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The Informational and Emotional Support Needs of Grandparents of Children with Pompe Disease

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The Informational and Emotional Support Needs of
Grandparents of Children with Pompe Disease

by

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Abstract

The complex roles and experiences of grandparents of children with various diagnoses have been described, but previous studies have not investigated the roles and experiences of grandparents of children with treatable, Mendelian conditions such as Pompe disease. The availability of treatment and heritable nature of Pompe introduce the possibility for unique grandparent roles, experiences and needs. This is a particularly timely investigation given the advent of newborn screening for Pompe. This study aimed to characterize grandparents' roles and involvement, identify grandparents' information and emotional support needs, and explore the psychosocial impact felt by grandparents of children with Pompe. An online questionnaire containing forced choice and open-ended questions was distributed through various Pompe disease organizations. Quantitative data and qualitative data were analyzed with descriptive statistics, statistical measures, and thematic analysis. Twenty-one grandparents of children diagnosed with Pompe before the age of 18 participated. Grandparents provided emotional support significantly more frequently than financial support ($p = 0.011$), long-term child care ($p < 0.0005$), medical assistance ($p < 0.0005$), and running errands ($p = 0.002$). Grandparents were satisfied with their grandchild's parents serving as primary sources of information and emotional support. Grandparents most valued information about treatment. Most participants learned about the genetics of Pompe ($n = 16$) and comprehended the genetic etiology ($n = 15$). While grandchildren's parents ($n = 18$), Internet resources ($n = 15$), and religion ($n = 12$) were common sources of emotional support, family and religion were most

important. Psychosocial impacts included altered travel and employment plans, frustration with the diagnosis, heightened awareness of grandchildren's limitations and medical needs, and the experience of double-grief. Though grandparents are large sources of emotional and practical support for families of children diagnosed with Pompe, they need considerable support themselves. However, specific resources for grandparents beyond their grandchild's parents are limited. Given the complex family system in which patients reside, these results warrant consideration of the unique support needs of extended family members surrounding the diagnosis of a genetic condition and facilitation of familial communication of complex medical information.

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Chapter 1: Background

1.1 Overview of Pompe Disease

Pompe disease, or Glycogen Storage Disease Type II, is an autosomal recessive condition that occurs in one in forty thousand people in the United States (Kishnani, Steiner, et al., 2006; Lim, Li, & Raben, 2014). This disease is caused by a deficiency in the lysosomal enzyme acid alpha-glucosidase (GAA) (Lim et al., 2014). When this enzyme is absent or improperly functioning, individuals are incapable of degrading glycogen into glucose. The inability to degrade glycogen causes glycogen accumulation in lysosomes, which in turn compromises tissue function (Kishnani, Steiner, et al., 2006; Lim et al., 2014).

The GAA enzyme becomes non-functional or absent when mutations in the *GAA* gene occur (Ko et al., 1999). For the disease to manifest, a patient must have alterations in the genetic sequence in both *GAA* alleles. A patient with Pompe disease will inherit one mutated *GAA* allele from each parent. While the parents of patients with Pompe disease are typically unaffected, each of their children has a twenty-five percent chance for having Pompe disease. Although there is only one causative gene for Pompe, the condition can be caused by a variety of alterations in the genetic sequence (Martiniuk, Mehler, Tzall, Meredith, & Hirschhorn, 1990).

The genetic heterogeneity that can cause Pompe disease reflects the range of clinical descriptions for the condition. Pompe disease has a variety of manifestations that

are described by the age of onset of the symptoms and the severity of the progression (Lim et al., 2014; Martiniuk et al., 1990; Slonim et al., 2000). Pompe disease is further described by one of three categories based on the onset of the patient's symptoms: classic infantile Pompe disease, non-classic infantile Pompe disease, and late-onset Pompe disease (Lim et al., 2014).

Classic infantile Pompe disease is the most severe form of Pompe disease. Infants typically develop symptoms within the first two months of life, and the disease progresses most rapidly in these patients. This subset of Pompe disease occurs when the GAA enzyme is absent or nearly absent, or when the GAA enzyme has less than 1% function (Van den Hout et al., 2003; Zampieri et al., 2011). Infants will present with hypotonia, generalized muscle weakness, feeding difficulties, failure to thrive, and respiratory distress. In this subset of Pompe disease, patients also experience some malfunctioning of the cardiac tissue, specifically cardiomegaly or hypertrophic cardiomyopathy (Lim et al., 2014; Van den Hout et al., 2003) . This enlargement of the heart can obstruct respiratory function and proper circulation. Without treatment, the disease progresses rapidly, and patients with classic infantile Pompe usually die within the first year of life (Kishnani, Hwu, et al., 2006; Lim et al., 2014; van den Hout et al., 2003).

Patients with non-classic infantile Pompe disease experience symptoms within the first year of life. These symptoms include hypotonia, respiratory insufficiency, and an accumulation of glycogen in muscle fibers. If these patients develop cardiomegaly, the heart enlargement is less severe than in classic infantile Pompe disease. Diagnosis with non-classic infantile Pompe disease confers a better prognosis, as these patients tend to

have longer life expectancies compared to individuals with classic infantile Pompe disease (Slonim et al., 2000).

Symptoms in individuals who are diagnosed with late-onset Pompe disease can begin any time from childhood into adulthood. Patients who develop late-onset Pompe disease have partial GAA enzyme activity, or GAA activity that is 2-40% of normal function (Zampieri et al., 2011). Muscle weakness, particularly in the lower extremities, and respiratory insufficiency are the most common symptoms experienced by these patients. Many patients become wheelchair dependent from the progressive muscle weakness. Cardiac implications are not frequently seen in patients with late-onset Pompe disease (Lim et al., 2014).

Although these symptoms of Pompe disease are striking, an individual may undergo numerous medical evaluations and tests to establish a diagnosis. Additional testing such as chest x-rays, electrocardiograms, urinary tetrasaccharide measurements, and blood tests to help establish a diagnosis of Pompe disease. Diagnostic testing in the form of GAA enzyme activity assays, muscle biopsies, or genetic testing are used to confirm a diagnosis of Pompe disease (Kishnani, Steiner, et al., 2006)

Once a diagnosis of Pompe disease has been established, treatment becomes a consideration. Treatment for Pompe includes a number of options to address the symptoms once they appear, and patients require a large, interprofessional medical team for comprehensive care (Kishnani, Steiner, et al., 2006). However, if a patient can be diagnosed before the presentation of these symptoms, treatment with enzyme replacement therapy (ERT) can prevent their appearance and substantially improve life expectancy and quality (Chien et al., 2015). Although ERT has proven to benefit patients,

the treatment is not a cure for Pompe disease. In fact, the response to ERT is quite variable, and a number of factors that contribute to determining a patient's response have been identified. Even patients who have favorable responses to ERT are not without additional health care needs. Some of the complications faced by long-term survivors of classic infantile Pompe disease include arrhythmia, muscle weakness, impaired gross motor function, speech impairments, and hearing impairments (Chien et al., 2015; Prater et al., 2012).

Beginning ERT as soon as possible is essential to improve patient outcome (Chien et al., 2009; Chien et al., 2011; Chien et al., 2015). Therefore, a reasonable conclusion is to screen for Pompe disease during newborn screening. Detection of Pompe disease has occurred as early as a few days after birth through the use of newborn screening methods that measure the activity of the GAA enzyme, enabling the detection of patients with both infantile and late onset Pompe disease (Chien et al., 2011; Chien et al., 2015).

Identification of patients with classic infantile Pompe disease allows these patients to receive treatment before the onset of observable symptoms, which dramatically improves the patient's prognosis (Chien et al., 2015). Detection of patients with later onset Pompe disease can improve the prognosis for these patients by encouraging frequent medical evaluations to monitor the patient's health. Regular medical evaluations can ensure that a patient begins ERT at the earliest and most appropriate time (Chien et al., 2011).

Although the ability to screen newborns for Pompe disease has existed since 2005, the United States had not recommended including this condition on newborn screening until 2015 (Burwell, 2015; Chien et al., 2015). While early diagnosis and treatment can

considerably improve a patient's physical outcome, this does not address the impact a genetic diagnosis has on a family.

1.2 Family Impact of Child Disability and Illness

1.2.1 Impact on the nuclear family. The birth of a child with any disability or life-threatening condition has a direct impact on the family in both positive and negative ways (Lee & Gardner, 2010; Povee, Roberts, Bourke, & Leonard, 2012; Whiting, 2014). Families report that raising a child with a disability has fostered increased tolerance, understanding, patience, compassion, and altruism (Lee & Gardner, 2010; Wikler, Wasow, & Hatfield, 1983). On the other hand, families also communicate a large number of challenges that accompany caring for a child with a disability or life-threatening condition. Parents experience shock, disbelief, loneliness, sadness, loss, guilt, anger, and frustration, even long after the initial diagnosis (Cameron, Snowdon, & Orr, 1992). In addition to the emotional impact on the parents, parental employment, family finances, parental mental and physical health, family relationships, social support, and time constraints are also challenges that families face when they have a child with a health condition. With the additional time and medical care that children with a life-threatening condition or disability require, parents may be forced to leave their jobs to ensure their child has proper care (Whiting, 2014). Parents often struggle with mental illness, fatigue, and back pain (Knapp, Madden, Curtis, Sloyer, & Shenkman, 2010; Whiting, 2014). Marital issues often arise in the face of raising a child who requires additional care (Whiting, 2014).

The impact of a child's disability is felt not only by the parents. Siblings of children with life-limiting conditions experience grief, worry, sadness, and heightened

senses of responsibility in caring for their sibling with a serious medical condition (Malcolm, Gibson, Adams, Anderson, & Forbat, 2014; Scherman, Gardner, Brown, & Schutter, 1995) . Additionally, these siblings acknowledge the social limitations they experience when their whole family cannot participate in certain activities (Malcolm et al., 2014).

1.2.2 Extended family impact. Members beyond the nuclear family also feel the impact of an ill child. It is well documented that grandparents of the sick child experience a plethora of emotions. When grandparents first learn about their grandchild’s diagnosis, they feel the same emotions that parents feel; anger, confusion, and sadness overwhelm grandparents with the introduction of a child with special needs to the family (Findler, 2014; Katz & Kessel, 2002; Miller, Buys, & Woodbridge, 2012; Scherman et al., 1995; Schilmoeller & Baranowski, 1998; Vadasy, Fewell, & Meyer, 1986; Woodbridge, Buys, & Miller, 2009). Feelings of despair and helplessness continue to plague grandparents for years after learning about their grandchild’s diagnosis (Katz & Kessel, 2002; Scherman et al., 1995).

The persistent sadness experienced by grandparents is often called “double grief.” Grandparents grieve not only for the shift in expectations for their grandchild but also for the stresses and burdens placed on their child, the parent (Hastings, 1997; Katz & Kessel, 2002; Ravindran & Rempel, 2011; Scherman et al., 1995; Vadasy et al., 1986; Woodbridge et al., 2009). Grandparents also worry about the future of their grandchild, including their grandchild’s ability to relate to others, to function independently, and to receive good educational programs and services (Scherman et al., 1995; Vadasy et al., 1986). The concern about family function in the future also exists. Grandparents see

themselves in an essential role in the care of their grandchild, and they worry about what will happen to the family when they are no longer able to provide support to the family (Hastings, 1997; Miller et al., 2012).

Just as the impact of a child with a disability or illness pervades the nuclear family, grandparental concerns and sadness reach beyond the grandchild and the parents. Grandparent grief may extend even further to “triple grief” as grandparents witness the impact that a grandchild’s illness has on their grandchildren who are not ill (Ravindran & Rempel, 2011). Any strain on family relationships serves as a stressor for grandparents as well (Miller et al., 2012). Furthermore, grandparents have concerns that relate more directly to themselves. For example, grandparents worry that they do not fully understand the cause of the disability and the implications it has for their grandchild (Katz & Kessel, 2002). Also, grandparents worry that all of the help they provide is taken for granted. For grandparents to be as involved as they are with their grandchildren, they often sacrifice their own plans for travel or work, and grandparents worry that the parents of the grandchild with special needs do not recognize these sacrifices they make (Miller et al., 2012).

When the cause of the grandchild’s disability is genetic, grandparents experience persistent guilt. This guilt is neither introduced nor amplified by family members. Instead, grandmothers can feel personally responsible and blame themselves for the transmission of the disease-causing genetic information (Lehmann, Speight, & Kerzin-Storrar, 2011).

Despite all of these emotions, grandparents rarely feel comfortable expressing themselves to the parents of the child with special needs. Grandparents feel compelled to

“hold their emotions” for the sake of the parents. Grandparents believe that the family needs optimism and positivity more than the family needs to understand the challenges that the grandparents are facing themselves (Miller et al., 2012).

Grandparents not only aim to convey positivity to their family; they do also benefit from being involved in the care of their grandchild with special needs. As grandparents spend more time with their grandchild, they develop acceptance and a positive perception of the grandchild (Katz & Kessel, 2002; Mirfin-Veitch, Bray, & Watson, 1997; Scherman et al., 1995; Schilmoeller & Baranowski, 1998; Vadasy et al., 1986). Grandparents commonly feel proud of their family when they have a grandchild with a disability. Both the family’s adaptation to the diagnosis and the grandchild’s accomplishment of certain goals instill a deep sense of pride in grandparents (Woodbridge et al., 2009). Grandparents also benefit from closer family bonds established by providing support to their families. Grandparental spousal relationships and extended family relationships are strengthened when grandparents are involved in the care of their grandchild with a disability or illness (Katz & Kessel, 2002; Scherman et al., 1995).

1.3 Roles and Involvement of Grandparents

The serial model of caregiving and the family systems models have identified grandparents as the most likely and as important sources of support to parents of children with disabilities (Green, 2001; Mirfin-Veitch et al., 1997). Evaluating the support given to parents of children with disabilities reveals that grandparents are, in fact, a large source of support for parents (Bruns & Foerster, 2011; Green, 2001; Heller, Hsieh, & Rowitz, 2000; Hornby & Ashworth, 1994; Katz & Kessel, 2002; Scherman et al., 1995). When

grandparents are involved in the care of a grandchild with a disability, they serve many roles, including supporter, liaison, and normalizer.

Grandparents provide support in two manners: instrumental and emotional (Lee & Gardner, 2010; Mirfin-Veitch et al., 1997; Seligman, Goodwin, Paschal, Applegate, & Lehman, 1997). Instrumental support occurs when grandparents provide assistance with practical tasks (Lee & Gardner, 2010). Grandparents are commonly involved in helping the parents of a sick child with medical care, respite care, household tasks, shopping, errands, child care for both sick children and their siblings, and financial aid (Green, 2001; Heller et al., 2000; Hornby & Ashworth, 1994; Katz & Kessel, 2002; Lee & Gardner, 2010; Ravindran & Rempel, 2011; Scherman et al., 1995). These practical acts of support are essential for preserving parental well-being (Green, 2001).

Although parents have an obvious need for assistance with practical tasks, the emotional support offered by grandparents is more important to parents (Findler, 2014; Lee & Gardner, 2010; Seligman et al., 1997). Emotional support from grandparents is described by their giving advice, being a confidant, being an encourager, and providing enjoyment (Heller et al., 2000). This emotional support from grandparents promotes parental well-being, father involvement, and better family adaptation (Heller et al., 2000; Seligman et al., 1997; Trute, 2003; Van Riper, 2007).

In addition to offering support to the parents, grandparents are responsible for maintaining family relationships. When a sick grandchild has siblings, grandparents foster well-child parent, sick-child parent, and sibling relationships. Grandparents will often care for the well-children to allow parents to attend medical appointments for the sick child. However, grandparents also encourage the parents to spend time with their

well-children by relieving the parents during long hospital stays. Furthermore, by taking well-children to visit their sick-siblings, grandparents foster sibling relationships (Ravindran & Rempel, 2011). Grandparents also manage relationships beyond the nuclear family; they act as an “interface” between the grandchild with a disability and the extended family (Miller et al., 2012).

While grandparents impact extended family relationships, they also alter the family’s perception of the sick or disabled grandchild. A grandparent can serve as a “role model” of appropriate behavior towards the disabled or sick grandchild. When a grandparent has a positive and accepting attitude towards the grandchild, extended family members are more likely to follow suit, and this acceptance of the child establishes parental well-being by normalizing the parents’ situation (Green, 2001; Lee & Gardner, 2010).

Although grandparents are known to play a pivotal role in supporting and benefitting the family when a child has a disability or illness, grandparental involvement can be a source of parental stress. The involvement of a grandparent in the care for a child with a disability or illness can introduce conflict or disagreement. When these disputes occur, maternal stress increases (Green, 2001; Hastings, Thomas, & Delwiche, 2002; Hastings, 1997). Furthermore, grandparents are often uncertain about the details of their grandchild’s diagnosis (Gardner, Scherman, Mobley, Brown, & Schutter, 1994; Katz & Kessel, 2002; Scherman et al., 1995). This uncertainty in grandparental understanding of the details of the diagnosis can cause grandparents to become a source of parental stress (Katz & Kessel, 2002; Lee & Gardner, 2010; Scherman et al., 1995; Vadasy et al., 1986).

Parental desires impact how involved grandparents are in their grandchild's care (Katz & Kessel, 2002). Previous parent-child relationships between the grandparent and parent determine the level of grandparental involvement. Grandparents fear overstepping boundaries. Previous tensions between the grandparent and parent reduce the likelihood of grandparental involvement (Mirfin-Veitch et al., 1997). A grandparent's poor understanding of their grandchild's diagnosis can hinder their involvement (Lee & Gardner, 2010; Mitchell, 2007). Other factors such as residential proximity, parents' marital status, and grandparent age and health influence grandparental support and involvement (Hornby & Ashworth, 1994; Lee & Gardner, 2010). Furthermore, the relationship of the grandparent to the sick or disabled grandchild can impact grandparental involvement. Grandmothers, and more specifically, maternal grandmothers are the most likely grandparents to be a source of support for the family (Bruns & Foerster, 2011; Hornby & Ashworth, 1994; Seligman et al., 1997; Shaw, 2005; Trute, 2003).

1.4 Resources for Grandparents

Despite the documented emotional toll a grandchild with a disability places on grandparents and the evidence that these grandparents both require and benefit from support, grandparents rarely receive any professional support (Hastings, 1997).

Support group involvement helps grandparents emotionally and promotes their involvement with their grandchild with special needs. Previous studies have found that grandparents benefit from discussing their emotions (Gardner et al., 1994). Grandparents who participate in support groups experience more positive emotions and are more

involved with their grandchild than grandparents who do not (Schilmoeller & Baranowski, 1998).

Grandparents have expressed an interest in support groups, counseling forums, and other opportunities to share information (Gardner et al., 1994). Grandparents have also shown themselves to be information-seeking; they claim that explanations of their grandchild's disability and information about their grandchild's progress are the most helpful support they receive from the parents of the child with special needs (Schilmoeller & Baranowski, 1998). Current literature recommends that support groups, grandparents' workshops, grandparent forums, family involvement programs, access to accurate information, and improving communication between grandparents and parents should all be available resources for grandparents (Lee & Gardner, 2010; Scherman et al., 1995). However, these resources are not commonly provided to grandparents (Scherman et al., 1995).

1.5 Support for Grandparents of Individuals with Pompe Disease

In the diagnosis and management guideline for Pompe, Kishnani, Steiner et al. (2006) acknowledged that the presence of an individual with Pompe disease introduces responsibility to extended family and friends. Furthermore, this guideline claims that health care providers should identify support for grandparents of individuals with Pompe disease. The identified resources for families managing Pompe disease in the guideline include the Muscular Dystrophy Association, Medicaid, and patient organizations such as the International Pompe Association, the Acid Maltase Deficiency Association, and the United Pompe Foundation (Kishnani, Steiner, et al., 2006). However, none of these resources offer support or resources specifically to grandparents.

1.6 Study Rationale

While a handful of literature exists to characterize grandparents' roles and experiences when a grandchild has a disability, a large gap in our knowledge of grandparent needs and experiences is still present. Grandparents of children with Down syndrome, Autism Spectrum Disorders, and cerebral palsy are the main focus of current literature. Current literature's focus on these populations has ignored a large number of grandparents. The conditions that have been studied are either not inherited or non-Mendelian and do not have available treatments. Therefore, we know little about grandparents' roles, experiences, and needs when they have a grandchild with an inherited, treatable diagnosis such as Pompe. The heritable nature of Pompe disease introduces the possibility for unique psychosocial impacts and emotional support needs, while the availability of treatment suggests that the informational needs and roles of grandparents in the family may be distinct from those of grandparents in other populations.

One study suggests that grandparents who carry genetic mutations that lead to their grandchild's diagnosis experience guilt in addition to the many other emotions felt by grandparents of children with special needs (Lehmann et al., 2011). However, current literature also suggests that the emotional impact of being a carrier of a genetic condition in the family is dependent upon the mode of inheritance (James, Hadley, Holtzman, & Winkelstein, 2006). Thus, defining the emotional impact of having a grandchild with an autosomal recessive condition, such as Pompe disease, is important and could provide insight into the experiences of a larger population of grandparents who have grandchildren with other autosomal recessive conditions such as cystic fibrosis, spinal

muscular atrophy, and many others. This study explored the psychosocial impacts of having a grandchild with Pompe disease through open-ended questions assessing life modifications. The emotional support needs of grandparents of children with Pompe were explored by asking grandparents to indicate their satisfaction with various sources of emotional support and to indicate their most valued sources of emotional support.

The available treatment for Pompe disease requires a greater number of medical appointments. With the added time and resources required to receive this treatment, it is possible that grandparents of children with Pompe may have slightly different roles than those that have been reported previously. Additionally, grandparents may have unique desires for information when their grandchild's condition can be treated. To understand grandparent roles in this unstudied population, our study investigated the frequency with which grandparents provide a variety of supports, grandparents' perspectives of their unique roles in their families, and any differences they have noticed between grandparenting their grandchild with Pompe and their healthy grandchildren. We assessed grandparents' satisfaction with the sources and type of information available and explored the most valuable information to grandparents. Furthermore, this study aimed to understand grandparents' use and comprehension of genetic information related to the etiology of their grandchild's diagnosis, which has not been reported in current literature.

The advent of newborn screening for Pompe disease makes studying the needs and experiences of grandparents of children with this specific treatable, autosomal recessive condition relevant and timely. Pompe disease was officially recommended for inclusion on newborn screening in the United States in 2015 (Burwell, 2015). This screening test will diagnose individuals with every form of Pompe disease within days of

their birth, meaning that more grandparents may be aware of their grandchild's diagnosis, even before symptom onset. With this heightened awareness of Pompe disease, having an understanding of how to better meet grandparent needs can allow healthcare providers to prepare families for what to expect and facilitate family psychosocial functioning.

The outcomes of this study are relevant in the field of genetic counseling, as a further understanding of the psychosocial impact of having a grandchild with a genetic condition will allow genetic counselors to provide appropriate resources to the families of children with a genetic condition. The importance for grandparental involvement in family functioning and adaptation following the birth of a grandchild with a disability is known through the literature for multiple different pediatric conditions (Green, 2001; Heller et al., 2000; Hornby & Ashworth, 1994; Katz & Kessel, 2002; Lee & Gardner, 2010; Ravindran & Rempel, 2011; Scherman et al., 1995; Seligman et al., 1997; Trute, 2003; Van Riper, 2007). Therefore, attending to these needs of grandparents of children with Pompe could help optimize family adjustment following the diagnosis of a genetic condition by facilitating and fostering grandparental adjustment to the genetic condition.

Chapter 2: The Informational and Emotional Support Needs of Grandparents of Children with Pompe Disease¹

2.1 Abstract

The complex roles and experiences of grandparents of children with various diagnoses have been described, but previous studies have not investigated the roles and experiences of grandparents of children with treatable, Mendelian conditions such as Pompe disease. The availability of treatment and heritable nature of Pompe introduce the possibility for unique grandparent roles, experiences and needs. This is a particularly timely investigation given the advent of newborn screening for Pompe. This study aimed to characterize grandparents' roles and involvement, identify grandparents' information and emotional support needs, and explore the psychosocial impact felt by grandparents of children with Pompe. An online questionnaire containing forced choice and open-ended questions was distributed through various Pompe disease organizations. Quantitative data and qualitative data were analyzed with descriptive statistics, statistical measures, and thematic analysis. Twenty-one grandparents of children diagnosed with Pompe before the age of 18 participated. Grandparents provided emotional support significantly more frequently than financial support ($p = 0.011$), long-term child care ($p < 0.0005$), medical assistance ($p < 0.0005$), and running errands ($p = 0.002$). Grandparents were satisfied with their grandchild's parents serving as primary sources of information and emotional

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support. Grandparents most valued information about treatment. Most participants learned about the genetics of Pompe ($n = 16$) and comprehended the genetic etiology ($n = 15$). While grandchildren's parents ($n = 18$), Internet resources ($n = 15$), and religion ($n = 12$) were common sources of emotional support, family and religion were most important. Psychosocial impacts included altered travel and employment plans, frustration with the diagnosis, heightened awareness of grandchildren's limitations and medical needs, and the experience of double-grief. Though grandparents are large sources of emotional and practical support for families of children diagnosed with Pompe, they need considerable support themselves. However, specific resources for grandparents beyond their grandchild's parents are limited. Given the complex family system in which patients reside, these results warrant consideration of the unique support needs of extended family members surrounding the diagnosis of a genetic condition and facilitation of familial communication of complex medical information.

2.2 Introduction

Pompe disease is an autosomal recessive lysosomal storage and glycogen storage disorder that affects one in forty thousand people in the United States (Kishnani, Steiner, et al., 2006; Lim et al., 2014). Pathogenic variants in the *GAA* gene cause insufficient glycogen degradation, leading to an accumulation of glycogen in lysosomes that compromises tissue function (Kishnani, Steiner, et al., 2006; Ko et al., 1999).

Of the three subtypes of Pompe disease, classic infantile Pompe disease (IPD) is the most severe, with onset of symptoms in infancy and rapid progression of hypotonia, generalized muscle weakness, failure to thrive, respiratory distress and hypertrophic cardiomyopathy. Without treatment, patients with IPD usually die in the first year of life

from cardiorespiratory failure (Kishnani, Hwu, et al., 2006; Lim et al., 2014; van den Hout et al., 2003). Non-classic or atypical infantile Pompe disease presents within the first year of life, but disease progression is slower and cardiac tissue is less commonly involved (Slonim et al., 2000). Late-onset Pompe disease may present at any point between childhood and adulthood with lower extremity muscle weakness and respiratory insufficiency (Lim et al., 2014).

Initiation of treatment with enzyme replacement therapy (ERT) before symptom onset can substantially improve life expectancy and quality of life (Chien et al., 2015). Though ERT has significant benefits for patients, this treatment is not a cure, and ERT has variable efficacy within the patient population. Even patients who respond well to ERT may face a plethora of additional medical complications, and a large medical team is required for comprehensive medical care (Chien et al., 2015; Kishnani, Steiner, et al., 2006; Prater et al., 2012).

The birth of a child with any disability or life-threatening condition impacts a family both positively and negatively (Lee & Gardner, 2010; Povee et al., 2012; Whiting, 2014). Parents benefit from increased tolerance, understanding, patience, compassion, and altruism even in the face of the shock, disbelief, loneliness, sadness, loss, anger, and frustration they experience at the time of their child's diagnosis (Cameron et al., 1992; Lee & Gardner, 2010; Wikler et al., 1983). On the other hand, parents face challenges with employment, finances, mental and physical health, family relationships, social supports, and time management (Knapp et al., 2010; Whiting, 2014).

Grandparents have been identified as large sources of support for families when children have additional medical needs (Bruns & Foerster, 2011; Green, 2001; Heller et

al., 2000; Hornby & Ashworth, 1994; Katz & Kessel, 2002; Scherman et al., 1995). Broadly speaking, grandparental roles in the family can be placed into two categories: emotional support or instrumental support. Grandparents provide emotional support by serving as confidants, providing encouragement, maintaining family relationships, and promoting grandchild acceptance (Green, 2001; Heller et al., 2000; Lee & Gardner, 2010; Miller et al., 2012; Ravindran & Rempel, 2011). Instrumental support includes financial assistance and fulfilling practical tasks such as child care and running errands (Lee & Gardner, 2010; Mirfin-Veitch et al., 1997; Seligman et al., 1997). Though instrumental support is necessary when a child has a complex medical condition, parents consistently deem grandparental emotional support more important (Findler, 2014; Lee & Gardner, 2010; Seligman et al., 1997). Parents value grandparental support, but grandparental involvement can be a source of parental stress, particularly if grandparents are uncertain about the detail of a grandchild's diagnosis (Gardner et al., 1994; Katz & Kessel, 2002; Scherman et al., 1995).

Grandparents are also largely impacted by a grandchild's diagnosis. Initially grandparents experience anger, confusion, and sadness when they learn of a grandchild's diagnosis. Feelings of despair and helplessness persist long after the initial diagnosis (Findler, 2014; Katz & Kessel, 2002; Miller et al., 2012; Scherman et al., 1995; Schilmoeller & Baranowski, 1998; Vadasy et al., 1986; Woodbridge et al., 2009). When the cause of a grandchild's condition is genetic, grandparents feel persistent guilt (Lehmann et al., 2011). The persistent sadness felt by grandparents is complex, and has been coined "double grief," which is grief felt not only for the shift in expectations for their grandchild but also for the stresses and burdens placed on their child (Hastings,

1997; Katz & Kessel, 2002; Ravindran & Rempel, 2011; Scherman et al., 1995; Vadasy et al., 1986; Woodbridge et al., 2009). Similar to parents, grandparents face challenges beyond the emotional impact of having a grandchild with a life-threatening condition as they alter life plans to be available for the family (Woodbridge, Buys, & Miller, 2011).

Despite the documented complex emotional experience grandparents endure when they have a grandchild with a disability, little support is available specifically for grandparents, and even less is available specifically for grandparents of children with Pompe disease. In the past, studies have discovered that grandparents are information seekers (Schilmoeller & Baranowski, 1998). For this reason, suggestions for resources and support for grandparents have included support groups, workshops, grandparent forums, family involvement programs, and access to accurate information (Lee & Gardner, 2010; Scherman et al., 1995).

This study aimed to 1) understand the roles and involvement of grandparents when a grandchild has Pompe disease, 2) explore grandparents' informational and emotional support needs, and 3) understand the impact of having a grandchild with Pompe disease. Investigating the roles, needs, and experiences of grandparents of children with Pompe provides insight into a new population of grandparents. Previous literature focuses mainly on grandparents of children with Down syndrome, Autism, and cerebral palsy, which does not address the needs and experiences of grandparents of children with inherited, treatable conditions such as Pompe and cystic fibrosis. The heritable nature of Pompe introduces the possibility for unique psychosocial impacts and emotional needs, while the available treatment for Pompe suggests that grandparent roles and informational needs may be distinct from previously studied populations (James et

al., 2006). Focusing on grandparents of children with Pompe is timely given the recent approval for inclusion of Pompe on newborn screening (Burwell, 2015). Having an understanding of how to better meet grandparent needs can allow healthcare providers to prepare families for what to expect and facilitate family psychosocial functioning.

2.3 Materials and Methods

2.3.1 Participants. This study received approval from the Institutional Review Board, Office of Research Compliance, of the University of South Carolina, Columbia, SC in June, 2015. The United Pompe Foundation (UPF), the Acid Maltase Deficiency Association (AMDA), the International Pompe Association (IPA), the UK branch of the Association for Glycogen Storage Disease (AGSD-UK), Children Living with Inherited Metabolic Diseases (CLIMB), the Australian Pompe Association, the United States branch of the Association for Glycogen Storage Disease (AGSDUS), Genzyme, and an individual manager of a Facebook Advocacy Group were contacted and asked to share a participation flyer and/or the survey link to the online survey hosted by surveymonkey.com (Appendices A, B). The UPF, AMDA, IPA, AGSD-UK, CLIMB, and the manager of a Facebook advocacy group all responded and agreed to share the information with members of their email lists, electronic mailing lists, and/or various Facebook support group affiliations. The links to the survey were shared in September, 2015, October, 2015, and January, 2016. The survey was open between September, 2015 and February, 2016. Eligible participants were grandparents of a child, living or deceased, diagnosed with Pompe disease before the age of 18.

2.3.2 Measure. The principal investigator conducted a pilot study of an initial draft of the survey in August, 2015, to assess survey question clarity and to gain

participant feedback over the phone using cognitive interviewing techniques and guided by a pilot interview guide (Appendix C; G. B. Willis, 1999; G. Willis, 2006).

Subsequently, survey questions were rephrased for clarity, and additional questions about grandparent-grandchild relationship strength and grandparent carrier status were added.

The final survey was distributed through the various means indicated above.

This online survey consisted of forced choice and open-ended questions. Questions included in the survey assessed demographic information, grandparents' roles and involvement in their families, grandparents' satisfaction with their sources of information and the type of information they received, grandparents' understanding of the genetics associated with Pompe, grandparents' satisfaction with their sources of emotional support, the psychosocial impact a grandchild's diagnosis of Pompe has on grandparents, and the specific informational and emotional support needs of grandparents. Participants rated their frequency of offering various supports as *Daily = 4, Weekly = 3, Monthly = 2, A few times a year = 1, or I do not do this = 0*. Average satisfaction with various sources of information, types of information, and sources of emotional support were calculated by participant responses to Likert scale questions: *Extremely Satisfied = 4, Satisfied = 3, Somewhat satisfied = 2, Not satisfied = 1, or I have not gotten information/emotional support from this source/I have not learned this = 0*. Questions to assess participant agreement and overall satisfaction were included as Likert scale questions: *Strongly Agree/Very Satisfied = 5, Agree/Satisfied = 4, Unsure/Neutral = 3, Disagree/Dissatisfied = 2, Strongly Disagree/Very Dissatisfied = 1*. Responding to each question was voluntary, which allowed participants to skip questions they did not wish to answer (Appendix D).

2.3.3 Data analysis. Quantitative analysis was performed using Statistical Package for the Social Sciences (SPSS), Version 23 of the SPSS system for Windows, Copyright ©2015 IBM (Aramonk, NY), and Microsoft Excel 2013, Copyright © 2013 Microsoft Corporation (Redmond, WA). Microsoft Excel was used for descriptive statistics, while SPSS was used for data analysis via Friedman test, Cronbach's alpha, independent-samples t- tests, and Pearson's product-moment correlation. A 0.05 level of significance was used for all analyses.

Investigation of any differences in frequency of supports provided by grandparents was completed through use of a Friedman test on participants' responses to question 18 (Appendix D). Pairwise comparisons were performed with a Bonferroni correction for multiple comparisons. Post hoc analysis was performed to identify which supports were offered with statistically significant different frequencies.

Single scores for participant involvement (Involvement Score) and participant satisfaction with types of information (Information Satisfaction Score) were calculated for use in later analyses. Before computing Involvement Scores and Information Satisfaction Scores for participants, Cronbach's alpha was calculated for Questions 18 and 25 to determine the internal consistency of these questions as they measured the underlying constructs of involvement and information satisfaction, respectively (Appendix D). Question 18 consisted of six sub-questions to assess the frequency with which participants provided various forms of support to their families. For participants with a living grandchild, these six sub-questions had a moderate level of internal consistency, as determined by a Cronbach's alpha of 0.689. Exclusion of the sub-question exploring participants' frequency of providing financial support, resulted in a Cronbach's

alpha of 0.733, indicating that the remaining five sub-questions to assess involvement had a high level of internal consistency. Thus, each participant with a living grandchild who reported a frequency for all five sub-questions (emotional support, respite care/short-term child care, primary child care/long-term child care, assistance with medical care/management for Pompe, and running errands/completing housework) had an Involvement score calculated by summing their frequencies of providing support: *I do not do this = 0, A few times a year = 1, Monthly = 2, Weekly = 3, and Daily = 4*. Therefore, Involvement Scores were calculated for 18 participants.

Question 25 consisted of ten sub-questions and was used to measure the underlying construct of information satisfaction. These ten sub-questions had a high level of internal consistency, as determined by a Cronbach's alpha of 0.886. Thus, Information Satisfaction Scores were calculated for participants who indicated a satisfaction for every sub-question by summing participants' responses: *I have not learned this = 0, Not satisfied = 1, Somewhat Satisfied = 2, Satisfied = 3, Extremely Satisfied = 4*.

As previous literature reports higher levels of involvement from maternal grandmothers, Involvement Scores were used in an independent-samples t-test to determine if any differences in involvement for participants in this study were statistically significant. Independent-samples t-tests were run for males and females and maternal and paternal grandparents.

Given the current literature's descriptions of relationships between grandparent uncertainty surrounding a grandchild's diagnosis and parental stress and grandparent involvement and family well-being, the data gathered from this study was analyzed for a possible relationship between grandparent satisfaction with information and grandparent

involvement (Heller et al., 2000; Katz & Kessel, 2002; Lee & Gardner, 2010; Scherman et al., 1995; Seligman et al., 1997; Trute, 2003; Vadasy et al., 1986; Van Riper, 2007). A Pearson's product-moment correlation was run to assess the relationship between participants' Information Satisfaction Scores and Involvement Scores. Preliminary analysis of a scatterplot of the data showed the relationship to be linear with one outlier. This one outlier was removed (Information Satisfaction Score = 0, Involvement Score = 15) before the data analysis by Pearson product-moment correlation.

Qualitative data was analyzed by the principal investigator to identify recurring themes using thematic analysis (Braun & Clarke, 2006). Themes were identified for each open-ended question individually and across questions.

2.4 Results

2.4.1 Participant demographics. A total of 30 participants began the survey. One participant did not meet the inclusion criteria, as he reported that his grandchild was diagnosed with Pompe at age 18; this response was not included in data analysis. Two responses from the same IP address with the same demographic information were received. One of these surveys was partially complete, while the other survey was complete. The partially completed survey was excluded with the assumption that both responses were from the same participant. An additional seven participants provided only demographic information and were also excluded. Of the remaining twenty-one responses, two provided only demographic information and information about their roles and involvement in the family. These two surveys were included because the answers provided would assist in answering an overarching study goal (i.e. define the role of

grandparent in the family when a grandchild has Pompe). Thus, twenty-one surveys were analyzed.

All of the participants reported Caucasian ethnicity ($n = 21$). Most respondents were married ($n = 17$); female ($n = 19$); from the United States ($n = 17$); and had some religious affiliation ($n = 19$). The average age of participants was 60.9 years ($n = 20$), and ranged from 50 to 74 years. Nearly half of the participants lived less than 30 miles from their grandchild ($n = 10$). More than half of the participants were maternal grandmothers ($n = 11$). (Table 2.1). Participants reported an average relationship strength with their grandchild diagnosed with Pompe of 4.4 out of 5, indicating that most grandparents felt they had a strong relationship with their grandchild.

Table 2.1 Participant Demographics

	Frequency
Gender	
Female	19
Male	2
Ethnicity	
Caucasian	21
Relationship	
Maternal	13
Paternal	8
Residence	
United States	17
Outside of the United States	4
Distance to Grandchild	
< 30 miles away	10
30-100 miles away	5
>100 miles away	6
In the same house	0
Religious Affiliation	
No religious affiliation	2
Religious affiliation	19
Relationship Status	
Married	17
Divorced	1
In a domestic partnership or civil union	1
Widowed	1

2.4.2 Demographics of grandchildren with Pompe. Participants were asked to report demographic information about their grandchild (Table 2.2). Most grandchildren were diagnosed with classic infantile Pompe disease ($n = 13$) at or before one year of age ($n = 19$). Twenty participants had living grandchildren whose current ages ranged from 0-3 months to 18 years. One participant had a grandchild with Pompe who died at age 12 years.

Table 2.2 Demographics of Grandchildren Diagnosed with Pompe Disease

		Frequency
Age at diagnosis		21
	0-3 months	9
	4-6 months	2
	7-12 months	3
	1 year	5
	2 years	1
	7 years	1
Living vs. deceased		21
	Living	20
	Deceased	1
Current age		19
	0-3 months	1
	7-9 months	1
	10-12 months	2
	1 year	4
	2 years	2
	4 years	1
	5 years	2
	6 years	2
	8 years	1
	9 years	1
	10 years	1
	18 years	1
Type of Pompe		21
	Classic Infantile	13
	Juvenile or Late onset	4
	Unsure	4

2.4.3 Type and frequency of support provided by grandparents. Only one participant was the primary caregiver for her grandchild; the remaining 20 participants

reported a primary role of support for the family. Participants were asked to indicate the frequency with which they provide a number of different supports to their families [Daily, Weekly, Monthly, A few times a year, and “I do not do this”]. Emotional support was the most commonly provided support by participants ($n = 19$), followed by financial support ($n = 16$), and respite care/short-term child care ($n = 16$). Though only one grandparent reported acting as the primary caregiver, five participants provided primary care for their grandchild. (Figure 2.1).

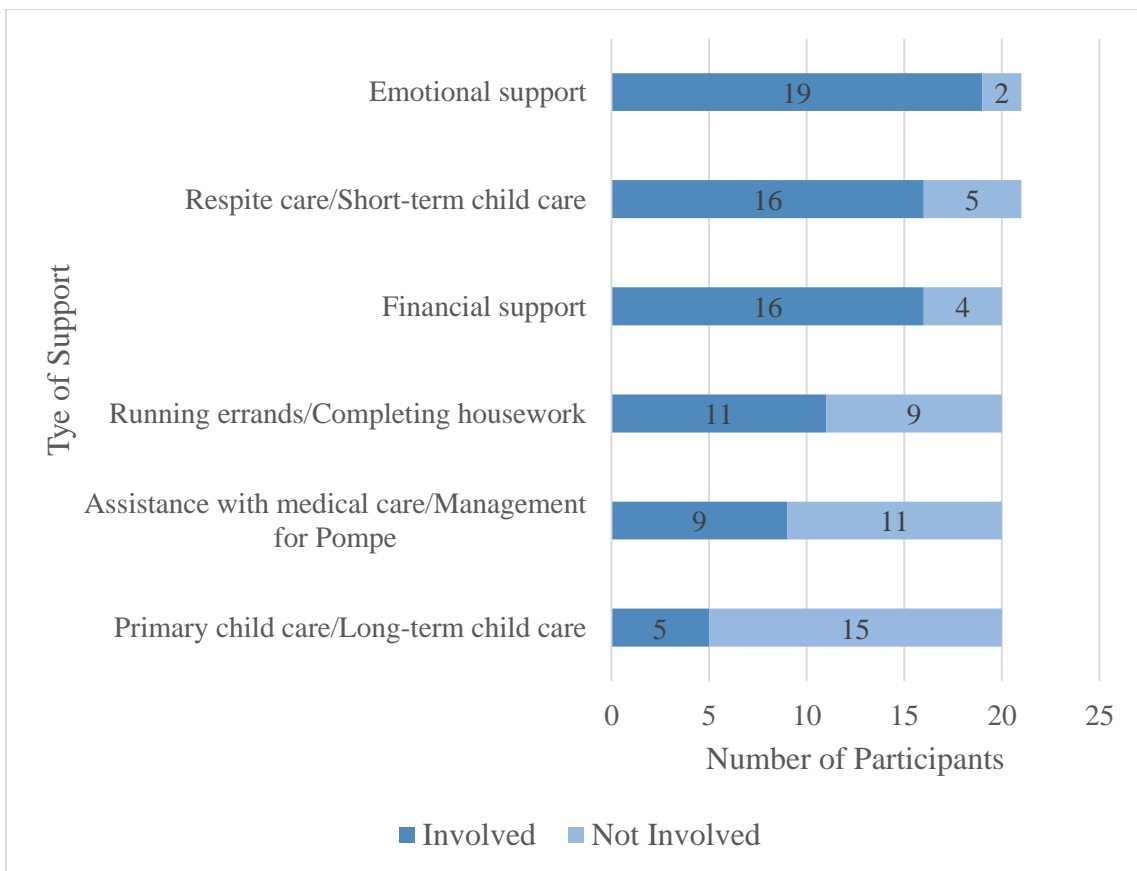


Figure 2.1 Types of Support Provided by Grandparents

Emotional support was also most frequently provided by participants. Nearly all participants provided emotional support at least weekly ($n = 17$) (Figure 2.2).

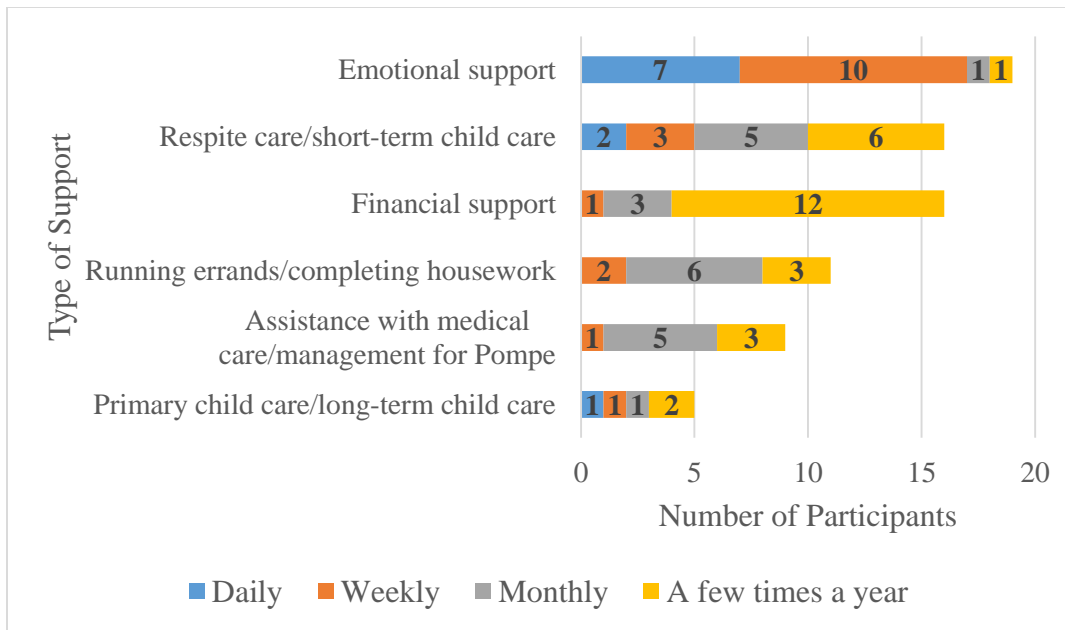


Figure 2.2 Frequencies of Grandparent Provision of Various Types of Support

All types of support provided by participants were compared pairwise. A statistically significant difference between the frequencies with which various supports were offered was identified $\chi^2(5) = 41.814, p < 0.0005$. Emotional support was offered statistically significantly more frequently than long-term child, medical assistance/management of Pompe, running errands, and financial support (Table 2.3).

Table 2.3 Statistically Significant Differences between Frequencies of Various Supports

Type of Support (Median frequency)	Type of Support (Median frequency)	p-value
Emotional support (4.0)	Long-term child care (1.0)	<0.0005
Emotional support (4.0)	Medical assistance/management of Pompe (1.0)	<0.0005
Emotional support (4.0)	Running errands/completing household tasks (2.0)	0.002
Emotional support (4.0)	Financial support (2.0)	0.011

Although most grandparents reported providing emotional support and reported providing this support to their families most frequently, when asked to describe their unique roles in the family, respondents focused more on the practical support they provide. In addition to describing the type of support they offer, participants also discussed the recipients of their support, including their grandchildren with Pompe, their children, their healthy grandchildren, and other family members (Table 2.4).

Table 2.4 Themes of Grandparents’ Perceived Unique Family Roles

Theme	Sub-theme	Frequency	Example quote
Support		<i>n</i> = 12	
	Practical support (financial, medical appointments, babysitting, etc.) only	4	<i>“I help as much as they need me. I travel to Dr appts and help with bi-weekly infusions. They have four children with our Pompe child being #4. They lead a very busy life.”</i>
	Emotional support only	1	<i>“Emotional support for children and grandchildren.”</i>
	Practical and Emotional support	5	<i>“I try to offer moral support to my daughter and her husband. My wife and I do help financially. We also babysit the older grandchild...”</i>
	Unspecified support	2	<i>“It is not hard for me to offer support to my daughter who is an amazing mother.”</i>
Recipient of support		10	
	Healthy grandchild	4	<i>“I also spend lots of time each month with her sister...doing things like school pick-ups, and caring for her during her sister’s Pompe treatments and appointments. She is only 7 and is very sensitive to her sister’s fears and discomfort.”</i>
	Grandchild with Pompe	5	<i>“Full time babysitter for her.”</i>
	Grandchild’s parent(s)	8	<i>“Intense support for my daughter emotionally.”</i>
	Family	2	<i>“I support my son’s family letting them know that I’m by their side 24/365.”</i>

In addition to understanding the roles and involvement grandparents have with their grandchild diagnosed with Pompe, we attempted to understand how grandparenting a child with Pompe may be different from grandparenting a child without Pompe. Eighteen participants responded to the question, but two respondents indicated that their grandchild with Pompe was their only grandchild. Therefore, sixteen responses addressed the question of whether or not grandparenting roles differed between unaffected and affected grandchildren. Half of these responses described a heightened awareness of their grandchild’s limitations and medical needs (Table 2.5).

Table 2.5 Perceived Differences between Grandparenting Grandchildren with Pompe and Healthy Grandchildren

Theme	Frequency	Example quotes
No difference	5	<i>“It isn’t—all are treated with the same unconditional love.”</i>
Increased awareness of medical needs/limitations	8	<i>“She is mobile with the help of braces but obviously needs more help.”</i> <i>“When they are at the house they have to be watched more closely than the others due to the balance issues and choking. So we have to be more aware of what they are doing, we don’t allow them to play in the bedroom alone, we keep them in our sight. Which we don’t do with the other kids. [sic]”</i>

2.4.3.1 Differences in involvement by gender and participant relationship.

Paternal grandmothers had the highest average Involvement Scores ($n = 7, M = 13$) and maternal grandfathers had the lowest average Involvement Score ($n = 2, M = 7$) (Figure 2.3). There were no statistically significant differences between average involvement scores for grandmothers and grandfathers, or for maternal grandmothers and paternal grandmothers, as calculated by independent-samples t-tests.

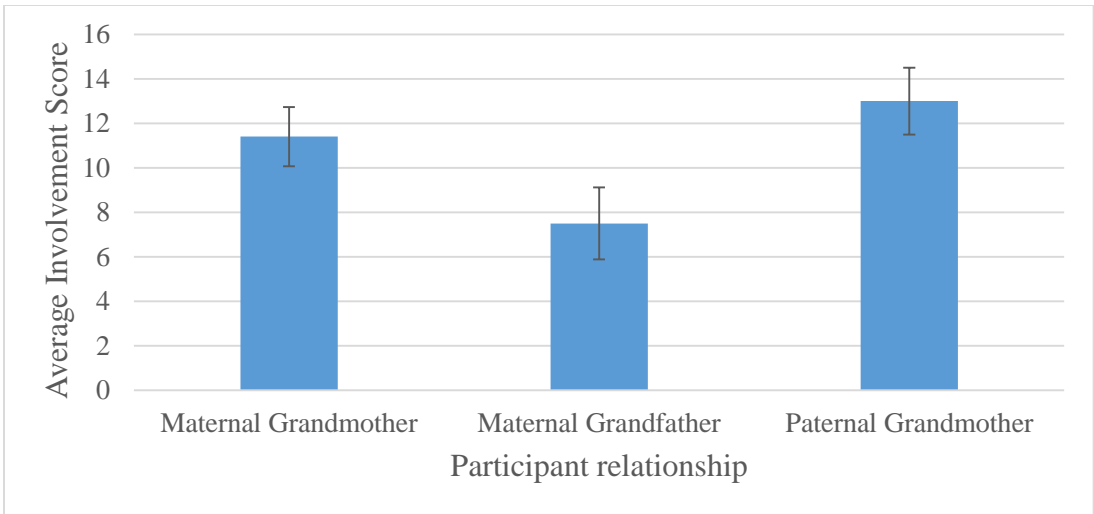


Figure 2.3 Average Involvement Score by Participant Relationship

2.4.4 Grandparents’ use and satisfaction with information

2.4.4.1 Use and satisfaction with sources of information. The most common source of information for participants was their grandchild’s parents ($n = 17$), followed by Internet research ($n = 13$) and pediatricians/family doctors/primary care physicians ($n = 9$). Genetics professionals were not common sources of information (Figure 2.4).

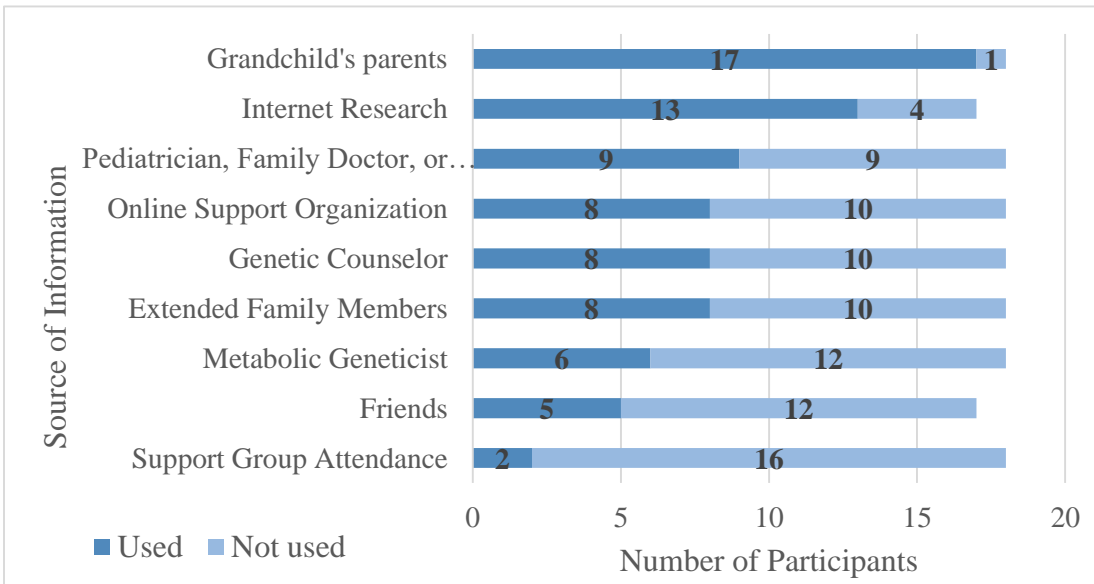


Figure 2.4 Sources of Information Used by Participants

In addition to being the most utilized source of information, grandparents were most satisfied with their grandchild’s parents being sources of information as well, with most participants reporting extreme satisfaction with their grandchild’s parents as sources of information ($n = 11$) (Figure 2.5).

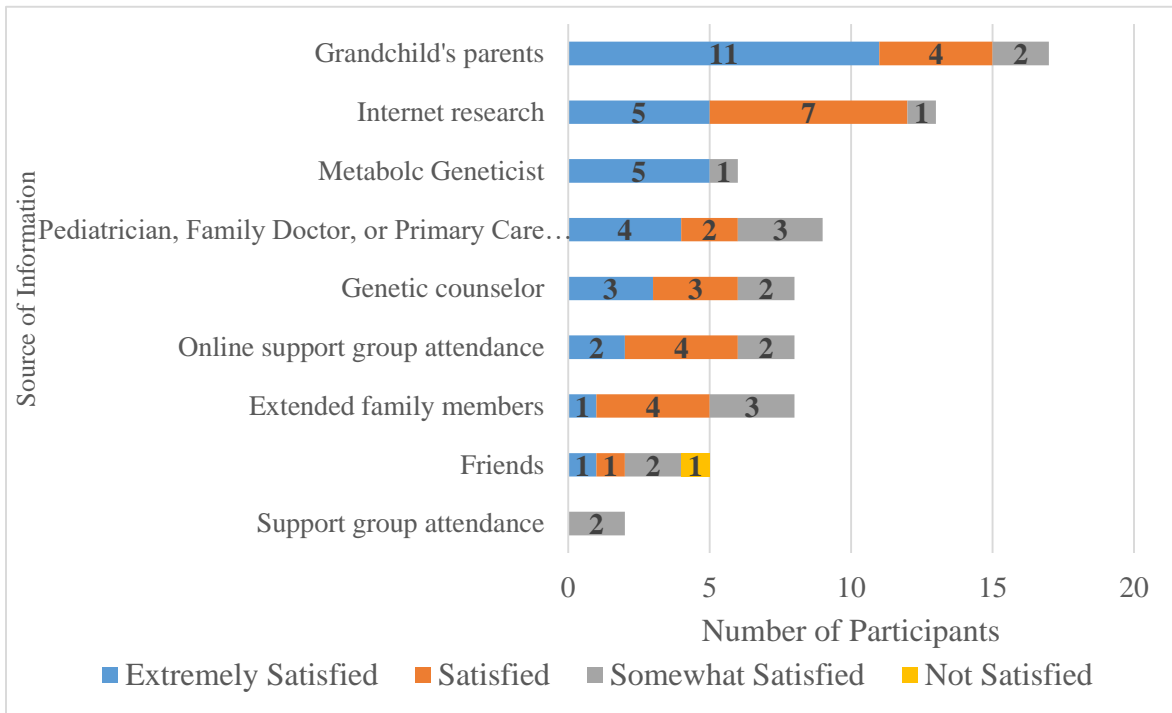


Figure 2.5 Participant Satisfaction with Sources of Information

2.4.4.2 Participant receipt and satisfaction with type of information. The majority of grandparents indicated that they had learned about genetic information related to Pompe ($n = 16$), medical complications associated with Pompe ($n = 17$), treatment options ($n = 17$), additional therapies ($n = 14$), the potential impact of Pompe on the family ($n = 15$), potential long-term outcomes for individuals with Pompe ($n = 13$), web-based information resources ($n = 14$), and printed information ($n = 13$). On the other hand, only a handful of grandparents had learned about the opportunity to speak with

other grandparents in a similar situation ($n = 4$) or referrals to individual/family counseling ($n = 3$) (Figure 2.6).

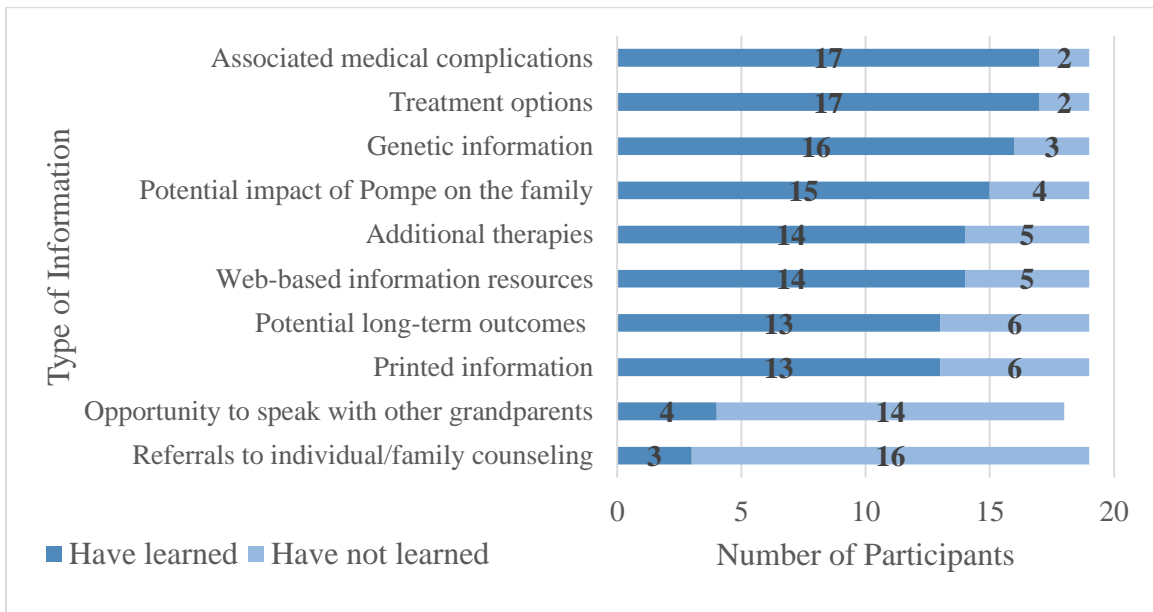


Figure 2.6 Types of Information Learned by Participants

Of the grandparents that received each type of information, participants were most satisfied with information about additional therapies for their grandchild and least satisfied with information they had received about referrals to individual/family counseling (Figure 2.7).

In addition to indicating their satisfaction with individual sources and types of information, respondents were asked to share their overall satisfaction with the information available to them as grandparents (Figure 2.8). Of the nineteen responses to this question, nearly half of the participants indicated being satisfied with the information available to them ($n = 9$). The average overall satisfaction with information was 3.6 out of 5 (Likert scale: 1-5), indicating that participants were between uncertain satisfaction and satisfaction with information.

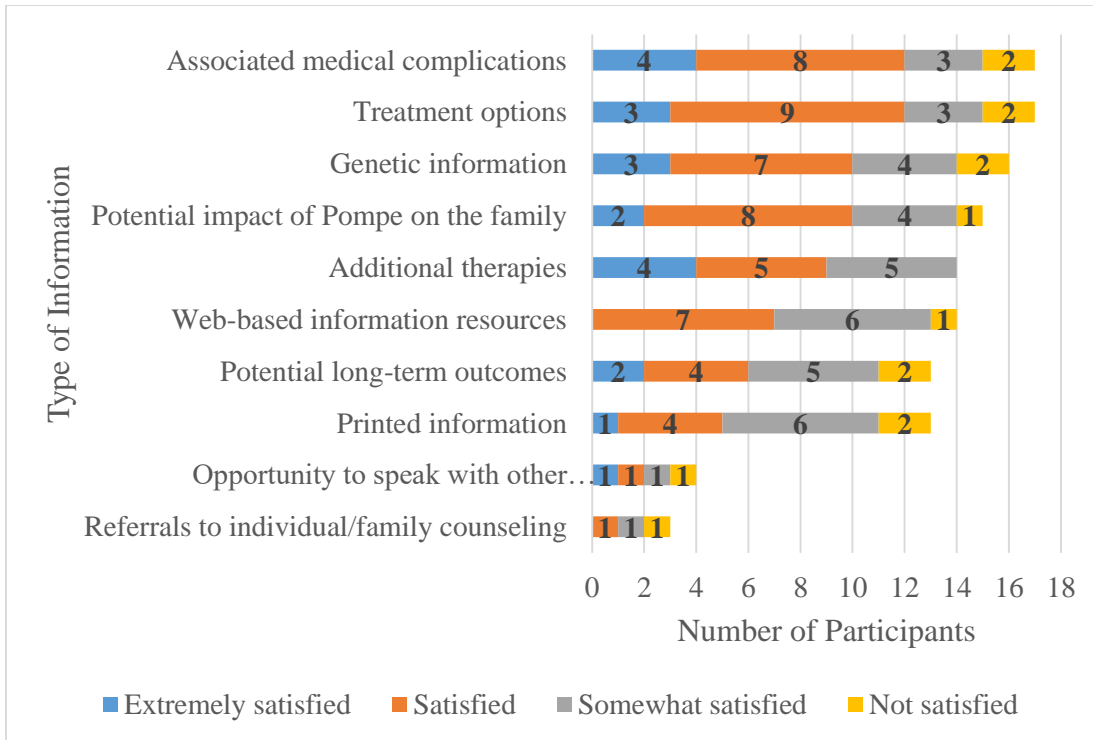


Figure 2.7 Participant Satisfaction with Types of Information Learned

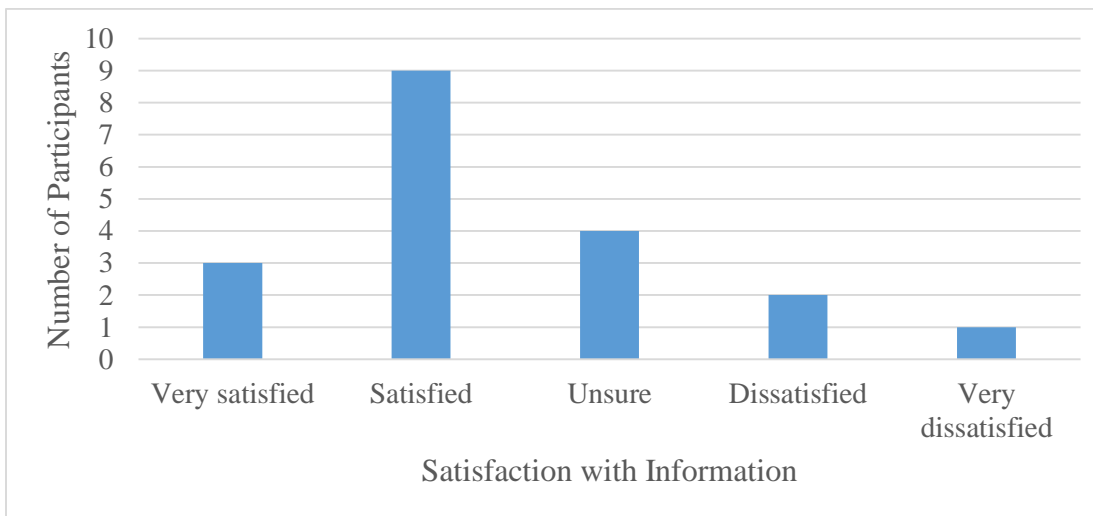


Figure 2.8 Participant Overall Satisfaction with Information Availability

Seventeen participants shared the most important information they had learned about their grandchild’s diagnosis. One common theme emerged from these responses.

Grandparents indicated that the most important information they had learned about Pompe was treatment. While treatment was a common theme, respondents focused on different topics surrounding treatment, including availability, utility, and effectiveness.

2.4.4.3 Genetic information. Most participants reported that they had learned about the genetics of Pompe ($n = 16$) (Figure 2.6), and of those participants that had learned about the genetics associated with Pompe, many were satisfied with the information they had gathered about the topic ($n = 7$) (Figure 2.7).

Only one respondent had undergone carrier testing. This participant did not answer questions regarding her carrier status or the impact carrier testing has had for her personally or for her relationship with her grandchild.

Participants were asked to report how well they felt they understood the genetics or inheritance of Pompe disease. Grandparents agreed that they understand the inheritance of Pompe with a 3.8 average agreement (Likert scale 1-5) (Figure 2.9).

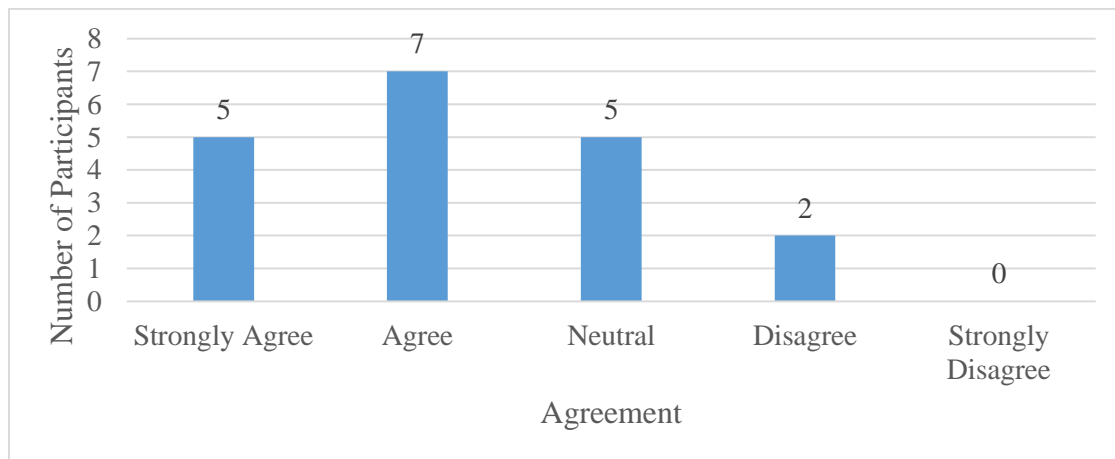


Figure 2.9 Participant Agreement with Comprehension of Genetics of Pompe

In response to a question about the inheritance of Pompe, three themes emerged (Table 2.6). Of the four participants who provided a risk assessment for two carriers to

have a child with Pompe, three provided the correct risk of 25%, while one participant reported a 50% chance for two carriers to have a child with Pompe.

Table 2.6 Themes from Participants’ Explanations of the Inheritance of Pompe

Theme	Frequency	Example quote
Uncertainty	3	“Not sure”
Carrier	15	“I know that my daughter, my granddaughter’s mother, carries the gene and my granddaughter’s father also carries the gene and my granddaughter got it from her parents.”
Risk assessment	4	“Both parents must carry the gene, and then there is a 1 in 4 chance that their child will have Pompe.”

2.4.4.4 Associations between grandparent involvement and satisfaction with information. There was a positive, statistically significant correlation between grandparent satisfaction with information and grandparent involvement, $r(12) = 0.66$, $p = 0.015$ indicating a strong relationship between grandparent satisfaction with information and involvement (Figure 2.10).

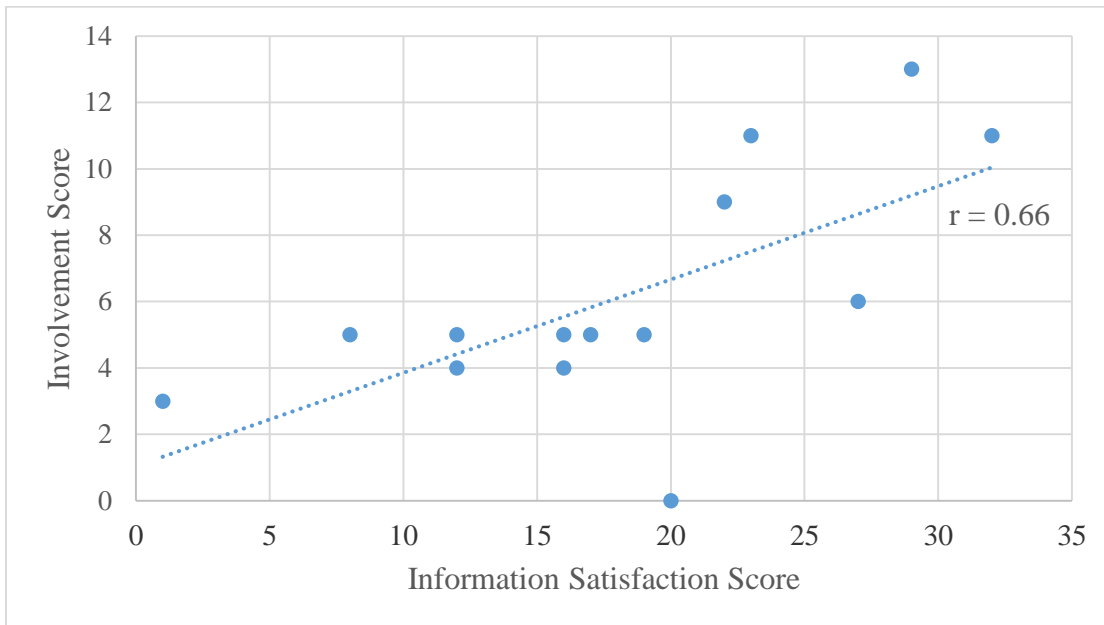


Figure 2.10 Relationship between Participant Satisfaction with Information and Involvement

2.4.5 Emotional support for grandparents of children with Pompe.

2.4.5.1 Use and satisfaction with sources of emotional support. Nineteen participants completed the question assessing their sources of emotional support. The majority of participants found emotional support from their grandchild's parents ($n = 18$). Other common sources of emotional support were spouses/significant others ($n = 17$), extended family ($n = 15$), Internet resources ($n = 15$), and religious or spiritual communities ($n = 12$). In-person support groups were the least commonly used source of emotional support ($n = 2$) (Figure 2.11).

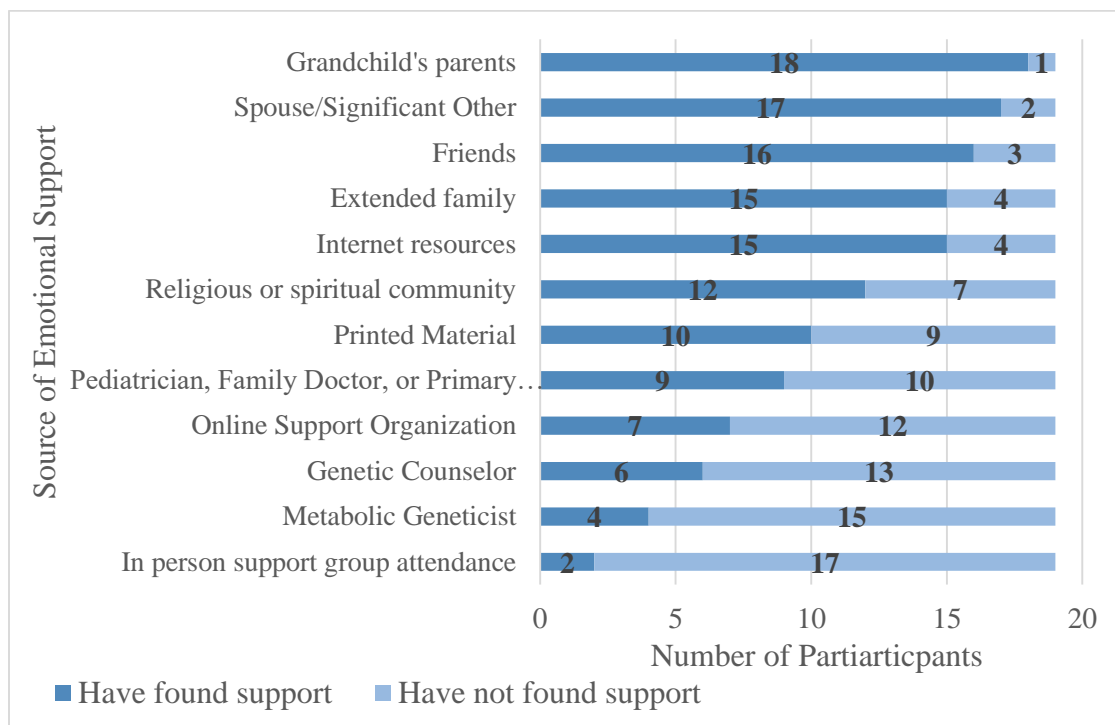


Figure 2.11 Sources of Emotional Support Used by Participants

In addition to being identified as common sources of emotional support, family and religion were also identified as the most important sources of emotional support for grandparents of children with Pompe (Table 2.7). Both family and religion were identified as the most important sources of emotional support in four responses.

Table 2.7 Sources of Support Most Valued by Participants

Theme	Frequency	Example quote
Family	7	“My main support is that we are a strong family unit.”
Religion	7	“My faith and church members.”

Moreover, emotional support from respondents’ grandchild’s parents was reported to be a highly satisfying source of emotional support, with the majority of participants who found support here being extremely satisfied ($n = 11$) (Figure 2.12).

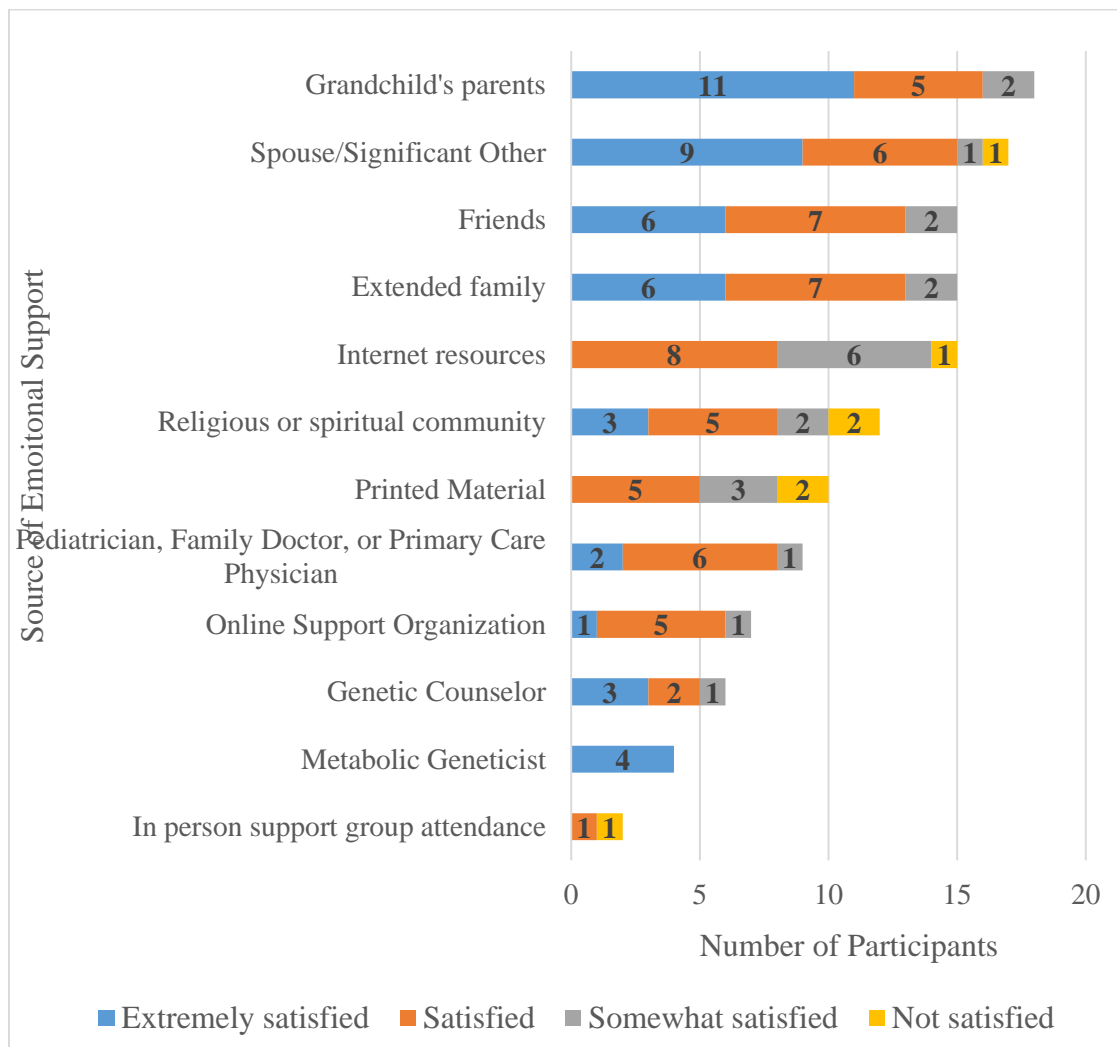


Figure 2.12 Participant Satisfaction with Sources of Emotional Support

In addition to reporting their satisfaction for each source of emotional support, participants rated their overall satisfaction with the emotional support they have received. The average overall satisfaction with information was 3.4 out of 5 (Likert Scale: 1-5), indicating that overall grandparents had some degree of uncertainty about their satisfaction with the emotional support they had received. Two participants reported being overall very satisfied, while eight reported being satisfied, five reported being unsure, and four reported being dissatisfied. Notably, none of the nineteen participants reported being very dissatisfied (Figure 2.13).

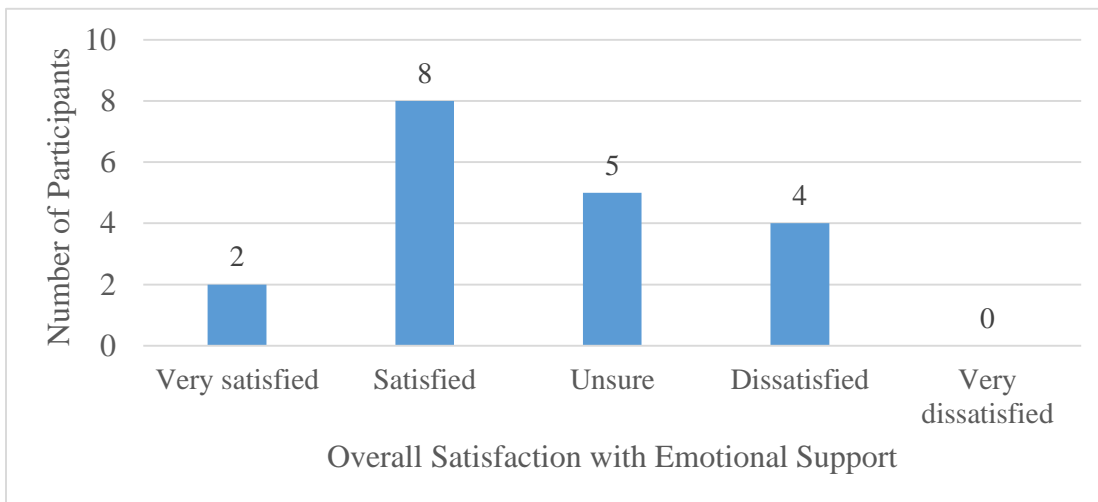


Figure 2.13 Participant Overall Satisfaction with Available Emotional Support

While none of the participants reported being very dissatisfied with the emotional support they had received, two participants commented in previous open-ended questions on the lack of emotional support. One of these participants commented:

In some people I have found excellent support. However, it is very hard for most people to understand the ramifications of Pompe. Most people have never heard of Pompe, including many in the medical community. Those of us who love Pompe patients feel very alone at times, and helpless

in the face of this disease. My husband and I are part of the support system, but often feel the need for support for ourselves. There is so much pain looking at the future for our sweet, brave, and strong granddaughter

2.4.6 Personal impact of having a grandchild with Pompe. Eighteen participants responded to a question exploring how grandparents’ lives are impacted by having a grandchild with Pompe. Travel was discussed by many participants in the context of increased travel to spend time with their family and in the context of reduced leisure travel to accommodate for additional financial costs and to be available for medical appointments. Impacts on work-life were mentioned and included postponing retirement, retiring early, or taking off more days from work. In response to another question allowing grandparents to share any other thoughts, three participants responded by expressing frustration with their grandchild’s diagnosis. The theme of double grief presented itself three times in three different questions throughout the survey (Table 2.8).

Table 2.8 Psychosocial Impacts of having a Grandchild with Pompe

Theme	Frequency	Example Quote
Travel	7	<i>“I travel more often to see them...I fly there 3-4 times a year and drive there 1-2 times a year.”</i> <i>“Vacations have been eliminated.”</i>
Impact on work life	3	<i>“I postponed my retirement in order to help financially.”</i> <i>“Retired but became full time babysitter.”</i>
Frustration with the diagnosis	3	<i>“This is very frustrating to see a small child full of needles and medical devices and trying to explain to him why...”</i>
Double grief	3	<i>“In reality though, my heart aches not only for my precious granddaughter, whom I love more than life, but my heart aches for my daughter and son-in-law. As a parent, I have always been able to take care of their aches and pains, but this time it is different. As a mother, it is very difficult to watch your child suffer and not be able to ‘make it better.’”</i>

Only one participant openly expressed the sadness directly related to her grandchild and her loss. Within her comment, she urged others to remember that Pompe affects an entire family. She shared:

When our grandchild died, we felt like we were kind of ignored when it came to condolences. We had lost someone we cared for and loved too but I think people forgot that we were hurting too. People would say things like, I know your son and daughter in law are just devastated. I feel that people could have been more aware that it was something that affected our whole family [sic].

Interestingly, the theme of carrier testing emerged when grandparents were given the opportunity to share other thoughts. Three different reasons for desiring genetic carrier testing for grandparents were expressed. First, carrier testing for grandparents was seen as a way to “advise other family members.” A different participant saw grandparental carrier testing as a “probable prevention of this life threatening disease.” The last reason offered to support genetic testing in grandparents was a way to understand where Pompe originated in the family.

2.5 Discussion

This is the first study to investigate the grandparent experience of having a grandchild with Pompe and to provide insight into the experiences of extended family members of children with autosomal recessive, treatable conditions. Specifically, this study explored the roles, involvement, informational and emotional support needs of grandparents of children with Pompe as well as the psychosocial impact of having a

grandchild with Pompe disease. Twenty-one participants' responses to an online survey mixed-mode survey were analyzed.

2.5.1 Grandparent roles and involvement. Participants most commonly and most frequently felt they provided emotional support to their family. In fact, participants provided emotional support to their family statistically significantly more frequently than they provided long-term child care, medical assistance/management of Pompe, running errands, and financial support. These findings support previous literature in which parents report that all grandparents provide emotional support more commonly than other types of support (Seligman et al., 1997). Two reasons could explain why participants in this study provide emotional support most commonly and most frequently. First, participants could have an understanding that emotional support is most valued by their family. Previous studies investigating parental perceptions of grandparent involvement revealed that parents find emotional support provided by grandparents most important (Findler, 2014; Lee & Gardner, 2010; Seligman et al., 1997). Second, participants may feel the most confident in a role that involves emotional support. A study by Gardner et al. (1994) found that grandparents feel most effective in roles that involve emotional support. Therefore, it is possible that participants in this study who share the same sentiments offer emotional support most commonly because it is the most valued and most comfortable role.

Interestingly, participants reported a difference between their most commonly offered support and their unique role in the family. When grandparents were asked to describe their unique roles in the family, participants more frequently described practical support roles as opposed to emotional support roles. In their descriptions of their unique

family roles, nine participants specifically mentioned practical support roles, while six specifically mentioned emotional support. Furthermore, four of the nine grandparents who explicitly described practical support mentioned only practical support as their unique role, whereas only one of the six participants who explicitly described emotional support mentioned only emotional support. Practical support roles included babysitting their grandchild with Pompe and assisting with medical management. Both of these roles require a solid understanding of the natural history, treatment, and management of Pompe disease for grandparents to properly care for their grandchild.

When asked to describe any differences between grandparenting their grandchild with Pompe and their unaffected grandchildren, half of participants described an increased awareness of the medical needs and limitations of their grandchild with Pompe. Grandparents stated that their grandchild “obviously needs more help” with mobility and must be “watched more closely than others due to balance issues and choking.” One participant even explained that her grandson “doesn’t get to come to [her] house because of the distance to the hospital.” A previous study identified this same phenomenon. In a study from 1994, thirty-two grandparents were asked to explain how their grandchild’s disability altered their relationship and identified that participants who reported changes in grandparenting a child with special needs mentioned needing to accept the restrictions related to their grandchild’s diagnosis (Gardner et al., 1994).

Taken together, grandparents’ perceived unique role of practical support in the family and their heightened awareness of the limitations and medical needs imposed upon their grandchild by a medical condition highlight the importance of enabling grandparent access to accurate information. If grandparents are fulfilling roles that require medical

management of Pompe, they should be appropriately educated about proper management. As grandparents demonstrated that their grandchild's medical needs and limitations alter their grandparenting role, grandparents should have access to accurate information to ensure that they understand the realities of the limitations associated with their grandchild's diagnosis.

One grandparent described the role she has with the unaffected sister of her grandchild with Pompe disease. She discussed how she does "things like school pick ups" and is aware of how this unaffected sister is "sensitive to her sister's fears and discomforts." This description reflects the experiences reported by grandparents of children with congenital heart defects. In a study by Ravindran & Rempel (2011), grandparents discussed the concern they have for the siblings of their ill grandchildren. The authors call this experience "triple grief," which elaborates upon the double grief grandparents feel by including the grief that grandparents feel for siblings of grandchildren with a life-threatening condition as well as their grief for their own child having to cope as the parent of child with a life-threatening condition.

Previous studies have identified that maternal grandparents are the most commonly involved grandparents (Bruns & Foerster, 2011; Hornby & Ashworth, 1994; Seligman et al., 1997; Shaw, 2005; Trute, 2003). Responses from paternal grandmothers in this study had the highest average Involvement Score, although there was no statistically significant differences between involvement of grandmothers and grandfathers or between maternal and paternal grandparent involvement. Given that statistical analysis did not verify an actual higher level of involvement by paternal grandmothers, it could be argued that this study is consistent with previous literature.

More than half of the participants were maternal grandmothers ($n = 11$). The method through which we distributed the survey would have selected for more involved grandparents, thus the higher frequency of maternal grandmothers who participated in the study could suggest that maternal grandmothers have a higher level of involvement.

2.5.2 Grandparent use and satisfaction with source and type of information.

Grandchildren's parents were the most frequently used source of information ($n = 17$) and a highly satisfying source of information, with the majority of participants who received information from their grandchild's parents being extremely satisfied ($n = 11$). This high level of satisfaction with family as sources of information and emotional support has been demonstrated before (Schilmoeller & Baranowski, 1998).

Understanding that grandparents rely upon family for information should increase our awareness that the information we present to our families travels from family member to family member. Therefore, we could consider better equipping patients with the tools they need to discuss complex medical concepts with their family members.

Medical professionals were less commonly reported as sources of information, which supports previous research. Limited interactions between health care providers and grandparents have been reported before (Findler, 2008; Lee & Gardner, 2010; Scherman et al., 1995; Vadasy et al., 1986). When participants did receive information from healthcare professionals, most participants were extremely satisfied or satisfied with the healthcare providers as sources of information, suggesting that medical professionals manage interactions with grandparents well. Therefore, healthcare providers should be encouraged to include grandparents more frequently.

Grandparents reported rarely receiving information about referrals to individual/family counseling and the opportunity to speak with other grandparents. The paucity of support groups for grandparents could explain why grandparents rarely received information about the opportunity to speak with other grandparents. On the other hand, most grandparents reported that they had received medical information about Pompe including genetics, medical complications, prognosis, treatment, and additional therapies. Information about the treatment for Pompe was identified as the most important information for participants. Overall, participants reported being satisfied with the information available to them as grandparents. Grandparent satisfaction with information is a unique finding to this study. Previous literature describes grandparents' desire for more information or a dissatisfaction with the information they received (Katz & Kessel, 2002; Vadasy et al., 1986). A possible explanation for this difference is the increased use of and familiarity with the Internet. Most grandparents ($n = 13$) in this study reported using the internet as a source of information. Since these previous studies documenting grandparents' desire for more information, Internet use has become more widespread in daily life. It is possible that the increased use of the Internet has helped to satisfy grandparents' desire for more information. Furthermore, this is an encouragement to ensure that accurate, appropriate information is available for grandparents on the Internet.

2.5.2.1 Genetics of Pompe. Participants in this study demonstrated an excellent understanding of the cause of their grandchild's condition. Fifteen out of eighteen participants attempted to explain the inheritance of Pompe disease, with 14 out of the 15 accurately explaining both autosomal recessive inheritance and recurrence risk. This level

of understanding has not been reported commonly in the literature (Katz & Kessel, 2002; Scherman et al., 1995). The differences in the conditions studied could explain the discrepancy between this study and previous literature. Cerebral palsy, severe intellectual disability, epilepsy, autism, and severe ADHD were some of the grandchild diagnoses included in these prior studies (Katz & Kessel, 2002; Scherman et al., 1995). In these previous studies, it is understandable that grandparents would express confusion about the cause of their grandchild's condition, as oftentimes the etiology for such conditions remains unknown even to medical professionals.

A study including participants more similar to participants in this study described a persistent guilt felt by grandmothers identified as carriers for X-linked conditions after having affected grandchildren. The guilt felt by these grandmothers suggests that they had a good understanding of the cause of their grandchild's condition (Lehmann et al., 2011). Therefore, it could be concluded that when the cause of a grandchild's condition is known, grandparents have a good understanding of that cause.

Again, the Internet could explain these more recent findings of better grandparent understanding of the cause of a grandchild's condition. However, better grandparent understanding of the etiology of their grandchild's diagnosis could also reflect healthcare providers' increasing awareness and understanding of how to effectively communicate genetic information. In recent years, a number of research studies have aimed to understand the communication of genetic information and how to best meet patient needs in such discussions (Gale, Pasalodos-Sanchez, Kerzin-Storarr, & Hall, 2010; Pieterse, van Dulmen, Beemer, Bensing, & Ausems, 2007). It is possible that in the time since

earlier studies, more effective communication of genetic information has evolved, thus resulting in better grandparent comprehension of genetic etiologies.

Though only one participant reported having had carrier testing for Pompe, three respondents did express an interest in carrier screening. The reasons grandparents offered for desiring this included obtaining useful information for family members, understanding the origin of their grandchild's diagnosis, and having information that may prevent a child from being born with Pompe disease. Participants in this study clearly had a good understanding of the utility of carrier screening and desired carrier screening for these reasons. Learning that this interest in carrier testing is present in the grandparent population brings into question how the current practice of carrier screening could meet this interest.

However, there was a discrepancy between grandparents' perceived understanding of the inheritance of Pompe and their comprehension of the inheritance of their grandchild's diagnosis. Seven of nineteen participants either disagreed that they understood the inheritance or were neutral about their understanding, yet fourteen grandparents accurately described the inheritance. Therefore, grandparents have received accurate information about this topic, yet grandparents may be uncertain about the information they have received. This perception of misunderstanding may be consistent with what has been reported in previous literature as a desire for more information about a grandchild's diagnosis (Katz & Kessel, 2002; Scherman et al., 1995).

2.5.2.2 Relationships between involvement and satisfaction with information.

This study described a positive, strong, statistically significant relationship between grandparent involvement and grandparent satisfaction with information. It is unclear at

this point if involvement leads to higher satisfaction with information, or if higher satisfaction with information leads to more involvement. However, knowing this relationship exists and given the research describing the benefits gained from grandparental involvement for both grandparents and the family as a whole, we should make an investment in understanding how to better satisfy grandparents informational desires to promote grandparent involvement (Green, 2001; Heller et al., 2000; Vadasy et al., 1986).

2.5.3 Satisfaction with emotional support. Family and religion were identified as common and important sources of emotional support for respondents, and grandparents were most satisfied with the emotional support they received from their grandchild's parents. These findings are consistent with a previous study that found that grandchildren's parents and religion were important sources of emotional support for grandparents (Hastings, 1997). Moreover, the high level of participant satisfaction with emotional support from their grandchild's parents affirms the same finding in a previous study (Schilmoeller & Baranowski, 1998).

Other sources of support used by our participants were used with different frequencies reported in existing literature. Nearly all of our participants found support from their significant other ($n = 17$), and only two participants found support from in person support groups. The number of participants who found support from their partners is considerably higher than 50% of participants who found emotional support from their spouses in a study by Scherman et al. (1995). Another study found that nearly 25% of their participants had found support from in-person support groups, while only two of our participants found support here (Schilmoeller & Baranowski, 1998).

Grandparents reported an average overall satisfaction with emotional support of 3.4, indicating that participants had some uncertainty about their satisfaction with the emotional support they had received. Interestingly, no participants gave detailed descriptions of positive experiences with emotional support, but two participants did openly discuss the lack of emotional support they had received. The overall uncertain satisfaction with emotional support coupled with these discussions about the lack of emotional support received suggests that there is room for increasing the awareness of grandparent support needs and the availability of emotional support resources.

2.5.4 Psychosocial impact of having a grandchild with Pompe disease. In previous literature, grandparents have consistently reported the experience of double grief, an experience characterized by grandparent grief for the shift in expectations for their grandchild and for the stresses and burdens placed on their child, the parent (Hastings, 1997; Katz & Kessel, 2002; Ravindran & Rempel, 2011; Scherman et al., 1995; Vadasy et al., 1986; Woodbridge et al., 2009). Though participants in this study were not specifically asked questions about double grief, three different respondents described double grief in three different questions, with one participant claiming that her “heart aches not only for [her] precious granddaughter...but [her] heart aches for [her] daughter and son-in-law.” This is the first study that documents the experience of double grief in grandparents of children with Pompe, which supports the idea that double grief is a universal experience for grandparents.

Grandparents in this study also described the impact they felt on their personal life, including changes in travel and employment plans. These findings support similar findings in previous literature, in which grandparents discussed the changes they made in

travel plans and retirement plans (Woodbridge et al., 2011). In both the previous literature and our study, participants describe these adjustments to their plans as voluntary and often unsolicited by the family. Furthermore, previous findings have identified frustrations that grandparents have from these changes in their plans, feeling as if they have been taken for granted (Miller et al., 2012). Understanding that grandparents may feel unappreciated as they willingly make these adjustments to their plans, even without being asked by the family, provides insight into the workings of the family system and could help uncover some of the underlying tension that may exist between grandparents and parents.

2.5.5 Implications for practice. Results from this study emphasize that additional efforts could be invested to further support members outside of the nuclear family. Here, we present suggestions for healthcare providers, especially genetics healthcare professionals.

- **Address grandparents directly.** Grandparents need and value both information about their grandchild's diagnosis and emotional support as they adjust to their grandchild's diagnosis. Though grandparent-genetic professional interactions are limited, grandparents generally report high levels of satisfaction with genetics healthcare providers as sources of information and emotional support. Therefore, genetics professionals have the skills to successfully address grandparent needs and should initiate these interactions. Addressing grandparent needs could be as simple as exploring grandparents' questions and acknowledging their unique roles and complex emotional experiences.

- **Prepare the family for grandparents' needs.** Grandparents rely heavily upon their grandchildren's parents for information and emotional support. At a time when parents need information and support themselves, family psychosocial functioning can be optimized by preparing families for the ways in which grandparents or other extended family members become involved and the new roles grandparents will rely upon parents to fill. To facilitate familial adaptation to a diagnosis and communication of complex medical information, be certain to provide families with appropriate informational resources and referrals. By extension, consider developing information resources about the diagnosis directed specifically toward grandparents.
- **Encourage communication between parents and grandparents.** Grandparents can be both a vital source of support and a stress for families when a child has a genetic condition. Parents should be encouraged to clearly communicate what help is needed and at what times, while grandparents should be encouraged to openly express when and how they are available for assistance.
- **Know the community.** Resources for grandparents are limited, so genetics professionals should take time to explore the options for grandparent support in their areas. Providing an outside source of support for grandparents could help alleviate stress between family members. Few grandparents learned about the opportunity to meet with other grandparents in similar situations. Because resources for grandparents may be limited, medical professionals may consider connecting grandparents in similar situations with each other.

2.5.6 Limitations and Future Studies. As Internet-based research, this study has common limitations including unknown response rates and ascertainment bias. As no support organizations specifically for grandparents of children with Pompe exist, information about the survey was shared through general support groups. Participants who saw this information independently and pursued the opportunity to participate in the survey would likely be highly motivated and involved grandparents. However, it is more likely that participants learned about the survey through their family members who are associated with these support organizations, which again selects for highly motivated and involved participants with good relationships with their family. This method of distribution prevented us from learning about the experiences of grandparents who are less involved. Moreover, the participant population was rather homogenous, as all participants reported Caucasian ethnicity. Therefore, the results from this survey may lack the ability to be generalized to the total population or to other ethnicities. Finally, the method of survey distribution prevents us from knowing who actually accessed the survey.

Additional research in this topic could include a more in-depth analysis of grandparent desires for information. Exploring grandparent experiences when a grandchild has a different treatable, autosomal recessive conditions such as cystic fibrosis could broaden our understanding of grandparent needs. The limited interactions between healthcare professionals and grandparents serves as another source of future research. Future studies could investigate possible reasons for limited interactions between healthcare professionals and grandparents. Research specifically focused on genetic counselors' interactions with grandparents could investigate genetic counselors' current

practices and involvement with grandparents. Studies including pairs of grandparents and parents together could provide more insight into the tensions that may be present between grandparents and parents. Most importantly, additional research that focuses on grandparents who are less involved to understand the reasons for their level of involvement could be pursued.

2.6 Conclusions.

Research regarding the roles, needs, and experiences of grandparents demonstrates a complex grandparent experience when a grandchild has special needs or a disability but has been restricted to grandparents of children with non-Mendelian conditions without treatment. Grandparents of children with autosomal recessive, treatable conditions have not been the focus of previous studies. In addition to being the first study to investigate the experiences of grandparents of children with Pompe, the results from this study address the gap in the literature by exploring the roles, needs, and experiences of grandparents of children with a treatable, autosomal recessive condition. Participants described their roles and involvement in their families, their experiences with gathering information about Pompe and receiving emotional support, and the psychosocial impact of having a grandchild with Pompe. Responses to a mixed mode survey allowed for the development of recommendations for clinical practice for genetics healthcare providers.

Grandparents provided emotional support most frequently to their families, yet they perceived their unique role in the family providing both practical and emotional support. Grandchildren's parents were primary sources of both information and emotional support for grandparents, and most grandparents reported extreme satisfaction

with their grandchild's parents as sources of information and emotional support.

Understanding that the information provided to the patient's parents is communicated throughout the family is a reminder that genetics services impact more than the direct recipients and highlights the importance of providing families with resources to communicate complex information to extended family members.

Grandparents described complex psychosocial impacts of having a grandchild with Pompe disease, including a heightened awareness of their grandchild's limitations and medical needs and the well-documented experience of double grief, emphasizing that impact of a child's diagnosis is felt beyond the nuclear family. Knowing that grandparents experience so many emotions should encourage genetics health care providers to be aware of available resources in the community to facilitate grandparent adjustment and adaptation to a grandchild's diagnosis.

Genetics healthcare professionals were not commonly utilized as sources of information or emotional support. However, when grandparents did receive information or emotional support from geneticists or genetic counselors, they generally reported being satisfied with genetics professionals as resources. Grandparent satisfaction with genetics professionals as sources of support serves to encourage healthcare providers to directly address grandparents and extended family members.

Healthcare providers, and especially genetic counselors, are dedicated to ensuring and facilitating optimum psychosocial functioning for patients. Understanding the roles, needs, and experiences of grandparents provides insight into the complex psychosocial context in which patients reside. Increased familiarity with and awareness of the needs and multifaceted experiences of grandparents will assist healthcare providers in more

consistently addressing grandparent needs and providing more comprehensive care for patients.

Chapter 3: Conclusions

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Appendix A – Invitational Letter to Participate in Survey

Dear Grandparent,

You are invited to participate in a research study for a Master's thesis project at the University Of South Carolina School Of Medicine. The objective of the study is to gain a better understanding of the needs of grandparents when their grandchild is diagnosed with Pompe disease. We would like for your input to

- Characterize the role and involvement of grandparents of individuals with Pompe
- Identify the informational and emotional needs of grandparents
- Explore the psychosocial impact of a diagnosis of Pompe on the grandparent

The online survey will take approximately 15-20 minutes to complete; the time to complete the survey will vary depending on the length of your responses. Your participation in this survey is anonymous. This means that no one will be able to connect your responses to your identity. Although we are unable to offer direct compensation for your participation in this study, we hope that your responses will benefit you indirectly through the increased awareness of grandparents' information and support needs.

We are happy to answer any questions you have about the study. You may contact me or my faculty advisor Janice Edwards using the contact information below. If you have any questions about your rights as a research participant, you may contact the Office of Research Compliance at the University of South Carolina at 803-777-7095.

Thank you for your consideration. If you would like to participate, please follow the link to the survey:

<https://www.surveymonkey.com/r/gparent-pompe>

Sincerely,

Natasha Rudy, BA, BS
Genetic Counseling Intern
University of South Carolina
Genetic Counseling Program
2 Medical Park, Suite 208
Columbia, SC 29203
Phone: 864-992-8469
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2 Medical Park, Suite 208
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Appendix B – Invitational Flyer to Participate in Survey

CALLING GRANDPARENTS OF CHILDREN WITH POMPE:

WE WANT TO HEAR YOUR THOUGHTS

Are you the grandparent of a child with Pompe? We would like to hear about your experience. Specifically, we want to learn more about the information and emotional support grandparents need when their grandchild has Pompe. If you would like to share your experiences and thoughts with us, please follow the link below to the online survey. There you can also find more details about the survey.

Not a grandparent of a child with Pompe? Please share this information with a grandparent you know.

SURVEY LINK:

www.surveymonkey.com/r/gparent-pompe

SURVEY DETAILS:

Who we want to hear from:

- Grandparents whose grandchild was diagnosed with Pompe at or before age 18
- Grandparents who have lost a grandchild with Pompe

Reason for the survey:

This study is part of a Master's Thesis project conducted through the University of [South Carolina School of Medicine](#). We want to bring awareness to the unique needs of grandparents.

CONTACT INFORMATION

Natasha Rudy, BA, BS
Phone: 864-992-8469
Email: rudynl@email.wofford.edu

Janice G, Edwards, MS, CGC
Phone: 803-545-5706
Email: jedwards@uscmed.sc.edu

Appendix C – Guide for Pilot Study

GUIDE: *Text in this font is to be read aloud to/asked of participant*

INSTRUCTIONS TO BE READ TO SUBJECT

You are being asked to review the following version of a web-based consent document and questionnaire. We are trying out these questions on a few people so that we can improve them. We are primarily interested in the way you arrive at your answer and any problems you encounter, such as problems with the wording of questions, questions that are difficult to understand, questions that are hard to answer, or questions that make little sense.

I will read the questions to you out loud. Then I would like for you to read the answer choices and answer the questions by “thinking aloud”—just tell me everything that comes to mind as you answer the questions, whether you think it is important or not. You can respond based on your personal experience of being a grandparent of a child with Pompe.

I will occasionally ask additional follow-up questions about how you come up with your answers and how you interpret the questions. If any question seems unclear, is hard to answer, or doesn’t make sense, please tell me and describe your reactions or opinions. If you are uncomfortable answering any question, please be sure to let me know. Do you have any questions before we start?

THINK-ALoud PRACTICE

Let’s begin with a practice question. Remember to think aloud as you answer. Try to visualize the place where you live, and think about how many windows there are in that place. As you move throughout your home to count the windows, tell me what you are seeing and thinking about.

OK, now let’s move on to the questionnaire we’re testing...

REVIEW OF THE CONSENT DOCUMENT

The first page of the questionnaire is the consent portion of web-based questionnaire. When participants log on to the survey URL, they will read this information before entering the question portion of the survey. Please read aloud the consent portion while thinking aloud. Be sure to tell me everything you are thinking, if something is unclear or does not make sense. Let’s get started:

The Information and Emotional Support Needs of Grandparents Whose Grandchild has a Genetic Condition: A Focus on Pompe Disease

Thank you for your interest in our study. This survey is for a Master's thesis project at the

University of South Carolina School of Medicine. The objective of this study is to gain a better understanding of the needs of grandparents when their grandchild is diagnosed with Pompe. We would like for your input to:

1. Characterize the role and involvement of grandparents of children with Pompe
2. Identify the informational and emotional support needs of grandparents
3. Explore the psychosocial impact of a diagnosis of Pompe on the grandparent.

This online survey will take approximately 15-20 minutes to complete; the time to complete the survey will vary depending on the length of your responses. Your participation in this survey is anonymous. We will not ask for any information that could identify you, and no one will be able to connect your responses to your identity. Although we are unable to offer direct compensation for your participation in this study, we hope that your responses will benefit you indirectly through the increased awareness of grandparents' information and emotional support needs.

We are happy to answer any questions you have about the study. You may contact Natasha Rudy or the faculty advisor Janice Edwards using the contact information below. If you have any questions about your rights as a research participant, you may contact the Office of Research Compliance at the University of South Carolina at 803-777-7095.

Thank you for your consideration. If you would like to participate, please click the "Next" button below.

By clicking the "Next" button, you are consenting to voluntarily participate in this anonymous research study.

If you have any questions about the survey or the research study, please contact
Natasha Rudy, BA, BS
Genetic Counseling Intern
University of South Carolina
Genetic Counseling Program
2 Medical Park, Suite 208
Columbia, SC 29203
Phone: 864-992-8469
Email: rudynl@email.wofford.edu

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Email: jedwards@uscmcd.sc.edu

1. *How easy or difficult was it to understand the information about the study?*
2. *Based on what you just read, what would you say is the purpose of the study?*
3. *What did you think about the amount of information provided? Too much? Too little?*
 - a. *What questions do you have that were not answered by reading this?*
4. *After reading this, are you inclined to participate in the study?*
5. *Do you have any concerns about participating in the study?*

Review of Web-based Survey Questions

Let's move on to review the web-based questionnaire. Here are some examples of problems with questions that you may help identify:

- *Any wording or text is too complicated*
- *Any wording that could be adjusted to minimize sensitivity*
- *The question uses technical terms that are undefined, unclear or vague*
- *There are multiple ways to interpret the question*
- *The question has more than one implied question (a double-barreled question)*
- *The question makes inappropriate assumptions about your experience*
- *Participants are unlikely to know the answer or have trouble remembering the details*
- *The question implies a socially acceptable response*
- *An open-ended question is inappropriate or difficult to answer*
- *The response choices do not match the question being asked*
- *There are multiple ways to interpret response choices or*
- *The meaning of response choices overlap*
- *Your answer choice is missing from the list of provided responses*
- *The order of the response choices does not make sense*
- *Or any other problem not identified above*

Any questions about those examples? Be sure to tell me everything you are thinking, if something is unclear or does not make sense. OK - Let's get started:

SECTION 1: GRANDPARENT DEMOGRAPHICS

1. What is your current age in years? _____

2. Please tell us what ethnicity you consider yourself from the list below. If none of these match, please put this in the other box
white
Black or African American
American Indian or Alaskan Native
Asian
Native Hawaiian or other Pacific Islander
From multiple races
Other _____

3. In what state do you live?

4. Please indicate your current relationship status:
 Married
 Widowed
 Divorced
 Separated
 In a domestic partnership or civil union
 Single, but cohabitating with a significant other

Single, never married

5. Do you identify with any of the following religions? (Please select all that apply.)

Protestantism

Catholicism

Christianity

Judaism

Islam

Buddhism

Hinduism

Native American

Inter/Non-denominational

No religion

Other _____

Overall, did you feel comfortable answering those questions?

Did you feel that your responses were represented in the answer choices?

SECTION 2: GRANDCHILD DEMOGRAPHICS

Please share with us some information about your grandchild with Pompe.

6. How old was your grandchild when he/she was diagnosed with Pompe? _____

7. What subtype of Pompe was your grandchild diagnosed with?

Classic infantile (Pompe with heart involvement)

Non-classic infantile or Atypical infantile (Pompe without heart involvement)

Juvenile or Late onset (diagnosed after the first year of life, without heart involvement)

Unsure

How confident are you in your answer to this question?

How familiar are you with the subtypes of Pompe?

What does “with heart involvement” mean to you?

8. Is your grandchild living or deceased?

Living

Deceased

SECTION 3: GRANDPARENT/GRANDCHILD RELATIONSHIP

Please share with us some information about your relationship with your grandchild.

9. Currently, how old is your grandchild with Pompe? _____

10. Please select your relationship to your grandchild?
- Maternal grandmother
 - Maternal grandfather
 - Paternal grandmother
 - Paternal grandfather

***What does the phrase “maternal grandmother” mean to you?
How would you explain how you are related to your grandchild?***

11. What is your primary role in your grandchild’s care? (Are you the legal guardian of your grandchild?)
- Legal guardian
 - Source of support for the family

How well do you feel these answer choices fit your response?

SECTION 4: UNDERSTANDING GRANDPARENTS’ ROLES

Grandparents often provide support in a number of ways: 1) practical support by running errands for the family or watching their grandchildren and 2) emotional support to the family by lending a listening ear, acting as a confidant, and providing encouragement or solace. Please provide some information about your role in the family (how you support your family)

12. How far do you live from your grandchild with Pompe disease?
- I live with my grandchild
 - Less than 30 miles
 - 30-100 miles
 - More than 100 miles

How did you choose your answer for this question?

13. In what ways are you involved in supporting your grandchild and other family members? For the ways in which you provide assistance, please indicate how frequently (often) you offer the specific support. If you do not perform a certain task, or if the task does not apply to your situation, please select, “Not applicable.”

***What does “respite care” mean to you?
How are you deciding how often you perform a task?
How well do you feel the answer choices fit your responses?
Are there other forms of support that you feel you offer that are not listed?***

14. How would you describe your unique role in your family unit?

Can you restate this question in your own words?

15. Is your grandparenting role for your grandchild with Pompe different from your role with grandchildren who do not have Pompe? If yes, please describe how your role is unique for your grandchild with Pompe.

No

Yes _____

SECTION 5: INFORMATIONAL NEEDS

Please tell us about the information you received regarding Pompe and your grandchild's diagnosis.

16. Please indicate your level of satisfaction with each resource that helped you collect information about Pompe. If you did not use a resource to gather information, please select, "Not applicable."

How easy or difficult is a question in this format to answer?

17. To the best of your recollection, please indicate your satisfaction with the information you received or gather about Pompe. If any of the following items were not addressed, please select, "I did not learn about this."

18. Overall, how satisfied have you been with the Pompe related information available to you as a grandparent?

Not at all satisfied Somewhat satisfied Satisfied Extremely satisfied

19. Please indicate (tell us) what information is essential for grandparents to have as they learn about Pompe.

Can you restate this question in your own words?

SECTION 6: GRANDPARENTS AND GENETICS

Currently, little is known about the needs of grandparents when a grandchild has a medical condition. We are interested to hear your thoughts about your experience with the genetics of Pompe.

20. How well do you feel you understand the genetics of Pompe?

Not at all Somewhat Mostly Completely

What does "somewhat" mean to you?

How did you choose your answer for this question?

21. Please describe your understanding of the genetics of Pompe, or how the condition is inherited.

Can you restate this question in your own words?

How would you ask someone to explain how a condition is passed through a family?

Given the known diagnosis of Pompe in your family, your or other relatives may have considered genetic testing to determine if you are a carrier for Pompe. Carriers themselves do not have the disease, and being a carrier does not affect individuals physically, mentally, or in any other way. The only consequence of being a carrier for Pompe is the possibility of transmitting the genetic trait to a child (passing the genetic trait to a child.)

What questions do you have about what you just read?

22. Have you had genetic testing to determine your carrier status for Pompe? If you have had carrier testing, please describe the impact that carrier testing has had on you.
- Unsure
- No
- Yes _____

SECTION 7: EMOTIONAL SUPPORT NEEDS

Please tell us about the emotional support you received as you learned about and adjusted to your grandchild's diagnosis.

23. Please indicate your level of satisfaction with each of the places you have found emotional support while adjusting to your grandchild's diagnosis. If you have not received support from a particular source, please select "Not applicable."
24. Overall, how satisfied have you been with the emotional support available to you as a grandparent of a child with Pompe?
- Not at all satisfied Somewhat satisfied Satisfied Extremely satisfied
25. What is your ideal version of emotional support for grandparents?
26. How are your emotional support needs as a grandparent of a child with Pompe different from the emotional support needs of grandparents whose grandchildren have Pompe?

SECTION 8: PSYCHOSOCIAL IMPACT

27. How has having a grandchild with Pompe impacted your personal life?
28. What else would you like us to know about being a grandparent of an individual (child) with Pompe?

Appendix D – Online Survey

The Informational and Emotional Support Needs of Grandparents of Children with Pompe disease

You are being asked to participate in this study because one of your grandchildren has been diagnosed with Pompe. The purpose of this study is to better understand grandparents' informational and emotional support needs when they have a grandchild with Pompe.

Your participation in this study will increase the awareness of grandparents' unique needs and help us provide the appropriate support to other grandparents in your situation. Your participation in the study is voluntary, and you can withdraw from the study at any time. Participating in the study involves completing an online survey. The survey is anonymous, meaning that we will not collect any personal information that could identify you or connect you to your responses. The survey should take 15-20 minutes to complete; the time to complete the survey will vary depending on the length of your responses. Questions in the survey will ask you about yourself and your grandchild, your role in your family, the sources and type of information you have received about Pompe, your experience with emotional support, and your satisfaction with the resources available to you as a grandparent. The survey also includes questions that ask about the impact your grandchild's diagnosis has had on you and your suggestions for providing information and support to grandparents.

This study is being conducted by Natasha Rudy, a genetic counseling student at the University of South Carolina Medical School for a Master's Thesis project. Janice Edwards, a genetic counselor at the University of South Carolina, is the thesis advisor for this study. If you have any questions about this study, you may contact us:

Natasha Rudy, BA, BS

Phone: 864-992-8469

Email: rudynl@email.wofford.edu

Janice G. Edwards, MS, CGC

Phone: 803-545-5706

Email: jedwards@usemed.sc.edu

For questions about your rights as a participant, you may contact the Office of Research Compliance at the University of South Carolina at 803-777-7095.

By accessing the online survey by clicking the "Next" button below, you are indicating your consent to participate in this study.

Thank you for sharing your experiences with us.

Thank you for your willingness to share your experience with us. Please tell us a little about yourself. For the following questions, please select the answer that *best* fits your situation.

1. What is your current age (in years)? _____
2. Are you male or female?
 - Male
 - Female
3. Please complete this sentence: My grandchild with Pompe is my
 - Daughter's child
 - Son's child
4. In what state do you live?
 - United States listing
 - Outside of the United States (please specify) _____
5. Please tell us your living arrangements regarding your grandchild with Pompe.
 - In the same house
 - Less than 30 miles away
 - 30-100 miles away
 - More than 100 miles away
6. What is your religious affiliation?
 - Buddhist
 - Catholic
 - Evangelical Christian
 - Hindu
 - Jewish
 - Mormon
 - Muslim
 - Protestant
 - No religious affiliation
 - Other (please specify) _____
7. What is your current relationship status:
 - Married
 - Widowed
 - Divorced
 - Separated
 - In a domestic partnership or civil union
 - Single, but cohabitating with significant other
 - Single, never married

8. What is your race/ethnicity? (Choose all that apply)

- Black or African-American
- White
- Asian
- American Indian or Alaskan Native
- Native Hawaiian or other Pacific Islander
- Other (please specify) _____

9. At what age was your grandchild diagnosed with Pompe? _____

10. What subtype of Pompe was your grandchild diagnosed with?

- Classic Infantile (diagnosed within the first year of life with heart involvement)
- Non-classic Infantile or Atypical Infantile (diagnosed within the first year of life without heart involvement)
- Juvenile or late-onset (diagnosed after the first year of life without heart involvement)
- Unsure

11. Is your grandchild living or deceased?

- Living
- Deceased

If answer to “Deceased,” skip to question 15.

Please tell us about your relationship with or grandchild.

12. Currently, how old is your grandchild with Pompe? _____

13. On a scale of 1-5 with 5 being the strongest, how strong is your relationship with your grandchild?

1 2 3 4 5

14. Are you the primary caregiver for your grandchild with Pompe?

- Yes
- No

Skip to question 18

We are so sorry for the loss you experienced. We would be honored if you would share some information about your relationship with your grandchild.

15. How old was your grandchild when they passed? _____

16. On a scale of 1-5 with 5 being the strongest, how strong was your relationship with your grandchild?

1 2 3 4 5

17. Were you the primary caregiver for your grandchild with Pompe?

- Yes
- No

Skip to question 21

Please provide some information about your roles within your family.

18. Please tell us how often you provide each type of support or assistance:

	Daily	Weekly	Monthly	A few times a year	I do not do this
Financial support					
Emotional support					
Respite care or short-term child care (babysitting or chaperoning)					
Primary child care or long-term child care					
Assistance with medical care/management for Pompe					
Running errands or completing housework					

19. How would you describe your unique role within your family after having a grandchild with Pompe? _____

20. How is grandparenting your grandchild with Pompe different from grandparenting your other grandchildren? _____

Skip to Question 24

Please provide some information about your roles in your family.

21. Please tell us how often you provided each support or assistance:

	Daily	Weekly	Monthly	A few times a year	I did not do this
Financial support					
Emotional support					
Respite care or short-term child care (babysitting or chaperoning)					

Primary child care or long-term child care					
Assistance with medical care/management for Pompe					
Running errands or completing housework					

22. How would you describe your unique role in your family? _____

23. How was grandparenting your grandchild with Pompe different from grandparenting your other grandchildren? _____

Please tell us about the information you received regarding Pompe and your grandchild’s diagnosis.

24. Please share your level of satisfaction with each *source of information* about Pompe. If you have not used a source for information, please select, “I have not gotten information from this source.”

	Extremely satisfied	Satisfied	Somewhat satisfied	Not satisfied	I have not gotten information from this source
My grandchild’s parent(s)					
Extended family members					
Friends					
Pediatrician, Family Care Doctor, or Primary Care Physician					
Metabolic Geneticist					
Genetic Counselor					
Internet Research					
Online Support Organization					
Support group attendance					

25. To the best of your recollection, please indicate your satisfaction with the information you have learned about Pompe. If any of the following items were not addressed, please select, “I have not learned about this.”

	Extremely satisfied	Satisfied	Somewhat satisfied	Not satisfied	I have not learned about this
Genetic information or information about the inheritance of Pompe					
Medical complications associated with Pompe					
Treatment options					
Therapies (physical therapy, occupational therapy, etc.)					
Potential impact of Pompe on the family					
Potential long-term outcomes for individuals with Pompe					
Web-based informational resources					
Printed information					
The opportunity to speak with other grandparents who have experienced a similar situation					
Referrals to individual/family counseling					

26. Overall, how satisfied have you been with the information you received about Pompe?

Very satisfied Satisfied Unsure Dissatisfied Very Dissatisfied

27. What has been the most important information you have learned about Pompe?

Please tell us about your experience learning about the genetics or inheritance of Pompe.

28. How much do you agree or disagree with this statement: “*I understand the genetic, or inheritance of Pompe.*”

Strongly Agree Agree Neutral Disagree Strongly Disagree

29. How would you explain the genetics of Pompe or how Pompe disease is inherited? _____

Given the known diagnosis of Pompe in your family, you may have considered genetic testing to determine if you are a carrier for Pompe. Carriers themselves do not have the disease, and being a carrier does not affect individuals physically, mentally, or in any other way. The only consequence of being a carrier for Pompe is that possibility of passing the genetic trait to a child.

30. Have you had genetic testing to determine your carrier status for Pompe?

- Yes
- No
- Unsure

If “No” or “Unsure,” skip to question 33

31. Were you found to be a carrier of a gene mutation for Pompe disease?

- Yes, I am a carrier
- No, I am not a carrier
- Unsure/ I do not wish to share

32. How has carrier testing impacted you or your relationship with your grandchild?

Please tell us about the emotional support you received as you learned about and adjusted to your grandchild’s diagnosis of Pompe.

33. Please tell us your level of satisfaction with each source of *emotional support* related to your grandchild’s diagnosis. If you have not received support from a particular source, please select “I have not found support here.”

	Extremely satisfied	Satisfied	Somewhat satisfied	Not satisfied	I have not found support here
My grandchild’s parent(s)					
Spouse or significant other					
Extended family					
Friends					
Pediatrician, Family Doctor, or Primary Care Physician					
Metabolic Geneticist					
Genetic Counselor					
Internet resources					
Printed material					
Online support organization					

In person support group attendance					
Religious or spiritual community					

34. Please describe the emotional support you have received. _____

35. Overall, how satisfied have you been with the emotional support available to you as a grandparent of a child with Pompe?

Very satisfied Satisfied Unsure Dissatisfied Very Dissatisfied

36. What type of emotional support has been the most important to you after learning about your grandchild's diagnosis? _____

37. Please describe any changes in your life that have come from having a grandchild with Pompe (travel plans, retirement plans, relationships, etc.). _____

38. What else would you like us to know about being a grandparent of an individual with Pompe? _____