are not and this has forced many similar studies to remain quite stagnant. A lot still has to be learned about the sibling relationship when one child has a disability and the other does not.

Hodapp, Glidden, & Kaiser (2005) discussed some of the issues with studies involving siblings of children or persons with disabilities. They claim that the biggest flaw to the methodology is that most research in this undeveloped area uses small samples of convenience. They also stress that sibling studies need to expand to more siblings, stating that the majority of families have more than one nondisabled sibling and that most studies only use the nondisabled sibling that is closest emotionally to the disabled sibling or has the most contact with their disabled sibling. This could lead to results that do not match the actual family experience if only the closest emotional sibling, likely to give the most positive responses, is included in empirical research studies. Finally, Hodapp et al. (2005) address two important variables that most studies fail to touch on, the first being differences based on age and the second being cultural implications. Noting that sibling relationships are going to look different both between and within groups is extremely significant especially when comparing samples that are from individualistic versus collectivistic cultures. Age is extremely significant, typically developing siblings have relationships that change throughout development, and they look quite different in adulthood than they do in childhood when siblings are often living together or sharing a room (Hodapp et al., 2005). They are going to be equally different, and it should be noted as such, for siblings that have disability as part of their relationship.

Overall, the last fifty years has seen a shift from solely medical and therapeutic interventions for children with disabilities in the developed world to a focus on the entire family

(Leonard, 2008). This has led to more access for children with disability and de-institutionalization of children with intellectual disabilities, legislation mandating a range of advocacy education and intervention policies, and the increasing entry of these children into schools and universities. As stated previously, a constant finding in the literature has been that the mother experiences more stress and depressive symptoms than other family members. When the family faces socio-economic challenges or the child has behavioral difficulties, these symptoms are even more heightened for mothers (Leonard, 2008). The earliest sibling studies reported almost entirely negative effects on the sibling that did not have a disability. More recent literature represents little to no difference in a range of social and behavioral constructs between siblings who do and do not have a brother or sister with an intellectual or physical disability (Leonard, 2008). Contemporary empirical evidence suggests that it is the overall family functioning that is more likely to determine sibling adjustment, although financial hardship remains a variable that most highly affects family function. A research study by Leonard (2008) also found both positive and negative effects when looking at the siblings of children with Down syndrome. The positive effects were related to personality characteristics, including an increased tolerance and awareness of difference, a caring and compassionate nature, increased maturity compared to their peers, and an increased appreciation of their own health and abilities. Some negative aspects were that family outings were limited, planned recreational activities were rare, and financial restraints limited family vacations, educational opportunities, and material possessions (Leonard, 2008). Some siblings reported a sense of embarrassment and ostracism by their peers.

Protective Factors

From the perspective of a counseling psychologist, examining protective factors that could help mothers and siblings of individuals affected with LHON, as well as other genetic diseases, could be extremely empowering for the population. Knowledge of such data could enable neuro-ophthalmologists performing the diagnosis as well as psychologists and other mental health practitioners providing counseling services, to know which resources within or about the community would be most efficient for family members of individuals recently impacted by an LHON diagnosis. It could be an enhancing starting point and provide a blueprint for what the most successful path to adjustment might be, based on how others have been found to deal and cope in a positive manner.

Spirituality.Ninety-two percent of the United States population is affiliated with a religion and 96% of Americans believe in God or a universal spirit (Elliot, Kilpatrick, & McCullough, 1999). Additionally, almost three quarters of Americans claim that their approach to life is grounded in their religious or spiritual faith (Elliot et al., 1999). The United States also reports levels of religiosity and spirituality that highly outweigh other industrialized nations. Historically, care for the ill or disabled was primarily the responsibility of the religious community, particularly from churches and synagogues. Yet, religious institutions have not always been entirely kind to the physically disabled as disability has often been termed the work of evil spirits and at times the disabled have either been banned or intentionally not included in certain services within religious communities (Elliot et al., 1999).

A study by the American Psychological Association's Division of Rehabilitation Psychology (Division 22) revealed that in contrast to the general public, most psychologists are not affiliated with religious organizations and the majority of them do not believe in God. However, 79% of the psychologists in the division still considered themselves spiritual. Training

in regard to religion and spirituality for psychologists, even rehabilitation psychologists, is minimal. Individuals that viewed disability as negative, prayed more whereas others found meaning and purpose in a spiritual sense as a result of their physical disability (Elliot et al., 1999).

An individual who is told that their vision will not improve faces physical and psychological adjustment. In order to adjust to visual impairment, just as with any other disability, internal and external resources are needed by the individual. Spirituality has been found to be a strong internal resource for the visually impaired (Duquette&Baril, 2011). Spirituality enables an individual to transcend their immediate circumstances. Spirituality allows for better adjustment by filling an existential void that may arise after an event such as vision loss (Duquette&Baril, 2011). Spirituality has been shown in disability research, including visual impairments, as a way of framing the condition in a positive light for the individual.

From a rehabilitation psychology perspective, spirituality takes on a motivational role as it encourages clients and families of clients with visual impairments to find personal meaning in an ordinarily difficult experience. It has been shown to motivate such individuals to achieve objectives and make connections between their beliefs (Duquette&Baril, 2011). Empirical research has shown that spirituality is a protective factor for individuals with visual impairments as they engage in activities of daily living. Duquette&Baril (2011) suggest that at the very least during rehabilitation, spirituality and religion should be taken into account when assessing the well being of the individual and/or their family members. Adjustment to vision loss is challenging as particular tasks of daily life are no longer possible in the same way they once were. Adopting new techniques and relying more on others can be stressful for both the individual with the visual impairment as well as for their immediate family members.

Depression, anxiety, and inadequacy may develop as a result. While there is an initial period of adjustment during the initial phases of acquired disability, this does not stop as other stressful life events later on in life may be complicated by the visual impairment (Duquette&Baril, 2011).

Due to negative perceptions of others, social isolation and stigmatization are very common for those in the visual impaired community, therefore requiring more reliance on the immediate support network of the family. Spirituality can counteract the existential emptiness following such a traumatic event for the family (Duquette&Baril, 2011). Spirituality has been shown to have a moderating effect, blunting the negative impact of visual impairment on quality of life (Duquette&Baril, 2011). Additionally, a key aspect to spirituality is that it has cross-cultural implications as a potential protective factor. Various studies throughout different racial and ethnic groups have shown spirituality to be helpful in the adaptation to vision loss. Further qualitative and quantitative research has shown that more often than not, people that were not spiritual prior to an event such as the onset of visual impairment in the family increased their levels of reported spirituality and religiosity following the event (Duquette&Baril, 2011)

Greater levels of religiousness and spirituality generally lead to better physical and mental health outcomes. The Lighthouse National Survey on Vision Loss (1995) indicated that, 13.5 million Americans 45 years of age and older report significant vision loss ranging from moderate impairment to total blindness. This study sampled 99 individuals that were middle aged and 95 that were older adults. After controlling for covariates using hierarchical multiple and logistic regressions, spirituality emerged as a significant independent predictor of better adaptation. Extrinsic religiousness was actually associated with lower adaptation to vision loss scores. The study also found that individuals from Protestant backgrounds were three times more likely to report faith based support as opposed to Catholics and Jews.

The Family-Spirituality-Psychological Well Being Model was developed and tested to explore how spirituality influences psychological well being among caregivers in the context of Korean family care-giving (Kim et al., 2011). The sample consisted of 157 Korean elder-family caregiver dyads. The study showed significant correlations between elders' and caregivers' spirituality and between elders' and caregivers' psychological well being. Elders' and caregivers' spirituality significantly influenced their own psychological well being (Kim et al., 2011). Spirituality levels of caregivers' did influence the well being of the elders that they were caring for, but the same could not be said vice-versa (Kim et al., 2011). The overall findings of this study suggest that spirituality levels of caregivers should be assessed within the family to provide holistic interventions. The literature indicated that spirituality plays an important role for adults in meeting the challenges of physical and psychological well being (Kim et al., 2011). It is important to note that this particular study used a Korean sample, a group that traditionally greatly values family support and relationships. Most Korean caregivers are family members of the individual with the disability, as they take care of others from home, making this an ideal population to examine for levels of spirituality within the context of family caregiving (Kim et al., 2011). Family member caregivers of older adults reported difficulties in family relationships and financial matters, lack of leisure time, and greater caregiving burden, which contributes to increased levels of loneliness and depression for the caregiver as well as diminished well being and mental health (Kim et al., 2011). Spirituality helped family caregivers find meaning and purpose in stressful situations, sustaining their psychological well being, alleviating their caregiver burden and depression, and strengthening family relationships (Kim et al., 2011).

While there is a great deal of social science research that examines quality of life, it has almost exclusively focused on the individual until recently. More current publications are giving

credence to the quality of life within the family. Poston & Turnbull (2004) conducted focus groups and individual interviews of 187 individual family members of children with a disability. Families described the importance of spirituality in their lives and participation in religious communities. As practitioners it may be important to provide spiritually sensitive support and enhance family spiritual well being. Poston & Turnbull (2004) reported that there is too much emphasis in medicine on healing the body and not the mind. One way to change emotional states is to focus on an individual's spirituality. Spirituality has a positive effect on health outcomes and healing. Spiritual tools (e.g. prayer) have a positive effect in conjunction with medical (medication) and psychological (counseling) tools as well (Poston & Turnbull, 2004).

Spirituality is an important protective factor in disability as the reaction to a disability can often begin as anger at God for the initial onset of the disability and then turn into a tool to cope with the disability itself. All participants spoke passionately about the presence of spirituality in their family, and parents in particular discussed the importance in having faith in something greater than themselves. About half of all respondents believed that their child was a gift from God, and found meaning in the disability (Poston & Turnbull, 2004).

The incidence of disability is higher among African Americans than any other group in the United States (Borum, 2008). Low income African American parent caretakers reported feeling less overwhelmed by a child with a disability than other ethnic caretakers of equally low income (Borum, 2008). For deaf children in particular, African Americans and Latinos expressed an overall low level of stress. That being said, a great deal of research has shown that African American parents are less likely to report the impact of child disability on the family than their Latino and White counterparts (Borum, 2008). This may be due to the unique history of African Americans in the United States in regard to experiences with coping during stressful situations.

A qualitative study of 14 non-deaf African American mothers with children that were deaf was conducted by Borum (2008) using thematic interviews. Thirteen of the fourteen13 respondents talked vividly about God and spirituality when asked about what their experience was like in raising a deaf child of African descent. The themes surrounding such a discussion include the themes of God being the creator of all and that God cares for, protects, and provides for his creation, that God is the ultimate parent, and the theme that spirituality in the African American community is not just related to God, but that it is an active force within the community. The overall results show that African American parents use spirituality as a source of hope and support in raising deaf children. Spirituality serves as a crucial source for which African American parents can tap into for coping and handling the stressors of appointments, learning a new language (sign language), and dealing with social stigma (Borum, 2008).

A study by Ekas, Whitman, & Shivers (2008) of 119 mothers with at least one child 18 years or younger diagnosed with Autism also assessed for spirituality. Ninety-five percent were Caucasian and 92% were middle or upper class. They were given a survey that assessed psychological well being in addition to levels of religiosity and spirituality. Spirituality is a coping resource used by many mothers of children with autism spectrum disorder. Results of this study revealed that spirituality was associated with better positive outcomes and, to a lesser extent, lower levels of negative outcomes. Religious activities were related to more negative outcomes and lower levels of positive outcomes than spirituality (Ekas et al., 2008).

Perceived social support. While having a child with a disability poses significant risks to the parents both physically and emotionally, this differs among racial and ethnic groups based on diverse cultural norms, resources, and support available to these parents (Ha, Greenberg, & Seltzer, 2011). It is also important to note that within group differences exist among these

populations. Parents of a child with a disability experience physical health symptoms, negative effects, and poorer psychological well being than the parents of nondisabled children. A study by Ha et al. (2011) contained 48 African American parents of children with disabilities and 144 comparison group parents of nondisabled children. Results showed that having a child with a disability is associated with more somatic symptoms. That being said, with greater positive social support from friends and family, the negative consequences on the mental health of the parents were greatly reduced (Ha et al., 2011). These data suggest that engaging other family members in positive interactions with parents of children with a disability will lead to better coping.

Stachour (1998) studied the relationship between quality and quantity of social support in illness. The results showed that quality of social support had a large impact on predicted illness level whereas quantity of social support played a smaller role in the prediction of illness level. The level of social support did not have an effect under low stress, but under high stress individuals who had high levels of perceived social support were healthier than individuals with low levels of perceived social support (Stachour, 1998).

Quality of life is an overarching psychosocial outcome in rehabilitation practice. After not gaining much attention throughout the history of the literature, quality of life outcomes are now highly emphasized, as of the last thirty years. Measuring such data is especially necessary in regard to persons with disabilities and their families. Various studies on quality of life have uncovered factors that have the most positive influence in the lives of people with visual impairments. Guerette & Miller-Smedema (2011) used a sample of 199 individuals diagnosed with a visual impairment. The relationship between perceived social support and multiple indicators of well being in adults with visual impairment and their families was investigated in

their study. The results supported other research, indicating a significant correlation between social support and depressive symptoms, life satisfaction, as well as physical, psychological, economic, family, and social well being. Age and social support were found to be significantly associated with depressive symptoms in the participants. As the ages of the participants increased, depressive symptoms decreased, suggesting that older participants were able to cope better with depressive tendencies (Guerette& Miller-Smedema, 2011). However, low levels of social support were associated with higher levels of depressive symptoms. These results are comparable to other studies that found high levels of depressive symptoms of individuals with visual impairments that had low levels of perceived social support. As practitioners, this may signal the necessity to help clients form a sense of community, whether in the form of a support group or a therapeutic group.

In another study of social support, Cimarolli&Boerner (2005) explored the link between social support and working-age adults with visual impairments. The most positive support reported by participants in this study was instrumental help from family members. Underestimation of participant capabilities was the most negative type of social support (Cimarolli&Boerner, 2005). This contradicts the idea that some individuals become more motivated by being underestimated and will seek to achieve more. This correlates with other research that has shown negative support to be derailing to the person with a disability. As stated, the most positive support was from family members. While this is a wonderful thing, the study fails to expand on the toll, responsibility, or burden that such a report could have on the actual family members. The participants in the study were 154 visually impaired adults who were first time applicants at a vision rehabilitation agency. A confounding issue with this study is that the researchers did not use a validated scale of perceived social support, but rather asked two open-

ended questions about social support, both positive and negative, that the participants had received. This leads to questions about the overall validity and significance of the results.

Role of Culture in Family Coping

The discipline of counseling psychology gives extreme importance to cultural considerations and the role that culture can have in the results of research studies. The powerful portion of this literature review was that it was quite diverse in its nature. From a cultural standpoint, there was a lot of diversity in terms of type of disability. Articles were not focused solely on the most common forms of disability, but made comparisons between them. Nearly every study advocated in their limitations sections for advancements in empirical research in the disability arena, as well as for studies that go beyond looking at psychological impact, but ask and wonder why similar results continue to display themselves throughout the data of various publications.

While the spirituality literature was somewhat weak in its methodology as being almost entirely qualitative, it was by far the most diverse in regard to race and ethnicity. The spirituality research focused on Caucasians, African-Americans, Latinos, and Asian groups. This was essential, as discussing spirituality without taking into account different cultures would have been a dramatic disservice to the topic. That is stated emphatically, because spirituality means different things to different cultures and its significance to a particular population of people can vary greatly as well.

A downside to the literature, as well as the present study, is that the majority of the research did a poor job in terms of reaching underserved populations of low socioeconomic status. While it is a downside of the studies that have been conducted, it is also a reality that

those of low socioeconomic status also have less access to both medical care and mental health services. These health disparities make it much more difficult for them to contribute to the literature if they do not have the access necessary to participate. This is a systemic problem throughout the fields of medicine and psychology that must continue to be addressed, and done so much more effectively than at the present time.

A strong positive aspect of these data was the balance in age and gender. Males and females were quite equally represented and all developmental levels were covered quite broadly from children with a disability all the way through to geriatric populations and their caregivers. This applied to the present study as there was a good amount of diversity in the age range of the respondents taking part in the internet based survey.

Finally, there was nothing in the literature relating to families made up of same-sex couples. All parenting research, styles, theories, and norms were based on heterosexual and traditional marital relationships. While this generalizes to the majority of the population in the world, it is leaving out a significant group of people that have families of their own and also struggle with the same issues heterosexual couples do. Many same-sex couples may have children with disabilities, may have genetic disease as part of their lives, and may have Leber's Hereditary Optic Neuropathy as well. The literature must do a more effective job in being inclusive to all populations, as they seem to have done for age, race, gender, and religious affiliation.

Summary and Conclusions

Overall, it should be noted that the empirical research and literature in this section is quite strong considering the low number of scientific inquiries into both the rare disease of

Leber's Hereditary Optic Neuropathy as well as disability and visual impairment psychological research as a whole.

The overarching goal of this chapter was to make connections between the impact of genetic disease, specifically the unique needs of mothers and siblings of a family member with LHON, and psychological well being. Through the use of data exploring the experience of mothers and siblings of individuals affected with both genetic diseases of various sorts and physical and developmental disability in general, it is very likely that some of the same risk factors and negative consequences psychologically could occur for mothers and siblings of individuals affected by a family member diagnosed with LHON. Since this has yet to be studied, no direct research could be reviewed, but peripheral studies that were similar in nature to the classic experience of those in the LHON community were explored in this thorough literature review.

As well, some of the largest protective factors psychologically, throughout peer reviewed literature, were spirituality and perceived social support. Studies examining these protective factors in individuals and their families with disabilities, as well as visual impairment specifically, were stressed to confirm that these same protective factors are likely to exist in mothers and siblings of families with LHON.

As with any research, there are holes in some of these studies and future follow up is necessary. For starters, so much remains a mystery about LHON itself. It has yet to be confirmed as to which genetic or environmental factors directly impact the onset of symptoms, or if there necessarily are any. It also has yet to be confirmed which protective factors could prevent someone from experiencing vision loss symptoms. If, as recently theorized, depression and

anxiety pertaining to psychological stress are potential risk factors for the development of vision loss in unaffected carriers, then the desire in the present study to find protective psychological factors is crucial to mothers and their unaffected children. LHON is a disease that can cause a lot of emotions. Mothers can feel a significant amount of guilt, blame, depression, and anxiety about transmitting the condition to their legally-blind children. They can also be experiencing psychological stress that could be both heightened and complicated if other family members are blaming them for transmitting the disease as well. The quality of their marital relationship could take a negative toll on them and the quality of the relationship with their affected child could decrease based on how their child responds to the news of how they were affected with this condition.

Similar responses apply for siblings as they could deal with anxiety and fear, not knowing whether or not they could become affected with vision loss symptoms at any point in their lives.

The various empirical and review of literature publications in this chapter normalize the experience of these mothers and siblings, as similar populations have experienced the same reactions, emotions, and probable responses to such a traumatic event as the onset of acquired disability or awareness of mutation via genetic testing.

In summary, the published literature provides a great deal of relevance, both LHON research and history, and research into other genetic diseases and disabilities and their impact on the family, that this is a noteworthy and necessary population to study. Further, the present research study will contribute significantly to this field of research within, not only the disciplines of counseling and psychology, but also to neuro-ophthamological professionals who

see patients, parents, and siblings with LHON in their clinical trials, research studies, and for check up and follow up appointments on a consistent basis.

The literature makes a strong case for the need for an integrative care model where disciplines collaborate to enhance the overall well being, both physically and psychologically, of their patients. Such a model would greatly benefit members of the LHON community, whether affected with vision loss symptoms or as unaffected carriers, as everyone with the disease undergoes both classic and unique experiences that they simply cannot manage without the help of a solid social support network, internal and external motivators (e.g. spirituality), and professionals (e.g. medical and mental health care practitioners).

Chapter III: Methodology

Study Design

This study employed both causal comparative, for comparing LHON-affected groups to the normative population, and correlational, for determining relationships among variables, designs to answer the study research questions and hypotheses. The survey was administered via SurveyMonkey, a publically available internet-based survey tool.

Methodology

Procedure. Participants were recruited from a publically available database originating from the National Institutes of Health (NIH) and various LHON related websites and social media outlets (please refer to Participants, below). The principal investigator provided all participants with the internet address for the survey portal. Upon accessing the site, participants were instructed to enter their initials and their month and year of birth to begin the survey. Respondents and their data remained anonymous. This approach ensured that each participant only took the survey once and there were no duplicate accounts created. Once the link was accessed the instructions, a demographic questionnaire, and four brief ratings scales were presented. Upon completing all items, participants were then thanked for their time and the survey tool then ended the survey. There was no subsequent follow-up with participants.

Protection of human subjects. This research study and its design received Seton Hall University Institutional Review Board (IRB) approval prior to initiation. Completion of the survey served as implicit consent to participate. Deception was not used in this study and there was no debriefing. The study was not expected to have any negative consequences for participants, nor were any reported. Information transmitted from SurveyMonkey was converted into Statistical Package for Social Sciences (SPSS) file format and stored on a USB memory key,

which was kept in a locked, secure location in the principal investigator's office. This information will be stored for a minimum of three years.

Participants

Participants were both male and female, over 18 years of age, and were the mother or a sibling of an individual that was diagnosed with Leber's Hereditary Optic Neuropathy (LHON). All participants were recruited via the internet through the National Institute of Health, NIH, North American Mitochondrial Disease Consortium (<http://rarediseasesnetwork.epi.usf.edu/NAMDC/register/registry.htm>) and the LHON.org (<http://www.LHON.org>) databases. Recruitment also took place on the Leber's Hereditary Optic Neuropathy (BLIND) Facebook Page and the LHON email listserv of Yahoo.com Groups. Additionally, participants were sent an email that described the study and provided an opportunity to participate in the research. Respondents were provided the option to voluntarily take part in the study and/or to forward the recruitment note to anyone they knew that fit the study inclusion criteria. There were no penalties for not participating in the research. Volunteer participants did not receive an incentive or compensation for their participation.

Instruments

The online internet survey featured a demographic questionnaire along with four brief validated assessments that have been found to be both appropriate and useful for populations similar to the population of interest for this study.

Multidimensional Scale of Perceived Social Support (MSPSS). The Multidimensional Scale of Perceived Social Support (Zimet, Dahlem, & Zimet, 1988) is a questionnaire developed for the purpose of measuring perceived social support from family, friends, and a significant other. The MSPSS consists of 12 items on a 7-point Likert scale ranging from "very strongly

disagree” to “very strongly agree.” Four items are dedicated to measure perceived social support from one particular source and the MSPSS measures the adequacy of support from three sources: family (items 3, 4, 8, 11), friends (items 6, 7, 9, 12), and significant other (items 1, 2, 5, 10). The items are divided into factor groups relating to the source of support, with scores ranging from 1 to 7. High scores indicate high levels of perceived social support. While this scale was initially used to assess undergraduate college students at Duke University, it has been proven to be valid and reliable among many different population groups (e.g. pregnant women, African-American adults, and Mexican-American youth). In a study of 222 African-American adolescents, high internal consistency was demonstrated, and factor analysis confirmed the three subscale structures of the MSPSS (Canty-Mitchell & Zimet, 2000). Correlations with a family caring scale also supported the discriminant validity of the family subscale (Canty-Mithcell&Zimet, 2000). The implications of these studies stress the cross-cultural utility of this instrument.

Another study featured 154 undergraduate students from an urban college in the United States. The results showed good internal reliability, and the factor analysis confirmed the subscale structure of the measure (Dahlem, Zimet, & Walker, 2006). This study also found no indication that social desirability bias influenced subject responses and while social support was related to depression, it was only in participants already experiencing high levels of life stress (Dahlem, Zimet, & Walker, 2006). The three subscales of this instrument (family, friends, and significant other) were found to have strong factorial validity (Zimet, Dahlem, & Zimet, 1988). Additional research concluded that the Multidimensional Scale of Perceived Social Support had good internal test/re-test reliability as well as moderate construct validity (Zimet, Dahlem, & Zimet, 1988). Within the sample collected for the present study, high internal consistency was

displayed for the total score (Cronbach's $\alpha = .918$), the friends subscale (Cronbach's $\alpha = .904$), the family subscale (Cronbach's $\alpha = .916$), and the significant other subscale (Cronbach's $\alpha = .929$),

Outcome Questionnaire-45 (OQ-45). The Outcome Questionnaire-45 (OQ-45) is a 45-item, 5-point Likert scale that is used to assess levels of psychological stress. In general, the measure is designed to assess baseline psychological functioning across variables (internal feelings, relationship conflict, tasks of daily living, etc.). The measure also assesses common symptoms of adult psychopathology. Lambert et al., (2004) reported that the Outcome Questionnaire-45 had high internal consistency ($r = .93$) and test/re-test reliability ($r = .84$). When correlated with measures used to test psychotherapeutic outcomes, concurrent validity was reported at high to moderate ($r = .50$ to $r = .85$). This measure has also been used with various populations across a wide range of settings from outpatient clinics to inpatient hospitals. There are three subscales of the OQ-45 and they measure symptom distress, quality of interpersonal relationships, and social role functioning (Lambert et al., 2004). Doerfler, Addis, & Moran (2002) found the OQ-45 was sensitive to change over a relatively short inpatient stay. Additionally, it was found to have convergent and divergent validity among its three subscales. Within the sample of the present study, high internal consistency was found for the total score (Cronbach's $\alpha = .950$), the symptom distress subscale (Cronbach's $\alpha = .929$), the quality of interpersonal relationships subscale (Cronbach's $\alpha = .863$), and the social role functioning subscale (Cronbach's $\alpha = .761$).

Impact of Event Scale (IES). The 15-item Impact of Event Scale, or IES, (Horowitz, Wilner, & Alvarez, 1979) was originally created for the study of bereaved individuals, but has also been used for exploring the psychological impact of a variety of traumas (Sundin & Horowitz, 2002). It has been termed as effective for assessment following both

traumatic events, as well as stressful life events that are less severe in nature. Results of research examining the psychometric properties of the IES indicated that the two factor structure of the IES is stable over different types of events (Sundin & Horowitz, 2002). It was also determined that the IES could discriminate between stress reactions at different times after the event, and that the IES has convergent validity with diagnosed post-traumatic stress disorder (Sundin & Horowitz, 2002). Various psychopharmacological studies have used the IES, giving evidence to its clinical relevance. The literature shows that the IES is a useful measure of stress reactions after a range of traumatic events, and it is valuable in detecting who require treatment for the coping of such events. Thewes, Meiser, & Hickie (2001) used a sample of 480 female hereditary breast cancer patients to test the psychometric properties of the IES. The results suggest good internal consistency (Cronbach's α , $r=0.84$ to $r=0.91$), and satisfactory test-retest reliability ($r=.80$). It was also found to have good face validity and be an acceptable instrument to women at increased risk for the development of hereditary breast cancer (Thewes, Meiser, & Hickie, 2001). The two factor (intrusion and avoidance) structure was replicated by factor analysis in this study. The total scale was found to have high reliability (Cronbach's $\alpha =.895$), as were the intrusion (Cronbach's $\alpha =.904$) and avoidance (Cronbach's $\alpha =.801$) subscales. Analysis of correlation coefficients between the IES, breast cancer related events, and attitude and other standardized measures of distress and general somatic concern, provide support for the concurrent and discriminative validity of the IES among women at increased risk of developing hereditary breast cancer (Thewes, Meiser, & Hickie, 2001). This indicates that the IES would likely be an effective instrument in assessing the impact of an event in regard to the onset of other hereditary diseases and conditions, such as LHON.

Spiritual Involvement and Beliefs Scale (SIBS). The Spiritual Involvement and Beliefs Scale (SIBS) is a 26-item, uni-dimensional measure that assesses spiritual involvement, activity and beliefs, making distinctions between spirituality and religiosity (Hatch et al., 1998). It was designed to be widely applicable across religious traditions in an attempt to assess actions as well as beliefs. The instrument was initially administered to 50 family practice patients and 33 family practice educators. The validity and reliability of the instrument were evaluated and found to be good (Hatch et al., 1998). The results showed high levels of internal consistency, strong test-retest reliability, a clear four-factor structure, and a high correlation with the Spiritual Well-Being Scale (Hatch et al., 1998). Maly & Day (2001) used a sample of 300 undergraduate students, 140 male and 160 female, to examine the psychometrics of the Spiritual Involvement and Beliefs Scale. Similar to Hatch et al. (1998), the results showed the measure to have high internal consistency and strong test-retest reliability. Within the sample of the present study, the total score had similarly high internal consistency (Cronbach's $\alpha = .788$)

Statistical Analyses Plan

The present study employed an *a priori* established statistical analysis plan (SAP) to prepare the study database, summarize descriptive statistics, and test study hypotheses. The following identified the SAP for this study.

Data preparation. All participant data was exported from the SurveyMonkey tool into the Statistical Program for the Social Sciences (Version 20.0). Upon transferring data, standard data validation procedures were conducted prior to formal statistical analysis. Specifically, the Explore function within SPSS was employed to generate statistics on extreme data points, potential outliers, and missing data. In addition, the Frequency function within the SPSS analysis package was used to generate histograms and measures of skew and kurtosis in order to

understand the distribution of primary study variables and their appropriateness for parametric statistical testing.

Descriptive statistics. Prior to conducting inferential analysis, descriptive statistics in the form of frequency counts, percentages, means, and standard deviations were generated to describe participant data and aggregate performance on measures. Appropriate tables of demographics were developed, aggregated by respondent type (mother vs. sibling) to summarize the characteristics of participants in this study. Further, a descriptive table describing overall and subscale performances on each of the primary study variables was generated.

Inferential analyses. The principal statistic used for inferential hypotheses within this study was a parametric, least squares multiple regression analysis (MRA), with standard entry of variables. Specifically, psychological well being was entered as the dependent, criterion variable and measures of spirituality and social support entered as the independent, predictor variables. Prior to inspection of regression coefficients, the analysis of variance (ANOVA) comparing regression to residual variance was observed for significance. Regression coefficients were then examined and interpreted. The MRA produced beta weights, r , r^2 , and R , to describe the linear relationships between predictor and criterion variables. For the purposes of this study, alpha was customary $p < 0.05$, where only p values observed to be less than 0.05 constituted statistical significance. Numerous regression models were generated to test, individually, the subscales of the dependent, criterion variables. Regression coefficients were then appropriately summarized and tabled.

Power analysis. In order to reduce the likelihood of Type II error and optimally assess study hypotheses, an *a priori* statistical power analysis was conducted to determine the number of participants required for this study. Publically available freeware, G-power, was used for this

purpose. Assuming a customary alpha of 0.05, a medium effect size 0.15, power of 0.80, and 4 simultaneous predictors, the total number of participants required was 85. Accordingly, assuming a 35% survey response rate, the present study attempted to recruit 245 participants to generate the necessary 85 participants for analysis and ultimately a total of 117 individuals participated in the study.

Chapter IV: Results

Participants

One hundred and seventeen participants completed the survey. The demographics for the entire sample are presented in Table 1. The largest age groups were 45-55 (29.9%, $n = 35$), 25-35 (22.2%, $n = 26$), and 56-64 (22.2%, $n = 26$). The majority were female (85.5%, $n = 100$), White (88.9%, $n = 104$) and employed (63.2%, $n = 74$). More than half were married (60.7%, $n = 71$). The number of children that participants had ranged from 0 (23.1%, $n = 27$) to 5 or more (5.1%, $n = 6$). Finally, the largest group of participants resided in the Northeast United States (24.8%, $n = 29$).

The frequency and percentages for those survey questions related to LHON are presented in Table 2. Over 50% ($n = 65$) were the mother of someone with LHON and the remainder were a sibling of someone with LHON (44%, $n = 52$). Most carried the 11778 LHON mutation (61.5%, $n = 72$). The length of time since the family member lost their vision varied and ranged from less than 1 year (6.8%, $n = 8$) to over 20 years (6.8%, $n = 8$). More than 50% indicated they were concerned about losing their own sight (51.3%, $n = 60$) and used the term legally blind to describe the family member with vision loss due to LHON (49.6%, $n = 58$).

Descriptive Statistics for the Measures

The descriptive statistics for the primary variables for the sample as a whole can be found in Table 3. IES (Impact of Event Scale) scores ranged from 0 to 64 with a mean score of 28.97 ($SD = 16.58$). MSPSS (Multidimensional Scale of Perceived Social Support) total scores ranged from 24 to 84 and the mean score was 64.62 ($SD = 12.42$). MSPSS Significant Other ranged from 4 to 28 and the mean score was 22.02 ($SD = 5.49$). MSPSS Family scores ranged from 7 to 28 and the mean score was 21.73 ($SD = 4.38$). MSPSS Friend scores ranged from 4 to

28 and the mean score was 21.73 ($SD = 12.42$). OQ-45 (Outcome Questionnaire-45) scores ranged from 9 to 112 and the mean score was 53.43 ($SD = 23.80$). Analyses indicated the data were normally distributed; Skewness and Kurtosis were not a problem for these variables.

The descriptive statistics for the primary variables for mothers can be found in Table 4. IES Total scores ranged from 1 to 64 with a mean score of 29.12 ($SD = 16.60$). IES Intrusive scores ranged from 0 to 35 with a mean score of 29.12 ($SD = 17.27$). IES Avoidant scores ranged from 0 to 30 with a mean score of 11.85 ($SD = 8.22$). MSPSS total scores ranged from 24 to 84 and the mean score was 65.15 ($SD = 12.96$). MSPSS Significant Other ranged from 4 to 28 and the mean score was 22.48 ($SD = 5.38$). MSPSS Family scores ranged from 7 to 28 and the mean score was 20.67 ($SD = 5.71$). MSPSS Friend scores ranged from 8 to 28 and the mean score was 22.00 ($SD = 4.26$). OQ-45 total scores ranged from 16 to 112 and the mean score was 48.65 ($SD = 24.40$). OQ-45 Symptom Distress scores ranged from 9 to 65 and the mean score was 29.16 ($SD = 14.47$). OQ-45 Interpersonal Relations scores ranged from 0 to 28 and the mean score was 10.80 ($SD = 6.98$). OQ-45 Social Role scores ranged from 1 to 22 and the mean score was 8.68 ($SD = 5.03$). SIBS scores ranged from 46 to 97 and the mean score was 66.86 ($SD = 11.67$).

The descriptive statistics for the primary variables for siblings can be found in Table 5. IES scores ranged from 0 to 60 with a mean score of 28.78 ($SD = 16.71$). IES Intrusive scores ranged from 0 to 35 with a mean score of 13.63 ($SD = 9.45$). IES Avoidant scores ranged from 0 to 34 with a mean score of 15.15 ($SD = 8.89$). MSPSS total scores ranged from 36 to 84 and the mean score was 63.95 ($SD = 11.80$). MSPSS Significant Other scores ranged from 8 to 28 and the mean score was 21.45 ($SD = 5.62$). MSPSS Family scores ranged from 9 to 28 and the mean score was 21.10 ($SD = 5.02$). MSPSS Friend scores ranged from 4 to 28 and the mean

Table 1

Frequency and Percentages for Participants' Demographic Characteristics

Variable	<i>n</i>	%
Age		
18-24	6	5.1
25-35	26	22.2
36-44	13	11.1
45-55	35	29.9
56-64	26	22.2
65+	11	9.4
Total	117	100.0
Gender		
Male	17	14.5
Female	100	85.5
Total	117	100.0
Race/Ethnicity		
White/Caucasian	104	88.9
Black/African-American	1	.9
Latino/Hispanic	10	8.5
Asian/Pacific Islander	2	1.7
Total	117	100.0
Current Relationship Status		
Single	19	16.2
In a Relationship	17	14.5
Married	71	60.7
Divorced	4	3.4
Widowed	2	1.7
Separated	4	3.4
Total	117	100.0
Number of Children		
0	27	23.1
1	19	16.2
2	35	29.9
3	23	19.7
4	7	6.0
5 or More	6	5.1
Total	117	100.0
Current Employment Status		
Unemployed	23	19.7

Employed	74	63.2
Self Employed	11	9.4
Student	9	7.7
Total	117	100.0
Region Currently Residing In		
Northeast United States	29	24.8
Southeast United States	18	15.4
Western United States	11	9.4
Midwest United States	14	12.0
Canada	6	5.1
Latin America/Caribbean	7	6.0
Europe	16	13.7
Australia or New Zealand	14	12.0
Asia	2	1.7
Total	117	100.0

Table 2

Frequency and Percentages for Participants' LHON Characteristics

Variable	<i>n</i>	%
Relationship to the person in the Family that Lost Vision due to LHON		
I am the mother of someone with LHON	65	56.0
I am the sibling (sister/brother) of someone with LHON	52	44.0
Total	117	100.0
LHON Mutation Carried		
11778	72	61.5
14484	19	16.2
3460	8	6.8
Other	18	15.4
Total	117	100.0
Length of Time Since the Family Member Lost Their Vision		
Less than 1 year	8	6.8
1-3 years	29	24.8
4-7 years	31	26.5
8-10 years	20	17.1
11-20 years	21	17.9
Over 20 years	8	6.8
Total	117	100.0
Concerned About Losing Own Sight		
Yes	60	51.3

No	57	48.7
Total	117	100.0

Term Used to Describe the Family Member With Vision Loss Due to

LHON

Blind	20	17.1
Legally-Blind	58	49.6
Visually Impaired	32	27.4
I do not use any term	7	6.0
Total	117	100.0

Table 3

Descriptive Statistics for the Measures (Entire Sample)

Scale	<i>N</i>	Min	Max	<i>M</i>	<i>SD</i>	Skewness	Kurtosis
IES total score	113	.00	64.00	28.97	16.58	.12	-.83
IES Intrusive	113	.00	35.00	15.63	10.07	.18	-1.03
IES Avoidance	113	.00	34.00	13.35	8.65	.40	-.73
SIBS Total	108	45.00	97.00	68.00	11.65	.34	-.27
MSPSS Total	104	24.00	84.00	64.62	12.42	-.80	.45
MSPSS Significant	104	4.00	28.00	22.02	5.49	-1.14	.80
Other							
MSPSS Family	104	7.00	28.00	20.86	5.40	-.80	-.02
MSPSS Friend	104	4.00	28.00	21.73	4.38	-1.12	2.37
OQ-45 Total Score	110	9.00	112.00	53.43	23.80	.45	-.45

Note. SD = Standard Deviation, M = Mean, Standard error for Skewness is .23, Standard Error for Kurtosis is .46

Table 4

Descriptive Statistics for the Measures (Mothers Only)

Scale	<i>N</i>	Min	Max	<i>M</i>	<i>SD</i>
IES total score	62	1.00	64.00	29.12	16.60
IES Intrusive	62	.00	35.00	17.27	10.34
IES Avoidance	62	.00	30.00	11.85	8.22
MSPSS Total	58	24.00	84.00	65.15	12.96
MSPSS Significant Other	58	4.00	28.00	22.48	5.38
MSPSS Family	58	7.00	28.00	20.67	5.71
MSPSS Friend	58	8.00	28.00	22.00	4.26
OQ-45 Total Score	60	16.00	112.00	48.65	24.40
OQ-45 Symptom Distress	60	9.00	65.00	29.16	14.47
OQ-45 Interpersonal Relations	60	.00	28.00	10.80	6.98
OQ-45 Social Role	60	1.00	22.00	8.68	5.03
SIBS Total score	59	46.00	97.00	66.86	11.67

Table 5

Descriptive Statistics for the Measures (Siblings Only)

Scale	<i>N</i>	Min	Max	<i>M</i>	<i>SD</i>
IES total score	51	.00	60.00	28.78	16.71
IES Intrusive	51	.00	35.00	13.62	9.45
IES Avoidance	51	.00	34.00	15.15	8.89
MSPSS Total	46	36.00	84.00	63.95	11.80
MSPSS Significant Other	46	8.00	28.00	21.45	5.62
MSPSS Family	46	9.00	28.00	21.10	5.02
MSPSS Friend	46	4.00	28.00	21.39	4.56
OQ-45 Total Score	50	9.00	111.00	59.18	21.95
OQ Symptom Distress	50	2.00	71.00	33.14	13.80
OQ Interpersonal Relations	50	2.00	39.00	14.42	6.96
OQ Social Role	50	3.00	19.00	11.62	3.97
SIBS Total score	49	45.00	96.00	69.34	11.58

score was 21.39 ($SD = 4.56$). OQ-45 total scores ranged from 9 to 111 and the mean score was 59.18 ($SD = 21.95$). OQ-45 Symptom Distress scores ranged from 2 to 71 and the mean score was 33.14 ($SD = 13.80$). OQ-45 Interpersonal Relations scores ranged from 2 to 39 and the mean score was 14.42 ($SD = 6.96$). OQ-45 Social Role scores ranged from 3 to 19 and the mean score was 11.62 ($SD = 3.97$). SIBS scores ranged from 45 to 96 and the mean score was 69.34 ($SD = 11.58$).

Reliability

As seen in Table 6, all of the measures were reliable as reflected by the Cronbach's alpha values that were computed based on the study sample. Cronbach's alpha is a measure of internal consistency. All of the measures used in this study had computed Cronbach's alphas greater than .76. As such, they are reliable.

Hypothesis Testing

A summary of the analyses for Hypotheses 1-3 for mothers of those with LHON is presented.

Results for Hypotheses 1-3: Mothers

Results for hypothesis I. Research Question 1 was what is the overall psychological well-being of mothers of children affected by vision loss symptoms secondary to Leber's Hereditary Optic Neuropathy? The researcher's hypothesis was that mothers of a child affected by vision loss symptoms due to Leber's Hereditary Optic Neuropathy (LHON) will have significantly higher levels of psychological distress, interpersonal problems, and social role dysfunction than a normative sample. In order to test this hypothesis, a series of one-sample independent t tests were conducted.

Table 6

Cronbach's Alpha (Reliability) for all Scales

Scale	# of items	Cronbach's Alpha
SIBS total	27	.78
IES Total Score	15	.89
IES Intrusive	7	.90
IES Avoidance	8	.80
MSPSS Total Score	12	.91
MSPSS Significant Other	4	.92
MSPSS Family	4	.91
MSPSS Friends	4	.90
OQ-45 Total Score	45	.95
OQ-45-Symptom Distress	25	.92
OQ-45-Interpersonal Relations	11	.86
OQ-45-Social Role	9	.76

The mean OQ- 45 Total score for mothers was 48.65 ($SD = 24.40$) and the normative sample mean for this subscale was 45.00. A one-sample t test was used to compare the average OQ- 45 Total scores for mothers to the mean from a normative sample (see Table 7). There was a lack of a statistically significant difference between the mothers' mean score and the normative mean score of 45 ($t(59) = 1.15, p = .251$). The mean OQ- 45 Interpersonal Relations score for mothers was 10.80 ($SD = 6.98$) and the normative sample mean for this subscale was 10.00. A one-sample t test was used to compare the average OQ- 45 Interpersonal Relations scores for mothers to the mean from a normative sample (see Table 7). The results indicated a lack of a statistically significant difference between the mothers' mean score and the normative mean score of 10.00 ($t(59) = 0.88, p = .378$).

The mean OQ- 45 Social Role score for mothers was 8.68 ($SD = 5.03$) and the normative sample mean for this subscale was 10.00. A one-sample t test was used to compare the average OQ- 45 Social Role scores for mothers to the mean from a normative sample (see Table 7). The results indicated a statistically significant difference between the mothers' mean score and the normative mean score ($t(59) = -2.02, p = .047$). Mothers' average Social Role score was significantly lower than that of the normative sample.

The mean OQ- 45 Symptoms Distress score for mothers was 29.16 ($SD = 14.47$) and the normative sample mean for this subscale was 25.00. A one-sample t test was used to compare the average OQ- 45 Symptoms Distress scores for mothers to the mean of 25.00 from a normative sample (see Table 7). The results indicated a statistically significant difference between the mothers' mean score and the normative mean score ($t(59) = 2.22, p = .030$). Mothers' average Symptoms Distress score was significantly higher than that of the normative sample.

Table 7

One Sample T Test for Mothers' OQ 45 Total Scores and Subscales

	<i>N</i>	Mean	<i>SD</i>	Mean	<i>t</i>	<i>df</i>	<i>P</i>
				difference			
OQ Total Score	60	48.65	24.40	0.80	1.15	59	.251
OQ Interpersonal	60	10.80	6.98	0.80	0.88	59	.378
Relations							
OQ Social Role	60	8.68	5.03	-1.31	-2.02	59	.047*
OQ Symptoms	60	29.16	14.47	4.16	2.22	59	.030*
Distress							

* $p < .05$

Given the findings, the researcher's hypothesis that mothers of a child affected by vision loss symptoms due to Leber's Hereditary Optic Neuropathy (LHON) will have significantly higher levels of psychological distress, interpersonal problems, and social role dysfunction than a normative sample was partially supported. More specifically, mothers had lower levels of social role dysfunction than a normative sample (Mothers' average Social Role score ($M = 8.68$) was significantly lower than that of the normative sample ($M = 10.00$)). Mothers had higher levels of psychological distress than a normative sample (mothers' average Symptom Distress score ($M = 29.16$) was significantly higher than that of the normative sample ($M = 25.00$)).

Summary of results for Hypothesis 1. Research Question 1 was what is the overall psychological well-being of mothers of children affected by vision loss symptoms secondary to Leber's Hereditary Optic Neuropathy? The researcher's hypothesis was that mothers of a child affected by vision loss symptoms due to Leber's Hereditary Optic Neuropathy (LHON) will have significantly higher levels of psychological distress, interpersonal problems, and social role dysfunction than a normative sample. The researcher's hypothesis was partially supported. More specifically, mothers had lower levels of social role dysfunction than a normative sample (Mothers' average Social Role score ($M = 8.68$) was significantly lower than that of the normative sample ($M = 10.00$)), which is the opposite of what was predicted. However, mothers had higher levels of psychological distress than a normative sample (mothers' average Symptom Distress score ($M = 29.16$) was significantly higher than that of the normative sample ($M = 25.00$)).

Results for hypotheses 2-3. Research Question 2 was does spirituality impact psychological well-being among mothers of children with LHON related vision loss? The researcher's hypothesis was that there will be a significant, positive relationship between

spirituality and psychological well-being among mothers of a child with LHON related vision loss. Research Question 3 was does Perceived Social Support impact psychological well-being among mothers of children with LHON related vision loss? The researcher's hypothesis was that there will be a significant, positive relationship between perceived social support and psychological well-being among mothers of a child with LHON related vision loss. A series of regression models were calculated to test these hypotheses. The results of the regression models are summarized below. The dependent variables in one set of regression models were the IES and the IES subscales. The dependent variables in the second set of regression models were the OQ-45 and the OQ-45 subscales.

Predicting the IES for mothers of those with LHON. The first set of regression models included IES and the IES subscales as the dependent variable. The first regression model examined the relationship between the MSPSS Total Score, the SIBS, and overall IES scores (the dependent variable). The independent variables were entered into the model simultaneously. The model as a whole was not statistically significant ($F(2, 65) = .365, p = .69$ and the model only accounted for 1% ($R^2 = .01$) of the variance in the overall IES scores. The regression coefficients appear in Table 8. None of the independent variables emerged as statistically significant predictor of IES scores. Given that the overall regression model was not statistically significant; the regression coefficients were not interpreted.

Table 8

Regression Coefficients for the Model Examining the Relationship between the MSPSS Total Score, the SIBS and the Overall IES Score

Model	<i>B</i>	Std. Error	β	<i>t</i>	<i>p</i>
SIBS Total score	.095	.194	.070	.491	.625
MSPSS Total score	-.086	.176	-.069	-.487	.629

The next regression model examined the relationship between the MSPSS subscales, the SIBS, and the IES (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was not statistically significant ($F(4, 53) = .789, p = .538$ and the model only accounted for 5% ($R^2 = .05$) of the variance in the IES scores. The regression coefficients appear in Table 9. None of the independent variables emerged as statistically significant predictors of IES scores. The overall regression model was not statistically significant and the regression coefficients were not interpreted.

The next regression model examined the relationship between the MSPSS subscales, the SIBS, and the IES Intrusive Subscale (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was not statistically significant ($F(4, 53) = .957, p = .439$ and the model only accounted for 6.7% ($R^2 = .067$) of the variance in the IES Intrusive Subscale. The regression coefficients appear in Table 10. None of the independent variables emerged as statistically significant predictors of IES Intrusive Subscale scores. Given that the overall regression model was not statistically significant; the regression coefficients were not interpreted.

The next regression model examined the relationship between the MSPSS subscales, the SIBS, and the IES Avoidant Subscale (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was not statistically significant ($F(4, 53) = 1.090, p = .371$ and the model only accounted for 7.6% ($R^2 = .076$) of the variance in the IES Avoidant Subscale. The regression coefficients appear in Table 11. None of the independent variables emerged as statistically significant predictors of IES Avoidant Subscale scores. Given that the overall regression model was not statistically significant; the regression coefficients were not interpreted.

Table 9

Regression Coefficients for the Model Examining the Relationship between the MSPSS

Subscales, the SIBS, and Overall IES Score

Model	<i>B</i>	Std. Error	β	<i>t</i>	<i>p</i>
SIBS Total score	.044	.201	.032	.044	.827
MSPSS Significant	.244	.580	.082	.421	.675
Other					
MSPSS Family	-.816	.567	-.292	-1.440	.156
MSPSS Friend	.525	.590	.140	.891	.377

Table 10

Regression Coefficients for the Model Examining the Relationship between the MSPSS

Subscales, the SIBS, and the IES Intrusive Subscale Scores

Model	<i>B</i>	Std. Error	β	<i>t</i>	<i>p</i>
SIBS Total score	-.016	.125	-.019	-.129	.898
MSPSS Significant Other	-.155	.363	-.083	-.427	.671
MSPSS Family	-.420	.355	-.239	-1.186	.241
MSPSS Friend	.275	.369	.116	.745	.460

Table 11

Regression Coefficients for the model Examining the Relationship between the MSPSS

Subscales, the SIBS, and the IES Avoidant Subscale Scores

Model	<i>B</i>	Std. Error	β	<i>t</i>	<i>p</i>
SIBS Total score	.060	.099	.089	.060	.544
MSPSS Significant	.399	.285	.270	1.398	.168
Other					
MSPSS Family	-.396	.279	-.284	-1.418	.162
MSPSS Friend	.251	.290	.134	.863	.392

Predicting the OQ-45 for mothers of those with LHON. The next set of regression models included OQ-45 and the OQ-45 subscales as the dependent variable.

The first regression model examined the relationship between the MSPSS total score, the SIBS, and the overall OQ-45 scores (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was statistically significant ($F(2, 55) = 9.61, p = .00$) and the model accounted for 25% ($R^2 = .25$) of the variance in the OQ-45. The regression coefficients appear in Table 12. The OQ-45 total score can be significantly predicted by the combination of MSPSS total score and SIBS total score. Specifically, the MSPSS total score is an independent, negative, statistically significant predictor of OQ-45 total score.

The next regression model examined the relationship between the MSPSS subscales, the SIBS, and the overall OQ-45 scores (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was statistically significant ($F(4, 53) = 5.466, p = .001$) and the model accounted for 29% ($R^2 = .29$) of the variance in the OQ-45. The regression coefficients appear in Table 13. The OQ-45 total score can be predicted by SIBS total score and the three subscales of the MSPSS. Specifically, the MSPSS Family subscale narrowly missed being an independent, negative statistically significant predictor of OQ-45 total score.

The next regression model examined the relationship between the MSPSS subscales, the SIBS, and the OQ-45 Symptom Distress Subscale scores (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was statistically significant ($F(4, 53) = 5.166, p = .001$) and the model accounted for 28% ($R^2 = .28$) of the variance in the Symptom Distress Subscale of the OQ-45. The regression coefficients appear

Table 12

Regression Coefficients for the Model Examining the Relationship between the MSPSS Total Score, the SIBS Total Score, and the Overall OQ-45 Scores

Model	<i>B</i>	Std. Error	β	<i>t</i>	<i>P</i>
SIBS Total Score	.104	.256	.050	.406	.686
MSPSS Total Score	-.919	.232	-.489	-3.956	.000

Table 13

Regression Coefficients for the Model Examining the Relationship between the MSPSS

Subscales, the SIBS, and Overall OQ-45 Scores

Model	<i>B</i>	Std. Error	β	<i>t</i>	<i>P</i>
SIBS Total score	.109	.264	.053	.411	.683
MSPSS Significant	-1.266	.765	-.280	-1.655	.104
Other					
MSPSS Family	-1.309	.747	-.307	-1.751	.086
MSPSS Friend	.242	.778	.042	.312	.757

in Table 14. An examination of the regression coefficients revealed that none of the independent variables were independent statistically significant predictors of the OQ-45 Symptom Distress Subscale scores.

The next regression model examined the relationship between the MSPSS subscales, the SIBS, and the OQ-45 Interpersonal Relations Subscale scores (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was statistically significant ($F(4, 53) = 6.733, p = .00$) and the model accounted for 33% ($R^2 = .33$) of the variance in the OQ-45 Interpersonal Relations Subscale scores. The regression coefficients appear in Table 15. The Interpersonal Relations Subscale of the OQ-45 score can be predicted by SIBS total score and the three subscales of the MSPSS. Specifically, the MSPSS Family subscale and the MSPSS Significant Other subscales were negative, statistically significant predictors of the OQ-45 Interpersonal Relations Subscale scores.

The final regression model examined the relationship between the MSPSS subscales, the SIBS, and the OQ-45 Social Role Subscale scores (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was not statistically significant ($F(4, 53) = 1.455, p = .299$) and the model accounted for 9.9% ($R^2 = .099$) of the variance in the Social Role Subscale. The regression coefficients appear in Table 16. None of the independent variables emerged as statistically significant predictors of the OQ-45 Social Role Subscale scores. As such, the regression coefficients were not interpreted.

Table 14

Regression Coefficients for the Model Examining the Relationship between the MSPSS

Subscales, the SIBS, and the OQ-45 Symptom Distress Subscale

Model	<i>B</i>	Std. Error	β	<i>t</i>	<i>P</i>
SIBS Total score	.099	.159	.080	.622	.536
MSPSS Significant Other	-.685	.460	-.254	-1.489	.142
MSPSS Family	-.708	.450	-.279	-1.575	.121
MSPSS Friend	-.040	.468	-.012	-.085	.932

Table 15

Regression Coefficients for the Model Examining the Relationship between the MSPSS

Subscales, the SIBS, and the OQ-45 Interpersonal Relations Subscale Scores

Model	<i>B</i>	Std. Error	β	<i>t</i>	<i>p</i>
SIBS Total score	-.004	.073	-.007	-.053	.958
MSPSS Significant	-.426	.210	-.332	-2.026	.048
Other					
MSPSS Family	-.450	.206	-.372	-2.187	.033
MSPSS Friend	.262	.214	.161	1.225	.226

Table 16

Regression Coefficients for the Model Examining the Relationship between the MSPSS

Subscales, the SIBS, and the OQ-45 Social Role Subscale Scores

Model	<i>B</i>	Std. Error	<i>B</i>	<i>t</i>	<i>P</i>
SIBS Total score	.014	.061	.032	.220	.827
MSPSS Significant	-.155	.178	-.166	-.870	.388
Other					
MSPSS Family	-.151	.174	-.172	-.867	.390
MSPSS Friend	.020	.181	.017	.112	.911

Summary of results for hypothesis II. Research Question 2 was does spirituality impact psychological well-being among mothers of children with LHON related vision loss? The researcher's hypothesis was that there will be a significant, positive relationship between spirituality and psychological well-being among mothers of a child with LHON related vision loss. The researcher's hypothesis was rejected given that spirituality was unrelated to mothers' adjustment and psychological well-being. In summary, the SIBS Total score did not predict: the IES Total score, IES Intrusive Subscale scores, IES Avoidant Subscale scores, overall OQ-45 scores, OQ-45 Symptom Distress Subscale score, OQ-45 Interpersonal Subscale score, or the OQ-45 Social Role Subscale score, when controlling for some combination of MSPSS scores.

Summary of results for hypothesis III. Research Question 3 was does Perceived Social Support impact psychological well-being among mothers of children with LHON related vision loss? The researcher's hypothesis was that there will be a significant, positive relationship between perceived social support and psychological well-being among mothers of a child with LHON related vision loss.

The researcher's hypothesis was partially supported given that Perceived Social Support; specifically social support from the family and from significant other, were negative predictors of mothers' psychological distress. That is, as social support increased, psychological distress decreased, as was predicted. In fact: (a) The MSPSS Total score was a negative, statistically significant predictor of the overall OQ-45 scores; (b) the MSPSS Family subscale is an independent, negative statistically significant predictor of OQ-45 total score; (c) the MSPSS subscales did not predict the OQ-45 Symptom Distress Subscale score; (d) the MSPSS Family subscale and the MSPSS Significant Other subscale were independent, negative statistically

significant predictors of the OQ-45 Interpersonal Subscale score; and (e) the MSPSS Family subscale the MSPSS subscales did not predict the OQ45 Social Role Subscale score.

Perceived Social Support did not predict mothers' adjustment. The findings for adjustment indicated that: (a) the MSPSS Total score did not predict the IES Total score; (b) the MSPSS Significant Other, MSPSS Family, and MSPSS Friend Scores did not predict the IES Total score; (c) the MSPSS Significant Other, MSPSS Family, and MSPSS Friend Scores did not predict the IES Intrusive Subscale scores; and (d) the MSPSS Significant Other, MSPSS Family, and MSPSS Friend Scores did not predict the IES Avoidant Subscale scores.

Results for Hypotheses 4-6: Siblings

A summary of the analyses for Hypotheses 4-6 for siblings of those with LHON is presented next.

Results for hypothesis IV. Research Question 4 was what is the overall psychological well-being of the siblings of people affected by vision loss symptoms secondary to LHON? The researcher's hypothesis was that the siblings of a person affected by vision loss symptoms due to LHON will have significantly higher levels of psychological distress, interpersonal problems, and social role dysfunction than a normative sample. In order to test this hypothesis, a series of one-sample independent *t* tests were conducted using the siblings mean scores and the mean scores from a normative sample.

The mean OQ-45 Total score for siblings was 59.18 ($SD = 21.95$) and the normative sample mean for this subscale was 45.00. A one-sample *t* test was used to compare the average OQ-45 Total scores for siblings to the mean from a normative sample. The results indicated a statistically significant difference between the siblings' mean score and the normative mean

score ($t(49) = 4.56, p = .000$). Siblings' average OQ- 45 Total score was significantly higher than that of the normative sample.

The mean OQ-45 Interpersonal Relations score for siblings was 14.42 ($SD = 6.96$) and the normative sample mean for this subscale was 10.00. A one-sample t test was used to compare the average OQ- 45 Interpersonal Relations scores for siblings to the mean from a normative sample. The results indicated a statistically significant difference between the siblings' mean score and the normative mean score ($t(49) = 4.48, p = .000$). Siblings' average Interpersonal Relations score was significantly higher than that of the normative sample.

The mean OQ- 45 Social Role score for siblings was 11.62 ($SD = 3.97$) and the normative sample mean for this subscale was 10.00. A one-sample t test was used to compare the average OQ- 45 Social Role scores for siblings to the mean from a normative sample. The results indicated a statistically significant difference between the siblings' mean score and the normative mean score ($t(49) = 2.88, p = .006$). Siblings' average Social Role score was significantly higher than that of the normative sample.

The mean OQ- 45 Symptom Distress score for siblings was 33.14 ($SD = 13.80$) and the normative sample mean for this subscale was 25.00. A one-sample t test was used to compare the average OQ- 45 Symptom Distress scores for siblings to the mean from a normative sample. The results indicated a statistically significant difference between the siblings' mean score and the normative mean score ($t(49) = 4.17, p = .000$). Siblings' average Symptom Distress score was significantly higher than that of the normative sample.

Summary of hypothesis IV. The researcher's hypothesis was that the siblings of a person affected by vision loss symptoms due to LHON will have significantly higher levels of psychological distress, interpersonal problems, and social role dysfunction than a normative

sample. The researcher's hypothesis was supported. Siblings had higher levels of psychological distress than a normative sample; siblings' average OQ-45 Total score ($M = 59.18$) was significantly higher than that of the normative sample ($M = 45.00$) and siblings' average Symptom Distress score ($M = 33.14$) was significantly higher than that of the normative sample ($M = 25.00$). Siblings had higher levels of interpersonal problems than a normative sample; siblings' average Interpersonal Relations ($M = 14.42$) score was significantly higher than that of the normative sample ($M = 10.00$). Siblings had higher levels of social role dysfunction than a normative sample; siblings' average Social Role score ($M = 11.62$) was significantly higher than that of the normative sample ($M = 10.00$).

Results for Hypothesis V and Hypothesis VI. Research Question 5 was does spirituality impact psychological well-being among siblings of people with LHON related vision loss? The researcher's hypothesis was that there will be a significant, positive relationship between spirituality and psychological well-being among siblings of a person with LHON related vision loss. Research Question 6 was does Perceived Social Support impact psychological well-being among siblings of people with LHON related vision loss? The researcher's hypothesis was that there will be a significant, positive relationship between perceived social support and psychological well-being among siblings of a person with LHON related vision loss. A series of regression models were calculated to test these hypotheses. The results of the regression models are summarized below. The dependent variable in one set of regression models were the IES and the IES subscales. The dependent variables in the second set of regression models were the OQ-45 and the OQ-45 subscales.

Predicting the IES for siblings of those with LHON. The first regression model examined the relationship between the MSPSS Total Score, the SIBS, and overall IES scores (the

dependent variable). The independent variables were entered into the model simultaneously. The model as a whole was not statistically significant ($F(2, 43) = 1.004, p = .375$ and the model only accounted for 4% ($R^2 = .04$) of the variance in the overall IES scores. None of the independent variables emerged as statistically significant predictor of IES scores. Given that the overall regression model was not statistically significant; the regression coefficients were not interpreted.

The next regression model examined the relationship between the MSPSS subscales, the SIBS, and the IES (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was not statistically significant ($F(4, 41) = .976, p = .431$ and the model only accounted for 8% ($R^2 = .08$) of the variance in the IES. None of the independent variables emerged as statistically significant predictors of IES scores.

The next regression model examined the relationship between the MSPSS subscales, the SIBS, and the IES Intrusive Subscale (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was not statistically significant ($F(4, 41) = 1.368, p = .262$ and the model only accounted for 11% ($R^2 = .11$) of the variance in the IES Intrusive Subscale. None of the independent variables emerged as statistically significant predictors of IES Intrusive Subscale scores. Given that the overall regression model was not statistically significant; the regression coefficients were not interpreted.

The next regression model examined the relationship between the MSPSS subscales, the SIBS, and the IES Avoidant Subscale (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was not statistically significant ($F(4, 41) = .492, p = .742$ and the model only accounted for 4% ($R^2 = .04$) of the

variance in the IES Avoidant Subscale. None of the independent variables emerged as statistically significant predictor of IES Avoidant Subscale scores. Given that the overall regression model was not statistically significant; the regression coefficients were not interpreted.

Predicting the OQ-45 for siblings of those with LHON. The next set of regression models included OQ-45 and the OQ-45 subscales as dependent variables. The first regression model examined the relationship between the MSPSS total score, the SIBS, and the overall OQ-45 scores (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was statistically significant ($F(2, 43) = 8.935, p = .001$) and the model accounted for 29% ($R^2 = .29$) of the variance in the OQ-45. The regression coefficients appear in Table 17. The OQ-45 total score can be significantly predicted by the combination of MSPSS total score and SIBS total score. Specifically, the MSPSS total score is an independent, negative, statistically significant predictor of the OQ-45 total score. The next regression model examined the relationship between the MSPSS subscales, the SIBS, and the overall OQ-45 scores (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was statistically significant ($F(4, 41) = 5.909, p = .001$) and the model accounted for 36% ($R^2 = .36$) of the variance in the OQ-45. The regression coefficients appear in Table 18. The OQ-45 total score can be predicted by SIBS total score and the three subscales of the MSPSS. Specifically, the MSPSS Family subscale is an independent, negative statistically significant predictor of OQ-45 total score.

Table 17

Regression Coefficients for the Model Examining the Relationship between the MSPSS Total Score, the SIBS Total Score, and the Overall OQ-45 Scores

Model	<i>B</i>	Std. Error	β	<i>t</i>	<i>p</i>
SIBS Total Score	.192	.262	.100	.733	.468
MSPSS Total Score	-.965	.263	-.500	-3.675	.001

Table 18

Regression Coefficients for the model Examining the Relationship between the MSPSS

Subscales, the SIBS, and Overall OQ45 Scores

Model	<i>B</i>	Std. Error	β	<i>t</i>	<i>p</i>
SIBS Total score	.065	.261	.034	.250	.804
MSPSS Significant	.086	.583	.021	.148	.883
Other					
MSPSS Family	-2.080	.668	-.459	-3.115	.003
MSPSS Friend	-1.267	.714	-.254	-1.775	.083

The next regression model examined the relationship between the MSPSS subscales, the SIBS, and the OQ-45 Symptom Distress Subscale scores (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was statistically significant ($F(4, 41) = 3.746, p = .001$ and the model accounted for 26% ($R^2 = .26$) of the variance in the Symptom Distress Subscale of the OQ-45. The regression coefficients appear in Table 19. An examination of the regression coefficients revealed that MSPSS Family was an independent, negative statistically significant predictor of the OQ-45 Symptom Distress Subscale scores.

The next regression model examined the relationship between the MSPSS subscales, the SIBS, and the OQ-45 Interpersonal Relations Subscale scores (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was statistically significant ($F(4, 41) = 5.511, p = .001$ and the model accounted for 35% ($R^2 = .35$) of the variance in the OQ-45 Interpersonal Relations Subscale scores. The regression coefficients appear in Table 20. The Interpersonal Relations Subscale of the OQ-45 score can be predicted by SIBS total score and the three subscales of the MSPSS. Specifically, the MSPSS Family Subscale is a negative, statistically significant predictor of the OQ-45 Interpersonal Relations Subscale scores.

The final regression model examined the relationship between the MSPSS subscales, the SIBS, and the OQ-45 Social Role Subscale scores (the dependent variable). All the independent variables were entered into the model simultaneously. The model as a whole was statistically significant ($F(4, 41) = 5.614, p = .001$ and the model accounted for 29% ($R^2 = .29$) of the variance in the Social Role Subscale. The regression coefficients appear in Table 21.

Table 19

Regression Coefficients for the Model Examining the Relationship between the MSPSS

Subscales, the SIBS, and the OQ45 Symptom Distress Subscale

Model	<i>B</i>	Std. Error	β	<i>t</i>	<i>p</i>
SIBS Total score	-.007	.177	-.006	-.039	.969
MSPSS Significant Other	.098	.394	.039	.250	.804
MSPSS Family	-1.047	.451	-.367	-2.322	.025
MSPSS Friend	-.878	.482	-.280	-1.821	.076

Table 20

Regression Coefficients for the Model Examining the Relationship between the MSPSS

Subscales, the SIBS, and the OQ45 Interpersonal Relations Subscale Scores

Model	<i>B</i>	Std. Error	β	<i>t</i>	<i>p</i>
SIBS Total score	-.001	.082	-.001	-.009	.993
MSPSS Significant Other	-.009	.183	-.007	-.050	.961
MSPSS Family	-.679	.209	-.483	-3.242	.002
MSPSS Friend	-.311	.224	-.201	-1.389	.172

Table 21

Regression Coefficients for the Model Examining the Relationship between the MSPSS

Subscales, the SIBS, and the OQ45 Social Role Subscale Scores

Model	<i>B</i>	Std. Error	β	<i>t</i>	<i>p</i>
SIBS Total score	.073	.047	.212	1.552	.128
MSPSS Significant Other	-.003	.105	-.004	-.028	.978
MSPSS Family	-.354	.120	-.439	-2.952	.005
MSPSS Friend	-.078	.128	-.088	-.609	.546

The MSPSS Family Subscale is a negative, statistically significant predictor of the OQ-45 Social Role Subscale scores. None of the other variables were statistically significant.

Summary of results for hypothesis V. The researcher's hypothesis was that there will be a significant, positive relationship between spirituality and psychological well-being among siblings of a person with LHON related vision loss. The researcher's hypothesis was rejected given that spirituality was unrelated to siblings' adjustment and psychological well-being. In summary, the SIBS Total score did not predict the: IES Total score, IES Intrusive Subscale scores, IES Avoidant Subscale scores, overall OQ-45 scores, OQ-45 Symptom Distress Subscale score, OQ-45 Interpersonal Subscale score, or OQ-45 Social Role Subscale score.

Summary of results for hypothesis VI. The researcher's hypothesis was that there will be a significant, positive relationship between perceived social support and psychological well-being among siblings of a person with LHON related vision loss. The researcher's hypothesis was partially supported given that Perceived Social Support; specifically social support from the family, was a negative predictor of siblings' psychological distress. That is, it is positively related to well being, the coefficients are negative because of the direction of the scoring of the scales, where high scores indicate distress. In fact: (a) the MSPSS total score was a negative, statistically significant predictor of the OQ-45 total score; (b) the MSPSS Family subscale was an independent, negative statistically significant predictor of OQ-45 total score; (c) the MSPSS Family subscale was an independent, negative statistically significant predictor OQ-45 Symptom Distress Subscale score; (d) the MSPSS Family subscale was an independent, negative statistically significant predictor of OQ-45 Interpersonal Subscale score; and (e) the MSPSS Family subscale was an independent, negative statistically significant predictor of the OQ-45 Social Role Subscale score.

Perceived Social Support did not predict siblings' adjustment. The findings for adjustment indicated that: (a) the MSPSS Total score did not predict the IES Total score; the MSPSS Significant Other, MSPSS Family, and MSPSS Friend Scores did not predict the IES Total score; (c) the MSPSS Significant Other, MSPSS Family, and MSPSS Friend Scores did not predict the IES Intrusive Subscale scores; and (d) the MSPSS Significant Other, MSPSS Family, and MSPSS Friend Scores did not predict the IES Avoidant Subscale scores.

Chapter V: Discussion

The current research was undertaken to explore the psychological well-being of mothers and siblings of an individual with vision loss symptoms due to Leber's Hereditary Optic Neuropathy and whether perceived social support and spiritual involvement and beliefs served as protective factors as these family members coped with their child or sibling acquiring a disability. In this chapter, the findings of the present study will be examined and interpreted in light of the existing research. Additionally, the limitations of the study will be discussed, clinical implications will be presented, and directions for future research will be suggested.

Discussion of the Results of the Hypotheses

Psychological well-being of mothers of children affected by vision loss symptoms secondary to Leber's Hereditary Optic Neuropathy. The first hypothesis in the present study stated that, "Mothers of a child affected by vision loss symptoms due to Leber's Hereditary Optic Neuropathy (LHON) will have significantly higher levels of psychological distress, interpersonal problems, and social role dysfunction than a normative sample." The Outcome Questionnaire-45 was the instrument used to test this hypothesis as its three subscales measure psychological distress, interpersonal problems, and social role dysfunction. The mean scores of a normative sample for these three subscales were compared to the mean scores of the 65 mothers who participated in this study using a series of one-sample *t* tests. The hypothesis was partially supported. Mothers had lower levels of social role dysfunction than the normative sample, which is the opposite of what was predicted. However, mothers had significantly higher levels of psychological distress than a normative sample. These findings could be interpreted in many ways. The fact that mothers of children with vision loss symptoms due to Leber's Hereditary

Optic Neuropathy had significantly lower levels of social role dysfunction than the normative sample could be attributed to the fact that their role as a mother and caretaker now takes on a more heightened sense of meaning and purpose than it did prior to their child's loss of sight. The case could even be made that these results were the opposite of what was predicted because many of these women may feel stronger in their role as a mother than the normative sample would as they assist their child in the adjustment process that comes with acquiring a disability. However, the comfort in the maternal role is contradicted by the degree of struggling reported by the mothers. That struggle is evident in the findings of this study which showed that symptom distress scores on the Outcome Questionnaire-45 were significantly higher for the 65 mothers that participated in this study than that of the normative sample. This finding could indicate that mothers of children with LHON go through a similar process of experiencing feelings of guilt, depression, and anxiety that the literature has shown occurs for mothers of children with other genetic conditions (James et al., 2006). While the results of this hypothesis are aligned with feelings of guilt, blame, and psychological distress similar to that in the James et al. (2006) study, the fact that the hypothesis was only partially supported shows the need for future research in this area. As Eddy & Engel (2008) noted, contradictory results when researching the psychological impact on mothers of having children with disabilities may be attributed to a multitude of factors such as how stable the condition is, whether it is congenital or acquired, and the level of impairment it may cause.

The impact of spirituality on psychological well-being among mothers of children with LHON related vision loss. The second hypothesis in the present study stated, "There will be a significant, positive relationship between spirituality and psychological well-being among mothers of a child with LHON related vision loss." In order to test this hypothesis, a series of

regressions were conducted to examine the relationship between the overall scores on the Spiritual Involvement and Beliefs Scale (SIBS) of the 65 mothers who participated in this study with their scores on the Impact of Event Scale (IES) and its two subscales (IES Avoidant and IES Intrusive) as the IES measures adjustment to a traumatic event. Additionally, regressions were conducted to examine the relationship between the overall scores, as well as scores on the three subscales (Symptom Distress, Social Role Dysfunction, and Interpersonal Problems) on the Outcome Questionnaire-45 with overall scores on the Spiritual Involvement and Beliefs Scale (SIBS) as the OQ-45 measures levels of psychological well-being.

This second hypothesis was not supported given that spirituality was unrelated to mothers' adjustment and psychological well-being. In summary, the SIBS Total score did not significantly predict: the IES Total score, IES Intrusive Subscale scores, IES Avoidant Subscale scores, overall OQ-45 scores, OQ-45 Symptom Distress Subscale score, OQ-45 Interpersonal Subscale score, or the OQ-45 Social Role Subscale score.

The fact that not a single overall score or subscale score in either dependent variable displayed a significant linear relationship with total scores on the Spiritual Involvement and Beliefs Scale is important to note. This finding implies that religion or spirituality has no impact on how well or poorly mothers of children with LHON related vision loss cope.

In the literature it has been found that spirituality allows for better adjustment by filling an existential void that may arise after an event such as vision loss (Duquette&Baril, 2011). Spirituality has been shown in disability research, including visual impairments, as a way of framing the condition in a positive light for the individual. Duquette&Baril (2011) also suggest that at the very least during rehabilitation, spirituality and religion should be taken into account

when assessing the well being of the individual and/or their family members. The present study may shed light on the need to take levels of religion and spirituality into account when assessing the well-being of an individual or their family members, but the present study may also shed light on why no assumptions should be made that spiritual involvements and beliefs are a required and necessary part of the adjustment to disability process. The experience of mothers with children with vision loss symptoms due to LHON is an incredibly unique and rare one, and the lack of a relationship between distress and spirituality in this population is a reminder of the diversity that exists under the umbrella label of disability research.

One important factor to consider when examining these results is that the sample of participants in this study was extremely homogenous in terms of gender, race, and ethnicity. Duquette&Baril (2011) pointed out that there are significant differences in how religion and spirituality are viewed across cultures and the current sample in this study may not have done that diversity justice.

The impact of perceived social support on psychological well-being among mothers of children with LHON related vision loss. The third hypothesis in the present study stated, “There will be a significant, positive relationship between perceived social support and psychological well-being among mothers of a child with LHON related vision loss.” In order to test this hypothesis, a series of multiple regression analyses were conducted to examine the relationship between the overall scores on the Multidimensional Scale of Perceived Social Support (MSPSS) and the three subscales of the MSPSS (family, friends, and significant other) of the 65 mothers who participated in this study with their scores on the Impact of Event Scale (IES) and its two subscales (IES Avoidant and IES Intrusive) as the IES measures adjustment to a traumatic event. Additionally, regressions were conducted to examine the relationship between

the overall scores, as well as scores on the three subscales (Symptom Distress, Social Role Dysfunction, and Interpersonal Problems) on the Outcome Questionnaire-45 with overall scores and subscale scores on the Multidimensional Scale of Perceived Social Support (MSPSS) as the OQ-45 measures levels of psychological well-being.

This third hypothesis was partially supported given that Perceived Social Support; specifically social support from the family and from significant other, were negative predictors of mothers' psychological distress. That is, as social support increased, psychological distress decreased, as was predicted. In fact, the MSPSS Total score was a negative, statistically significant predictor of the overall OQ-45 scores, the MSPSS Family subscale is an independent, negative statistically significant predictor of OQ-45 total score, the MSPSS subscales did not predict the OQ-45 Symptom Distress Subscale score, the MSPSS Family subscale and the MSPSS Significant Other subscale were independent, negative statistically significant predictors of the OQ-45 Interpersonal Subscale score, and the MSPSS Family subscale, the MSPSS subscales did not predict the OQ-45 Social Role Subscale score.

Perceived Social Support did not predict mothers' adjustment. The findings for adjustment indicated that the MSPSS Total score did not predict the IES Total score, the MSPSS Significant Other, MSPSS Family, and MSPSS Friend Scores did not predict the IES Total score, the MSPSS Significant Other, MSPSS Family, and MSPSS Friend Scores did not predict the IES Intrusive Subscale scores, and the MSPSS Significant Other, MSPSS Family, and MSPSS Friend Scores did not predict the IES Avoidant Subscale scores.

Therefore, the data in this present study indicate that adjustment as measured by the Impact of Event Scale and its two subscales (Avoidant and Intrusive) is not enhanced by high

levels of perceived social support from family, friends or a significant other for mothers who have a child with vision loss symptoms due to LHON. However, perceived social support from family, friends, and a significant other showed significant results in the regression model when examining the levels of psychological well being of mothers of a child with vision loss symptoms due to LHON. The data in the presented study showed that the more perceived social support mothers of children with LHON experienced from their family and from a significant other, the more their levels of distress went down. This is evidence that despite having significant levels of psychological stress as shown in the results of the first hypothesis in this study, having strong social support in the environment will significantly aid in decreasing those levels of stress.

The results for this hypothesis appear to be aligned with the literature. For example, while it is true that parents of a child with a disability experience physical health symptoms, negative effects, and poorer psychological well being than the parents of nondisabled children, Ha et al. (2011) found that having a child with a disability is associated with more somatic symptoms. However, with greater positive social support from friends and family, the negative consequences on the mental health of the parents were greatly reduced (Ha et al., 2011). These data suggest that engaging other family members in positive interactions with parents of children with a disability will lead to better coping. This appears to be the case in the present study as mothers of a child with vision loss symptoms due to LHON saw their levels of psychological distress go down in the presence of perceived social support from both family members and their respective significant others.

Psychological well-being of the siblings of people affected by vision loss symptoms secondary to LHON. The fourth hypothesis in the present study stated that, “The siblings of a

person affected by vision loss symptoms due to LHON will have significantly higher levels of psychological distress, interpersonal problems, and social role dysfunction than a normative sample. The Outcome Questionnaire-45 was the instrument used to test this hypothesis as its three subscales measure psychological distress, interpersonal problems, and social role dysfunction. The mean scores of a normative sample for these three subscales were compared to the mean scores of the 52 siblings who participated in this study using a series of one-sample *t* tests.

This fourth hypothesis was supported. Siblings had higher levels of psychological distress than a normative sample; siblings' average OQ-45 Total score was significantly higher than that of the normative sample and siblings' average Symptom Distress score was significantly higher than that of the normative sample. Siblings had higher levels of interpersonal problems than a normative sample; as well as higher levels of social role dysfunction.

When examining the results of this hypothesis, it can be interpreted that siblings of a person affected by vision loss symptoms secondary to LHON have a very difficult time witnessing their brother or sister acquire a disability. Unlike mothers of a child with vision loss symptoms due to LHON from the first hypothesis of the present study, siblings do not just have significantly higher levels of symptom distress but also experience significantly higher levels of interpersonal problems and social role dysfunction. It was hypothesized that mothers may have a stronger sense of their social role as caretakers to a child with an acquired disability, but for siblings social role could become more complex as they now recognize that as an unaffected carrier, they too could lose their vision due to LHON at any point in their lives just as their affected sibling did. The fear and anxiety associated with that reality make it understandable why siblings would experience high levels of psychological distress. Interpersonal problems could be

due to feelings that cause them to withdraw, isolate, or act out as the now non-disabled family member carrying this genetic condition. Another possible explanation for these results is that many people affected by LHON are young adults, and classic behaviors of young adults involve social situations which individuals carrying LHON mutations are urged to stay away from in order to prevent the loss of their vision (e.g., alcohol use, being in the presence of smoke, etc.).

According to Stoneman (2005) a great deal of research into sibling relationships focuses on the negative, making the assumption that having a sibling with a disability must be a bad thing for the typically developing sibling. This conclusion is made on the basis that siblings of children with disabilities would have a low self-concept and exhibit increased behavior problems and psychological stress. While Stoneman (2005) notes that the literature makes such assumptions, the data from the present study, at least, indicate that such symptoms are real and very present in the siblings of people affected by vision loss symptoms due to LHON as scores are significantly higher than the normative sample in all of the domains that Stoneman (2005) mentions.

There has also been conflicting research in regard to depression and loneliness, with the limited studies finding no differences between depression in children that have siblings with disabilities and children that do not have siblings with disabilities, whereas research without a comparison group has found high levels of depression and loneliness among individuals that have a sibling with a disability. However, when examining the quality of the relationship, siblings of children with disabilities most often report positive, satisfying, nurturing, and warm relationships with their siblings. Siblings of children with disabilities often take on the dominant role, serving as teacher, manager, and helper. Such a dominant role is assumed regardless of whether the sibling is older or younger (Stoneman, 2005). In the present study, the quality of the

relationship among siblings was not measured or analyzed. However, while it has been noted throughout the literature that scholarly work in this area is very limited, the relationship when one sibling has a disability and the other does not can be even more complex when the disability is acquired rather than congenital, as such a distinction now signifies a potential for a drastic change to the relationship. Additionally, the needs of siblings in the LHON community are even more unique than anything the limited literature has considered previously. The siblings in the LHON community also face the concern that they could someday develop the same symptoms as their sibling due to the genetic risk and the course of the development of symptoms. These siblings know that the experience might someday be their own experience as well.

The impact of spirituality on psychological well-being among siblings of people with LHON related vision loss. The fifth hypothesis in the present study stated that, “There will be a significant, positive relationship between spirituality and psychological well-being among siblings of a person with LHON related vision loss.” In order to test this hypothesis, a series of multiple regression analyses were conducted to examine the relationship between the overall scores on the Spiritual Involvement and Beliefs Scale (SIBS) of the 52 siblings who participated in this study with their scores on the Impact of Event Scale (IES) and its two subscales (IES Avoidant and IES Intrusive) as the IES measures adjustment to a traumatic event. Additionally, regressions were conducted to examine the relationship between the overall scores, as well as scores on the three subscales (Symptom Distress, Social Role Dysfunction, and Interpersonal Problems) on the Outcome Questionnaire-45 with overall scores on the Spiritual Involvement and Beliefs Scale (SIBS) as the OQ-45 measures levels of psychological well-being.

This fifth hypothesis was not supported given that spirituality was unrelated to siblings’ adjustment and psychological well-being. In summary, the SIBS Total score did not predict the:

IES Total score, IES Intrusive Subscale scores, IES Avoidant Subscale scores, overall OQ-45 scores, OQ-45 Symptom Distress Subscale score, OQ-45 Interpersonal Subscale score, or OQ-45 Social Role Subscale score.

Similar to the second hypothesis of the present study which examined the impact of spirituality on mothers of a child affected with vision loss symptoms due to LHON, siblings of a person affected with vision loss symptoms of LHON also showed no improved levels of psychological well-being or adjustment based on levels of spiritual involvement and beliefs. Therefore, it is necessary to note that at least in the present study levels of spiritual involvement and beliefs did not have a significant impact on the entire sample. While existing literature seems to suggest otherwise, the literature in this arena has almost always focused on the individual with the disability rather than the family. More current publications are giving credence to the quality of life within the family. Poston & Turnbull (2004) conducted focus groups and individual interviews of family members of individuals with disabilities. Families described the importance of spirituality in their lives and participation in religious communities. Poston & Turnbull (2004) reported that there is too much emphasis in medicine on healing the body and not the mind. One way to change emotional states is to focus on an individual's spirituality. Spirituality has a positive effect on health outcomes and healing. Spiritual tools (e.g. prayer) have a positive effect in conjunction with medical (medication) and psychological (counseling) tools as well (Poston & Turnbull, 2004). Spirituality is an important protective factor in disability as the reaction to a disability can often begin as anger at God for the initial onset of the disability and then turn into a tool to cope with the disability itself. While these data are important and likely were very true for the Poston & Turnbull (2004) study, the present study did not find spiritual involvement or beliefs to be a significant predictor of psychological well-being or adjustment. Again, there could

be various factors that caused this outcome but the reality that LHON is extremely rare, is acquired rather than congenital, and leaves siblings with an awareness that they could lose their vision as well at any time in their lives, may make spirituality less of a protective factor than initially hypothesized. While a substantial body of the literature suggests that an overwhelming majority of people are either religious or spiritual, those numbers dip significantly in younger generations and most siblings of people affected by vision loss symptoms due to LHON are part of that demographic as the classic cases of this condition occur in young adulthood.

The impact of perceived social support on psychological well-being among siblings of people with LHON related vision loss. The sixth and final hypothesis in the present study stated that, “There will be a significant, positive relationship between perceived social support and psychological well-being among siblings of a person with LHON related vision loss.” In order to test this hypothesis, a series of multiple regression analyses were conducted to examine the relationship between the overall scores on the Multidimensional Scale of Perceived Social Support (MSPSS) and the three subscales of the MSPSS (family, friends, and significant other) of the 52 siblings who participated in this study with their scores on the Impact of Event Scale (IES) and its two subscales (IES Avoidant and IES Intrusive) as the IES measures adjustment to a traumatic event. Additionally, multiple regression analyses were conducted to examine the relationship between the overall scores, as well as scores on the three subscales (Symptom Distress, Social Role Dysfunction, and Interpersonal Problems) on the Outcome Questionnaire-45 with overall scores and subscale scores on the Multidimensional Scale of Perceived Social Support (MSPSS) as the OQ-45 measures levels of psychological well-being.

This sixth hypothesis was partially supported given that Perceived Social Support; specifically social support from the family, was a negative predictor of siblings’ psychological

distress. That is, it is positively related to well being, the coefficients are negative because of the direction of the scoring of the scales, where high scores indicate distress. In fact, the MSPSS total score was a negative, statistically significant predictor of the OQ-45 total score, the MSPSS Family subscale was an independent, negative statistically significant predictor of OQ-45 total score, the MSPSS Family subscale was an independent, negative statistically significant predictor of the OQ-45 Symptom Distress subscale score, the MSPSS Family subscale was an independent, negative statistically significant predictor of OQ-45 Interpersonal subscale score, and the MSPSS Family subscale was an independent, negative statistically significant predictor of the OQ-45 Social Role subscale score.

Perceived Social Support did not predict siblings' adjustment. The findings for adjustment indicated that the MSPSS Total score did not predict the IES Total score, the MSPSS Significant Other, MSPSS Family, and MSPSS Friend Scores did not predict the IES Total score, the MSPSS Significant Other, MSPSS Family, and MSPSS Friend scores did not predict the IES Intrusive subscale scores; and the MSPSS Significant Other, MSPSS Family, and MSPSS Friend scores did not predict the IES Avoidant subscale scores.

These data show many similarities to the data presented in hypothesis three of the present study. Such results can be interpreted as meaning that while the fourth hypothesis of this present study indicated that siblings have significantly higher levels of symptom distress, interpersonal problems, and social role dysfunction than a normative sample does, with high levels of perceived social support overall and more specifically from their family members, levels of symptom distress, interpersonal problems, and social role dysfunction drop drastically and significantly. Such information is crucial when it comes to a family member acquiring a genetic disability as family support can assist the coping process and proactive approaches to

treatment can be implemented and suggested. Similar to all other independent variables in the present study, perceived social support was not an accurate predictor of adjustment, at least based on scores from the Impact of Event Scale and its two subscales (IES Avoidant and IES Intrusive).

These results that perceived social support, most specifically from the family, can be a protective factor for siblings of a person affected by vision loss symptoms due to LHON align with results from previous studies that examined the relationship between social support and levels of psychological well-being of the visually impaired. For example, Guerette & Miller-Smedema (2011) researched a sample of individuals diagnosed with a visual impairment. The relationship between perceived social support and multiple indicators of well-being in adults with visual impairment and their families was investigated in their study. The results supported other research, indicating a significant correlation existed between social support and depressive symptoms, life satisfaction, as well as physical, psychological, economic, family, and social well-being. Age and social support were found to be significantly associated with depressive symptoms in the participants. As well, low levels of social support were associated with higher levels of depressive symptoms (Guerette & Miller-Smedema, 2011). These results are comparable to other studies that found high levels of depressive symptoms of individuals with visual impairments that had low levels of perceived social support. Cimarolli & Boerner (2005) explored the link between social support and working-age adults with visual impairments. The most positive support reported by participants in this study was instrumental help from family members (Cimarolli & Boerner, 2005). In summary, the data involved in the third and sixth hypotheses suggest that perceived social support is not only beneficial to the individual with the visual impairment as prior studies have noted, but that it is also a strong protective factor for

unaffected family members of the individual with the visual impairment or disability and for unaffected carriers of genetic diseases such as Leber's Hereditary Optic Neuropathy.

Clinical and Practical Implications

The overall results of the present study have significant and important clinical and practical implications for researchers, neuro-ophthalmologists, psychologists, mental health practitioners, and families.

As Sadun & Carelli (2002) noted, despite Leber's Hereditary Optic Neuropathy having been discovered over a century ago and that it was uncovered to be a hereditary condition in 1988, it remains a mysterious condition as it is still unknown why only 50% of men carrying the genetic mutation lose their sight or why an even smaller percentage of females lose vision. For this reason, various theories have been suggested as to what could trigger the onset of vision loss. Some of these have included alcohol use, smoking, or exposure to certain chemicals (Sadun & Carelli, 2002). As theorized by Newman (2009), a possible trigger could be psychological stress, as empirical data across disciplines have shown stress to lead to physical impairments of various kinds. While anecdotal and not based on empirical data, Newman (2009) theorized this could be the case in the LHON population as well. Levels of psychological distress in mothers and siblings of family members with vision loss symptoms due to LHON were significantly higher than the normative sample in this particular study. This knowledge emphasizes the need for neuro-ophthalmologists diagnosing LHON to have an awareness that psychological stress is not just a reality for the individual losing their sight, but also a challenge for unaffected carriers that could be susceptible to losing their vision at any time as well. This would enable neuro-

ophthalmologists to make proper referrals to psychologists and other mental health practitioners when they make such diagnoses.

The results of this study also point out a few important protective factors. The results showed that social support from a significant other and family members helped mothers' levels of psychological well-being and that siblings coped best when there was significant social support from the family. A great deal of the literature, as presented, suggested that the onset of a disability within a family member can negatively impact the marital relationship and the family dynamic as a whole. However, when those relationships are perceived to remain strong and intact, levels of psychological stress, social role dysfunction, and interpersonal problems, significantly decreased for both mothers and siblings of individuals with vision loss symptoms due to LHON. For psychologists and other mental health practitioners, this data is crucial as it stresses resources that can best help family members outside of counseling and psychological services to best cope during the adjustment process.

Lastly, this study has groundbreaking implications for the future direction of professional psychology as a whole. The field of counseling psychology has made a commitment to serving underserved and underrepresented groups and this data contributes to the continued need to further study rare diseases and conditions. Additionally, the emphasis on an integrative care model has become a core goal of professional psychology. Exploratory research such as the present study enables such communication between researchers and clinicians across various disciplines to occur and it illustrates the significance that physical conditions can have on psychological well-being and vice versa.

Limitations

As with any research study, the present investigation had some methodological limitations. Since Leber's Hereditary Optic Neuropathy is such a rare condition, this is a difficult population to access and recruit. Therefore, an online internet based survey was the best and most appropriate way to access a sample of mothers and siblings of LHON affected individuals. Internet survey methodologies, however, have their own limitations such as not reaching those without access to a computer or the internet. There are also tens of thousands of affected and unaffected carriers of LHON that might have access to the technology, but be unaware of the LHON internet community that includes various social networking sites (e.g., Facebook and Yahoo Groups) as well as structured and organized databases. Thus, the sample for study in this research was quite homogenous with 88.9% of participants self-identifying as White/Caucasian. While LHON does not discriminate and is featured throughout the various countries and cultures of the world, most individuals that participate in clinical trials at medical centers, as well as social science research pertaining to this disease, are Caucasian. An oversight of the principal researcher in this study was also in the Demographic Questionnaire in which the options for self-identifying as Biracial or Multiracial were omitted. This could have caused many individuals to have felt excluded from the present study or that their experience with LHON was not acknowledged in the same way as those that most closely identified with one of the four traditional racial and/or ethnic groups listed in the Demographic Questionnaire. The reality that the majority of participants in both clinical trial and social science research are Caucasian could be attributed to a variety of factors that include access to healthcare, access to transportation, or socioeconomic status. This precluded a large portion of the international LHON community

from participating in this study. Finally, this survey was offered only in English, thereby limiting recruitment from nations that do not speak English.

Another limitation of this study was the lack of attention to fathers. In the limitations of previous research section in the review of the literature chapter, it was explicitly stated that fathers are often completely disregarded in empirical studies that have examined the impact of a child's disability on the family dynamic. This does not seem to support a family systems approach. Due to the exploratory nature of this study, the primary goal was to examine how mothers feel about transmitting a condition that causes their children to become legally-blind, and how siblings feel about the possibility that this could happen to them. Since fathers do not contribute genetically to the disease, they were not studied within this research. Future studies, however, are recommended to focus on the quality of the relationship between married couples that have a child diagnosed with LHON. In addition, research might address the role the father plays, given that the majority of individuals affected with vision loss are male (four times more likely to become blind than their female counterparts).

Finally, the present study was limited in its focus on males. Given that males experience this disease much more frequently, this leaves open the possibility that unaffected siblings could be almost exclusively female. For the most part this was the case in the sample of participants for the present study as 85.5% were female.

Despite the numerous understood limitations of this study, the benefits of conducting this research are considerable relative to the methodological considerations.

Future Directions

As stated, this was an exploratory study and the first research study to examine the psychological well-being of unaffected carriers of Leber's Hereditary Optic Neuropathy (LHON). The results of the present study set the stage for future research. Future studies may benefit from a more integrative model in which psychological instruments and data are collected in person, perhaps in conjunction with clinical trials. This would enable researchers to get a more diverse sample in terms of race, ethnicity, gender, and age that could more deeply uncover the experiences of unaffected mothers and siblings who are carriers, but also individuals who have lost their sight due to LHON. As well, future studies would be best served to provide translation services or valid and reliable instruments that are multilingual in order to reach a more widespread demographic in regard to both language and geography.

The results of this study indicated that perceived social support is a significant protective factor for unaffected carriers of LHON. While spirituality was not found to be a significant protective factor in this particular study, future studies may want to examine alternative measures and instruments to better measure spirituality as it has been shown in many previous studies to positively impact coping, adjustment, and overall levels of psychological well-being among family members dealing with rare diseases, chronic illness, and disability within their family.

In addition, this was a quantitative research study. Considering this area of research is unique, rare, and relatively new, it would be equally beneficial to conduct qualitative research to hear a detailed account of the experiences of those impacted by Leber's Hereditary Optic Neuropathy either due to their own vision loss or as unaffected carriers who have seen a loved one lose their sight.

Lastly and perhaps most importantly, the present study displayed that this is a particular population in need of future research but as noted in the review of the literature there is currently no theoretical model that defines the identity development of individuals with disabilities. Similar models have had powerful influence on the clinical treatment of various other underserved, non-majority groups, and if future studies can more clearly define and provide a framework for the experience of those diagnosed with chronic illness and physical disabilities and impairments it can lead to more effective structured and evidence based counseling and psychological treatment modalities moving forward and into the future.

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Appendix A

SOLICITATION EMAIL

My name is Jonathan Dator and as someone with Leber's Hereditary Optic Neuropathy (LHON) and a Counseling Psychology Ph.D. candidate, I recognize many of the challenges of adjusting to this condition. While there is so much to explore on the experiences of families impacted by LHON, there is no published research on the psychological well being of family members of individuals diagnosed with this condition. This project aims to add knowledge to a field that can ultimately tailor current resources and create new ones for the unique needs of family members of individuals diagnosed with Leber's Hereditary Optic Neuropathy.

To make your participation as brief as possible, I have created a survey that will only take about **20 minutes** or less to complete.

If you are 18 years or older and are either an unaffected mother or sibling of someone that has lost vision due to a diagnosis of Leber's Hereditary Optic Neuropathy (LHON), I invite you to take part in this survey. As someone with LHON, I am interested in understanding the challenges that come with adjusting to this condition and how factors like social support and spiritual involvement and beliefs can aid in the process. Your participation in this study is completely voluntary. You may withdraw from the study at any time without consequence. Your anonymity will be maintained throughout all aspects of the study. Any publication of the data from this study will in no way identify you and results will be reported in combined form only. All material will be collected in the strictest confidence. Completed responses to questionnaires will be kept in a secure location and will be accessible only to myself and my academic advisor, Dr. Laura K. Palmer. The data will be stored electronically on a USB memory key and kept in a locked, secure physical setting.

Your participation provides useful information on the adjustment that occurs for the family following an LHON diagnosis. Personally, this study may also be useful to increase your self-awareness. This survey includes a demographic form, and four scales to measure how the vision loss of your child or sibling has impacted you and the role that social support and spiritual

involvement and beliefs play in adjustment. To begin the survey, click the following link:
<http://www.research.net/s/lhonstudy>

This project has been reviewed and approved by the Seton Hall University Institutional Review Board (IRB) for Human Subjects Research. Questions about the research subject's rights should be directed to the Director of the IRB at SetonHallUniversity, Dr. Mary F. Ruzicka, Ph.D. at (973)-313-6314. Thank you for your consideration to participate.

Sincerely,

Jonathan W. Dator, M.Ed.

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Appendix B

INFORMED CONSENT

Researcher's Affiliation:

This study is being conducted by Jonathan W. Dator, a doctoral candidate in the Counseling Psychology Ph.D. program in the Department of Professional Psychology and Family Therapy, in the Seton Hall University College of Education and Human Services.

Purpose and Duration of Research:

This project aims to add knowledge to a field that can ultimately tailor current resources and create new ones for the unique needs of family members of individuals with Leber's Hereditary Optic Neuropathy.

To make your participation as brief as possible, this survey should take about 20 minutes to complete.

Instruments:

Participants will complete 5 instruments during this survey. (1)Demographic Questionnaire, (2)The Impact of Event Scale, (3)The Outcome Questionnaire 45, (4) The Spiritual Involvement and Beliefs Scale, (5) The Multidimensional Scale of Perceived Social Support.

Procedures and Voluntary Participation:

If you are 18 years or older, are the mother or sibling of someone who has lost vision due to Leber's Hereditary Optic Neuropathy (LHON) and have been diagnosed with LHON, you are eligible to take part in this survey. Participation in this study is completely voluntary. You may withdraw from the study at any time without consequence.

Anonymity Preservation and Confidentiality Maintenance:

Your anonymity will be maintained throughout all aspects of the study. Any publication of the data from this study will in no way identify you and results will be reported in combined form only. All materials will be collected in the strictest confidence. Completed responses to questionnaires will be kept in a secure location and will be accessible only to myself and my academic advisor, Dr. Laura Palmer. The data will be stored electronically on a USB memory key and kept in a locked, secure physical setting.

Anticipated Risks and Discomfort:

There are no significant risks or discomforts likely to be associated with this study. However, participants who do experience significant distress are urged to use the American Psychological Association's psychologist locator to request a referral to a psychologist in your area through the following website: <http://locator.apa.org/>.

Benefits to Research:

Participation provides useful information in further understanding some of the challenges facing individuals affected by vision loss due to Leber's Hereditary Optic Neuropathy and their families. Personally, this study may also be useful to increase your self-awareness

Alternative Procedures:

This study does not involve any clinical treatment; therefore, there are no relevant alternative procedures.

Accommodations:

If this online survey is not compatible with your Assistive Technology, or you would prefer to be administered the questions in an alternative format, please email Jonathan W. Dator and accommodations will be provided.

Contact Information:

If participants have questions regarding the research process or would like to have a copy of the results, please contact Jonathan W. Dator. If participants have questions regarding their rights as research participants, the Director of Seton Hall University Institutional Review Board (IRB), Dr. Mary Ruzicka, may be reached at 973-313-6314.

Jonathan W. Dator, M.Ed., Principal Researcher
Jonathan.Dator@student.shu.edu
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Dr. Laura K. Palmer, Ph.D., ABPP, Faculty Advisor
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Consent to participate is indicated by completing these assessments, and participants are affirming that they are at least 18 years old.

I agree to participate in the study as described above. I do NOT agree to participate in the study as described above

Appendix C

Demographic Questionnaire

Demographic Information: Some of the following questions may seem quite personal. Please understand that we are not trying to pry into your lives but rather need to know this information for statistical purposes. Your responses will be kept strictly confidential. It is very important that you answer every question. Thank you very much.

1. Which of the following age groups do you fit into?
 18-24
 25-35
 36-44
 45-55
 56-64
 65+
2. What is your gender?
 Male
 Female
3. Which Race/Ethnicity do you most strongly identify as?
 White/Caucasian
 Black/African-American
 Latino/Hispanic
 Asian/Pacific Islander
4. What is your current relationship status?
 Single
 In a Relationship
 Married
 Divorced
 Widowed
 Separated
5. How many children do you have?
 0
 1
 2
 3
 4
 5 or more
6. What is your current employment status?
 Unemployed
 Employed
 Self Employed
 Student
7. What is your relationship to the person in your family that has lost vision due to LHON?
 I am the mother of someone with LHON
 I am the sibling (sister/brother) of someone with LHON

8. Which LHON Mutation do you carry?
- 11778
 - 14484
 - 3460
 - 15257
 - Other
9. How many years has it been since your family member lost their vision?
- Less than 1 year
 - 1-3 years
 - 4-7 years
 - 7-10 years
 - 10-20 years
 - Over 20 years
10. Are you concerned that you will lose your sight as well?
- Yes
 - No
11. What term do you use to describe your family member with vision loss due to LHON?
- Blind
 - Legally-Blind
 - Visually Impaired
 - I do not use any term
12. In what region do you currently reside?
- Northeast United States
 - Southeast United States
 - Western United States
 - Midwest United States
 - Canada
 - Latin America/Caribbean
 - Europe
 - Africa
 - Australia or New Zealand
 - Asia

Appendix D

Impact of Event Scale (IES)

List the Date of the Event_____

Describe the Event: Family Member Diagnosed with LHON

*Below is a list of comments made by people after stressful life events. Please mark each item, indicating how frequently these comments were true for you **during the past seven days**. If they did not occur during that time please click the "not at all" column.*

Select only one answer per row.

	Not at all	Rarely	Sometimes	Often
1. I thought about it when I didn't mean to.	0	1	3	5
2. I avoided letting myself get upset when I thought about it or was reminded about it.	0	1	3	5
3. I tried to remove it from memory.	0	1	3	5
4. I had trouble falling asleep or staying asleep because of pictures or thoughts about it that came to my mind.	0	1	3	5
5. I had waves of strong feelings about it.	0	1	3	5
6. I had dreams about it.	0	1	3	5
7. I stayed away from reminders about it.	0	1	3	5
8. I felt as if it hadn't happened or was unreal.	0	1	3	5
9. I tried not to talk about it.	0	1	3	5
10. Pictures about it popped into my mind.	0	1	3	5
11. Other things kept making me think about it.	0	1	3	5
12. I was aware that I still had a lot of feelings about it, but I didn't deal with them.	0	1	3	5
13. I tried not to think about it.	0	1	3	5
14. Any reminder brought back feelings about it.	0	1	3	5
15. My feelings about it were kind of numb.	0	1	3	5
	<u> 0 </u>	+ <u> </u>	+ <u> </u>	+ <u> </u> = <u> </u>

Scoring: Total each column and add together for a total stress score.

For example, every item marked in the "not at all" column is valued at 0. In the "rarely" column, each item is valued at a 1. In the "sometimes" column every item marked has a value of 3 and in the "often" column each item is valued at 5. Add the totals from each of the columns to get the total stress score.

Appendix E

Outcome Questionnaire (OQ-45)

Instructions: Looking back over the last week, including today, help us understand how you have been feeling. Read each item carefully and mark the items that best describe your current situation. For this questionnaire, work is defined as employment, school, housework, volunteer work, and so forth.

0=Never 1=Rarely 2=Sometimes 3=Frequently 4=Almost always

1. ___ I get along well with others.

2. ___ I tire quickly.

3. ___ I feel no interest in things.

4. ___ I feel stressed at work/school.

5. ___ I blame myself for things.

6. ___ I feel irritated.

7. ___ I feel unhappy in my marriage/significant relationship.

8. ___ I have thoughts of ending my life.

9. ___ I feel weak.

10. ___ I feel fearful.

11. ___ After heavy drinking, I need a drink the next morning to get going. (If you do not drink, mark "never").

12. ___ I find my work/school satisfying.

13. ___ I am a happy person.

14. ___ I work/study too much.

15. ___ I feel worthless.

16. ___ I am concerned about family troubles.

17. ___ I have an unfulfilling sex life.

18. ___ I feel lonely.

19. ___ I have frequent arguments.

20. ___ I feel loved and wanted.

21. ___ I enjoy my spare time.

22. ___ I have difficulty concentrating.

23. ___ I feel hopeless about the future.

24. ___ I like myself.

25. ___ Disturbing thoughts come into my mind that I cannot get rid of.

26. ___ I feel annoyed by people who criticize my drinking (or drug use). (If not applicable, mark "never").

27. ___ I have an upset stomach.

28. ___ I am not working/studying as well as I used to.

29. ___ My heart pounds too much.

30. ___ I have trouble getting along with friends and close acquaintances.

31. ___ I am satisfied with my life.

32. ___ I have trouble at work/school because of drinking or drug use. (If not applicable, mark “never”).

33. ___ I feel that something bad is going to happen.

34. ___ I have sore muscles.

35. ___ I feel afraid of open spaces, of driving, or being on busses, subways, and so forth.

36. ___ I feel nervous.

37. ___ I feel my love relationships are full and complete.

38. ___ I feel that I am not doing well at work/school.

39. ___ I have too many disagreements at work/school.

40. ___ I feel something is wrong with my mind.

41. ___ I have trouble falling asleep or staying asleep.

42. ___ I feel blue.

43. ___ I am satisfied with my relationships with others.

44. ___ I feel angry enough at work/school to do something I might regret.

45. ___ I have headaches.

Please indicate how often you do the following:

Always Usually Sometimes Rarely Never

20. When I wrong someone, I make an effort to apologize. -----

21. When I am ashamed of something I have done, I tell -----

22. I solve my problems without using spiritual resources. -----

23. I examine my actions to see if they reflect my values. -----

24. During the last WEEK, I prayed. . . (check one)

- 10 or more times.
- 7-9 times.
- 1-3 times.
- 4-6 times.
- 0 times.

25. During the last WEEK, I meditated... (check one)

- 10 or more times.
- 7-9 times.
- 4-6 times
- 1-3 times.
- 0 times.

26. Last MONTH, I participated in spiritual activities with at least one other person... (check one)

- more than 15 times.
- 11-15 times.
- 6-10 times.
- 1-5 times.
- 0 times.

Appendix G

Multidimensional Scale of Perceived Social Support

Instructions: We are interested in how you feel about the following statements. Read each statement carefully. Indicate how you feel about each statement.

Circle the "1" if you **Very Strongly Disagree**

Circle the "2" if you **Strongly Disagree**

Circle the "3" if you **Mildly Disagree**

Circle the "4" if you are **Neutral**

Circle the "5" if you **Mildly Agree**

Circle the "6" if you **Strongly Agree**

- | | | | | | | | | | |
|-----|--|---|---|---|---|---|---|---|-----|
| 1. | There is a special person who is around when I am in need. | 1 | 2 | 3 | 4 | 5 | 6 | 7 | SO |
| 2. | There is a special person with whom I can share my joys and sorrows. | 1 | 2 | 3 | 4 | 5 | 6 | 7 | SO |
| 3. | My family really tries to help me. | 1 | 2 | 3 | 4 | 5 | 6 | 7 | Fam |
| 4. | I get the emotional help and support I need from my family. | 1 | 2 | 3 | 4 | 5 | 6 | 7 | Fam |
| 5. | I have a special person who is a real source of comfort to me. | 1 | 2 | 3 | 4 | 5 | 6 | 7 | SO |
| 6. | My friends really try to help me. | 1 | 2 | 3 | 4 | 5 | 6 | 7 | Fri |
| 7. | I can count on my friends when things go wrong. | 1 | 2 | 3 | 4 | 5 | 6 | 7 | Fri |
| 8. | I can talk about my problems with my family. | 1 | 2 | 3 | 4 | 5 | 6 | 7 | Fam |
| 9. | I have friends with whom I can share my joys and sorrows. | 1 | 2 | 3 | 4 | 5 | 6 | 7 | Fri |
| 10. | There is a special person in my life who cares about my feelings. | 1 | 2 | 3 | 4 | 5 | 6 | 7 | SO |
| 11. | My family is willing to help me make decisions. | 1 | 2 | 3 | 4 | 5 | 6 | 7 | Fam |
| 12. | I can talk about my problems with my friends. | 1 | 2 | 3 | 4 | 5 | 6 | 7 | Fri |

Circle the "7" if you **Very Strongly Agree**