KLIPPEL-FEIL SYNDROME: A STUDY OF PARENTS' EXPERIENCES OF DIAGNOSIS, HEALTH SERVICE USE, AND ONLINE SUPPORT IN A RARE DISEASE POPULATION

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DEDICATION

In honor of my children, Sarah, Caleb, and Hannah, and all parents of children diagnosed with rare diseases.

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ABSTRACT

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KLIPPEL-FEIL SYNDROME: A STUDY OF PARENTS' EXPERIENCES OF DIAGNOSIS, HEALTH SERVICE USE, AND ONLINE SUPPORT IN A RARE DISEASE POPULATION

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This study describes the experience of diagnosis, health service use, and online support for parents who have a child diagnosed with Klippel-Feil Syndrome. The survey drew questions from the EurordisCare 2 and 3 surveys. A total of 15 parents participated in the study; parents were recruited from a closed online Facebook support group for parents of KFS children. This study describes psychosocial issues relevant to KFS, such as challenges, adjustments, and resources. Uncertainty in KFS treatment and management are delineated, including discussion of ways parents experience uncertainty. Access to physicians with KFS experience and travel were found to be barriers in care for the parents in this study. The positive role of a closed online support group and how the group fosters resilience are discussed.

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CHAPTER I

INTRODUCTION

Healthcare in the United States is a major political and social issue. During the 2016 fiscal year, the National Institutes of Health (NIH, 2016) received approximately $32.3 billion for medical research (NIH, 2016). In a country that puts healthcare reform at the political forefront and dedicates billions of dollars toward improving the health of its citizens, it could be difficult to imagine that there is a disease for which few if any treatments are available. However, the lack of available treatments and lack of disease specific information are the reality for patients and families who find themselves or a family member diagnosed with a rare disease (Anderson, Elliot, & Zurynski, 2013; Kole & Faurisson, 2009; Schieppati, Henter, Daina, & Aperia, 2008; Shire, 2013).

Rare disease as a classification creates a context that influences every aspect of disease treatment and management. Patients diagnosed with a rare disease face many difficulties obtaining a correct diagnosis (Anderson et al., 2013; Kole & Faurisson, 2009; Schieppati et al., 2008; Shire, 2013), which delays the time it takes for patients and families to understand a loved one's disease and collectively adjust emotionally and psychologically. While the prolonged experience of getting a diagnosis can leave rare disease patients and families in a state of ambiguity, very often a confirmed diagnosis

may bring the realization that the medical community simply does not know enough about a disease to provide definitive answers for patients (Anderson et al., 2013; Kole & Faurisson, 2009; Shire, 2013). Although legislation such as the Orphan Drug Act of 1983 along with local and national support organizations have joined forces to advance the plight of rare diseases, many rare diseases remain orphans in research and drug development (Griggs et al., 2009). The average lay person may recognize some rare diseases that have garnered much public support through fundraising and increased public awareness such as cystic fibrosis, yet thousands of others remain under-researched and unknown to society at large. Living with a rare disease is arguably a social determinant of health disparity due to the many disease barriers faced by rare disease patients (Williams, 2011).

Klippel-Feil Syndrome

Klippel-Feil Syndrome (KFS) is an orphaned rare disease that was first clinically described in 1912 (Klippel & Feil, 1912). Current research on KFS has been limited to medical research on the disease only; as with many other orphaned rare diseases, none of the research on KFS has evaluated the impact of the disease on either patients or their families. Due to cervical problems, KFS patients have an increased risk of injury to the neck and are typically restricted in their physical activity; more severely impacted patients need to be regularly monitored by doctors (National Organization of Rare Disorders [NORD], 2016a). Researchers have identified that scoliosis is often associated with KFS (NORD, 2016a; Samartzis, Kalluri, Herman, Lubicky, & Shen, 2011;

Thomsen, Schneider, Weber, Johannisson, & Niethard, 1997; Xue et al., 2014). Scoliosis, a disfiguring disorder, affects the individual in two ways: psychosocial issues (Merenda, Costello, Santangelo, & Mulcahey, 2011; Payne et al., 1997; Reichel & Schanz, 2003) and decreased health related quality of life (Danielsson & Nachemson, 2001; Freidel et al., 2002). On the other hand, KFS can be genetically transmitted in families (NORD, 2016a), with family communication about genetic inheritance creating an additional difficulty for families (Gallo, Angst, Knafl, Hadley, & Smith, 2005; McAllister et al., 2007; Metcalfe, Plumridge, Coad, Shanks, & Gill, 2011). The genetic implications for a person diagnosed in late adulthood may be quite different when compared to younger counterparts who must consider the risk of passing on the disorder when procreating. Although KFS can be genetically inherited, it presents most often as a spontaneous occurrence in a family (NORD, 2016a).

In rare disease, the patient's experience of diagnosis in terms of the number of doctors consulted, time from initial symptoms until diagnosis, and the patient's access to disease information and experienced physicians have been identified as significant for understanding issues and barriers faced by patients (Kole & Faurisson, 2009; Schieppati et al., 2008; Shire, 2013). Further, the psychosocial demands of any disease depend on the interaction between the disease, the patient, and the family (Rolland, 1994). However, there is no research to elaborate the experience of diagnosis for the KFS population. Additionally, there is no research on KFS that explores the use of health professionals and services for this population, nor a focus on patients' psychosocial

wellbeing. The experience of diagnosis and health service use are critically important contextual issues for rare disease patients; understanding the parameters of these issues is important for appreciating the psychosocial impact of KFS.

Online Support Groups

The use of online support groups for patients with KFS has not been explored. However, research on the use of online support groups for other patients with a variety of other chronic illnesses is promising. Broadly, online support groups have been shown to support patients' need for access to information about their disease beyond information provided by health professionals, with a preference for information from individuals with lived experience (Mo & Coulson, 2014). The use of online support groups also provides patients with access to others' experiences and contact with other patients in their own social networks when, otherwise, patients do not have face-to-face experiences (Mo & Coulson, 2014). Online support groups also serve to legitimize emotional experiences that can be invalidated in an individual's social world (Varga & Paulus, 2014).

The use of online support groups in disease treatment are significant because perceived emotional support from an online support group has been shown to shape perceptions of healthcare services (Nambisan, Gustafson, Hawkins, & Pingree, 2014). Understanding the unique role of online support groups in treating a particular disease is important for establishing practices that improve health services and contribute to improvements in practices that enhance patient experiences in online communities (Nambisan et al., 2014). Although some research of online support groups in other rare

diseases have evaluated participant posts and explored factors related to participant posts (Coulson, Buchannan, Aubeeluck, 2007; Lasker, Sogolow, & Sharim, 2005), there are no studies that evaluate the use of online support in KFS.

Statement of the Problem

As a rare disease, Klippel-Feil Syndrome (KFS) has been understudied. Although KFS is not a fatal disease, it is chronic and incurable. Despite an extensive review of multiple databases available through the TWU library, there is a dearth of literature examining the experience of diagnosis or health service use by this population. Further, the author was unable to locate psychosocial or healthcare research for KFS. The significant psychosocial implications for scoliosis (Danielsson & Nachemson, 2001; Freidel et al., 2002; Payne et al., 1997; Merenda et al., 2011; Reichel & Schanz, 2003) may indicate that the burden for the KFS population is even higher when physical deformity proliferates. Understanding the experiences of the KFS population is an imperative prerequisite to future research addressing the specific needs of this population and the rare disease community as a whole.

Statement of Purpose

The purpose of this study is to describe the experience of diagnosis, health service use, and online support group use as reported by the parents of children with Klippel Feil Syndrome (KFS). This study seeks to describe the psychosocial impact of KFS on families related to diagnosis and health service use. Further, this study seeks to evaluate the use of online support groups as a resource in disease treatment and management.

This research seeks to address the enormous deficit of rare disease research and provide a foundation for future psychosocial and healthcare-related research for KFS patients and families. Among researched rare diseases, those classified as congenital malformations have historically received the fewest number of research publications (Heemstra, Van Weely, Buller, Leufkens, & De Vreuh, 2009). Additionally, this research carries implications for providers who work with individuals and families in the rare disease community.

Theoretical Framework

Rolland's (1994) Family Systems Illness Model is significant for elaborating a biopsychosocial illness perspective. Rolland asserted that disease onset, course, outcome, degree of incapacitation, and degree of uncertainty are important for understanding the psychosocial impact on the affected individual and family over time. According to Rolland's typology, KFS has an acute onset, since the disease is present at birth even if symptomology abates and individuals go undiagnosed. During the onset of an acute illness, families must adjust rapidly by learning to exchange roles flexibly, utilize resources, and solve problems (Rolland, 1994). For individuals diagnosed with KFS, the diagnosis can be accompanied by the need to see additional specialists for further testing (NORD, 2016a), thereby forcing a family to dedicate time, money, and travel for disease treatment while members alter roles to meet family needs. Due to the variability in KFS presentation (NORD, 2016a), the course of KFS can be progressive, constant, or relapsing depending on the individual (Rolland, 1994). As a disease, KFS is nonfatal but

the disease is differentially incapacitating depending on individual presentation and associated conditions (NORD, 2016a).

Most significantly, however, Rolland (1994) asserts that the degree of uncertainty or unpredictability is an overarching concept that permeates onset, course, outcome, and incapacitation. The degree of ambiguity in disease typology greatly impacts families and can erode resiliency (Rolland, 1994). The ambiguity that exists in the context of rare disease is significant for understanding the psychosocial impact of a rare diagnosis. For KFS, research focusing on long term health outcomes (Theiss, Smith, & Winter, 1997) is limited, and medical research is limited to case reports (Hsu, Manabat, Huffnagle, & Huffnagle, 2011; Matsumoto, Wakahara, & Shimizu, 2006; O'Donnel & Seupaul, 2008; Patel, Gupta, Bajaj, & Tagore, 2014; Serdiuk & Bosek, 2012) with few researchers focused on advancing KFS disease knowledge (Samartzis et al., 2011; Samartzis, Kalluri, Herman, Lubicky, & Shen, 2016; Thomsen et al., 1997) or genetics (Bayrakli1 et al., 2013; Tassabehji et al., 2008).

Regardless of the degree of ambiguity KFS families experience (Rolland, 1994), it is imperative to elaborate family processes that underscore a family's ability to face the challenge of living with a family member diagnosed with a rare disease. According to Walsh's (2003) concept of resiliency, it cannot be assumed that all families experiencing a KFS diagnosis succumb to dysfunctional patterns because of the ambiguous context or experience of rare disease. Walsh asserts that key processes in family resilience are related to family belief systems, organizational patterns, and family communication.

While some families may certainly be challenged to face the experience of a rare diagnosis, others may use the experience as an impetus for positive change that not only helps them meet current challenges but buffers them against future ones (Walsh, 2003).

Organizationally, KFS families face healthcare expenses for testing and care from specialty physicians that last a lifetime and tap into family financial resources (Klippel-Feil Syndrome Freedom, 2017), which are also greatly impacted by public policy and health insurance coverage. Travel to appointments may require extended stays and missed time from work and school that force members to alter roles, while mobilizing additional resources such as extended family or friends to fulfill responsibilities. Belief systems are altered when KFS families are forced to realign their hopes and dreams for a future that reflects realistic and fulfilling expectations of a child or adult with restricted physical activity and physical deformity. Furthermore, belief systems and family communication must adapt to allow for open discussion of genetic risk, future procreation, and issues of physical appearance.

KFS family members must be able to openly discuss their emotions and beliefs about the presence of KFS in their lives in a way that supports each family member with empathy and mutual respect, while propelling the family forward with a positive outlook that reflects share values and purpose. It is important to elucidate processes that build upon existing family strengths in order to mobilize resources that foster positive family adaptation (Walsh, 2003), especially with families that risk being overwhelmed by rare disease difficulties. A problem saturated focus from a mental health perspective, may

only reinforce a deficit perspective that prevents families and clinicians from recognizing strengths that fuel positive adaptation and by extension resilience (Walsh, 2016).

Research Questions

The three research questions below are based on the psychosocial perspective as described in Rolland's (1994) Family Systems Illness Model:

1. According to Rolland's (1994) Family Systems Illness Model, with respect to onset, course, and degree of uncertainty what is the experience of diagnosis for KFS patients and families?
2. According to Rolland's (1994) Family Systems Illness Model, with respect to disease treatment and management what barriers do KFS patients and families face in health service use?
3. Based on Walsh's (2003) concept of resiliency what role do online support groups play in KFS disease treatment and management?

The table below outlines the relationship between the research purposes, research questions and survey questions (see Table 1).

Table 1

Relationship Between Research Purpose, Research Questions, and Survey Questions

Purpose	Research Question	Quantitative Survey Questions	Qualitative Survey Questions
Describe the experience of diagnosis of KFS parents.	1. According to Rolland's (1994) Family Systems Illness Model, with respect to onset, course, and degree of uncertainty what is the experience of diagnosis for KFS patients and families?	1, 2, 3, 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17, 18, 19, 20, 21, 22, 23, 26	25, 40
Describe the experience of health service use of KFS parents.	2. According to Rolland's (1994) Family Systems Illness Model, with respect to disease treatment and management what barriers do KFS patients and families face in health service use?	4, 5, 28, 29, 30, 31, 32, 33, 34, 35, 36, 37	38, 39
Describe the purpose of online support group use of KFS parents.	3. Based on Walsh's (2003) concept of resiliency what role do online support groups play in KFS disease treatment and management?	24, 26	27

Role of the Researcher

The researcher is the mother to three children, two of the children are diagnosed with KFS, a 17-year-old daughter and 15-year-old son. The researcher's personal experience with KFS contributes to her knowledge, interpretation, and insight about the psychosocial impact of parenting a child living with a rare diagnosis. The researcher acknowledges that her personal experience can bias the interpretation of quantitative and qualitative data in the study and plans to limit her bias by including two additional coders to the research team. The researcher's faculty advisor and a doctoral student in the Family Therapy Program at Texas Woman's University will be the two additional coders.

Definition of Terms

Klippel-Feil Syndrome (KFS). Klippel Feil Syndrome is a rare disease characterized by fusions in the cervical spine that occur as a congenital malformation (NORD, 2016a). The cervical fusion is a congenital malformation that occurs during the third to eighth week of gestation (Thomsen et al., 1997).

Orphans. Rare diseases are called orphans because they have been neglected and marginalized in treatment and research (Schieppati et al., 2008).

Rare disease. A disease is considered rare in the United States if it affects fewer than 200,000 people, or if it affects more than 200,000 people but the development and marketing of a drug for the disease is cost prohibitive (Orphan Drug Act, 1983).

Scoliosis- a deviation in the spine causing the backbone to move laterally, a curve in the spine (Scoliosis, 2014).

Torticollis- tendency for the head to be held to one side (Torticollis, 2014).

Assumptions

1. Participants in the closed Facebook support group recruited for this study will be honest in their responses to the online questionnaire.
2. Participants from the closed Facebook support group will join this research project only if they have a child with KFS.
3. Participants on the closed Facebook support group are over age 18 and have access to a computer.
4. Participants from the closed online support group who volunteer for this study will be similar to those support group members who do not participate.

Delimitations

1. Participants are limited to residents of the United States only.
2. Participants are limited to only those members in the closed online parent support group.
3. Participants are limited to the parents of children diagnosed with KFS by a medical professional.

Summary

First identified by Klippel and Feil in 1912, Klippel Feil Syndrome (KFS) is a rare disease characterized by fusions in the cervical spine that occur as a congenital malformation (NORD, 2016a). KFS is most commonly associated with scoliosis (NORD, 2016a; Samartzis et al., 2011; Thomsen et al., 1997; Xue et al., 2014), although

KFS can present with a plethora of associated anomalies and medical conditions (NORD, 2016a) that cause it to be very different from scoliosis. KFS individuals are typically restricted from physical activities that could increase the risk of injury to the cervical spine (NORD, 2016a). To date, medical case studies have provided the most current research into KFS. The purpose of the present qualitative study is to gather biopsychosocial research about KFS that examines experiences of diagnosis and health service use among KFS patients and families. Additionally, this study seeks to understand the role of a closed online support group in disease treatment and management among KFS patients and families. Furthermore, this study will maintain a focus on the psychosocial impact (Rolland, 1994) of illness, while appreciating factors that contribute to family resilience (Walsh, 2003). Overall, this study will address deficits in rare disease research and carries implications for KFS patients and families as well as the rare disease community.

CHAPTER II

REVIEW OF LITERATURE

This review of literature on rare disease will set the context for the focus of this paper on Klippel Feil Syndrome (KFS), a rare disease that has generated research in the medical field. It was found that KFS research is currently limited to medical research with a majority of that research focusing on case reports. In fact, a review of research that focused on rare diseases yielded the finding that diseases classified as congenital malformations have historically received the fewest number of research publications among rare diseases (Heemstra et al., 2009).

This researcher examined common psychosocial issues that are experienced by patients with a rare disease diagnosis and then summarized psychosocial issues that have been identified as specific to scoliosis, which is the most common complication of KFS, (NORD, 2016a; Samartzis et al., 2011; Thomsen et al., 1997; Xue et al., 2014). However, the researcher did not find any published journal articles that examined psychosocial issues for KFS patients and families in a thorough exploration of databases available at TWU. There has been no research to explore how patients and their families experience KFS, a congential malformation, nor is there research on how the patient was diagnosed, the patient's access to care, or psychosocial issues reported by this population.

Context of Rare Disease

In the examination of any rare disease, understanding context is imperative. In the United States, a disease is considered rare if it affects fewer than 200,000 people, or if it affects more than 200,000 people but the development and marketing of a drug for the disease is cost prohibitive (Orphan Drug Act, 1983). Although low prevalence rates determine rare status, there are an estimated 30 million Americans, or 1 in 10, who have a rare disease (National Organization of Rare Disorders [NORD], 2016b). Of the 7,000 known rare diseases, less than 500 have some type of government approved treatment (NORD, 2016b). The reality for people who do have a rare disease is that the vast majority of rare diseases have no cure (Schieppati et al., 2008).

Rare diseases are called orphans because they have been neglected and marginalized in treatment and research (Schieppati et al., 2008). Currently, there is a lack of research on the social and health care implications of rare disease patients and families living in the United States (Williams, 2011). In a historical effort to entice pharmaceutical companies to develop drug treatments or designate existing drugs for new uses, the U.S government established the Orphan Drug Act of 1983 (Orphan Drug Act, 1983). The act provides incentives for drug development and allows increased flexibility for companies to perform clinical trials (Orphan Drug Act, 1983). Although the Orphan Drug Act of 1983 and government grant programs have created opportunities for research into rare disease, researchers face challenges when investigating rare diseases, such as difficulty recruiting research participants, securing funding, and getting support from

pharmaceutical companies that stand to profit more from drugs developed for common diseases (Griggs et al., 2009). Due to the smaller number of people in the population with a specific rare disease, Institutional Review Boards and the U.S. Health Insurance Portability and Accountability Act (HIPAA) may add to the complexity of protecting research participants or creating research registries (Griggs et al., 2009). For example, in a study reviewed for this research, a rare disease was classified as a very rare syndrome because naming the disease could violate the privacy of the 5 known patients in the country where the research took place (Huyard, 2009). Furthermore, a universal system that could be used internationally to code and register rare disease patients does not exist (Schieppati et al., 2008).

Rare disease research is replete with information about difficulties related to patient diagnosis. Rare disease patients and families face delays in diagnosis, typically receive a misdiagnosis before an accurate diagnosis is determined, and report seeing multiple doctors before getting a correct diagnosis (Anderson et al., 2013; Kole & Faurisson, 2009; Schieppati et al., 2008; Shire, 2013). For example, in the Eurordis survey of almost 12,000 rare disease patients in Europe, participants affected by Ehler-Danlos Syndrome ranged from consulting more than five to more than 20 physicians before receiving a correct diagnosis (Kole & Faurisson, 2009). Additionally, 7% of all patients in the Eurordis survey reported that psychological or psychiatric treatments were inappropriately administered after a psychiatric diagnosis was used to explain the patient's symptoms, which further delayed a correct diagnosis and contributed to a loss of

confidence in healthcare providers (Kole & Faurisson, 2009). The Eurordis survey, overall, found that rare disease patients face many barriers, including:

(1) lack of scientific knowledge of their disease

(2) lack of access to correct diagnosis

(3) delays in diagnosis

(4) lack of appropriate multidisciplinary healthcare

(5) lack of quality information and support at the time of diagnosis

(6) undue social consequences

(7) inequities and difficulties in access to treatment, rehabilitation, and care

(8) dissatisfaction with and loss of confidence in medical and social services, and

(9) rejection by healthcare professionals. (Kole & Faurisson, 2009, p. 20)

In a report by Shire (2013) that evaluated the impact of rare disease using online surveys in a sample of 466 rare disease patients in the United States and the United Kingdom, U.S. patients waited an average of 7.6 years to get an accurate diagnosis, while visiting an average of 8 doctors and receiving 2 to 3 misdiagnoses. Overall, Shire (2013) found three major needs from the study: (1) a need for collaboration among physicians and specialists along with more physicians who have experience treating rare diseases; (2) a need for patient access to resources that assist them in coping with the emotional burden related to disease treatment and management; and**,** (3) more research needed to improve disease knowledge and treatment options.

Clearly, rare disease creates a context that imposes many barriers to patients and families. In addition, rare diseases as a whole have been understudied in medical research. The deficits in research are so great that Williams (2011) affirms rare disease fits the definition of a health disparity outlined in the Health Disparities Act of 2000 and should be included in the definition in order to advance research that addresses disparities in healthcare access and quality for rare disease patients and families. With thousands and thousands of rare diseases, however, progress in the treatment and development of strategies aimed at reducing healthcare barriers has been slow. As a rare disease, Klippel-Feil Syndrome is no exception.

Klippel-Feil Syndrome

The National Organization of Rare Disorders (2016a) defines Klippel-Feil Syndrome (KFS) as a rare disorder primarily characterized by the fusion of two or more vertebrae in the cervical spine or neck. The cervical fusion is a congenital malformation that occurs during the third to eighth week of gestation (Thomsen et al., 1997). Maurice Klippel and Andre Feil were French neurologists who first described the clinical presentation of the disorder in 1912 (Klippel & Feil, 1912). In 1919, Feil later classified the disorder into three groups (as cited in Samartzis et al., 2011). KFS is classified as Type I, Type II, or Type III; types are distinguished by the location or combination of fusions in the cervical, thoracic, and lumbar spine (Thomsen et al., 1997). Although the true incidence of the disorder is unknown, it is estimated that KFS occurs in one of every 42,000-50,000 live births, with 65% of cases occurring in females (NORD, 2016a). The

symptomatic presentation of KFS is defined by a triad of features, which include a short neck, limited range of motion in the head and neck, and a low hairline on the back of the neck (NORD, 2016a). Although a majority of KFS patients will present with the classic triad, not all KFS patients have all three symptoms (Samartzis et al., 2016). Additionally, KFS is associated with complications in other bodily systems due to its congenital presentation (NORD, 2016a; Xue et al., 2014). Complications related to KFS include additional skeletal abnormalities, Sprengel's deformity, spina bifida occulta, hearing impairment, eye abnormalities, craniofacial abnormalities, torticollis, cleft palate, heart defects, kidney defects, and neurologic complications due to spinal injury or age related deterioration of the spine (NORD, 2016a). The most commonly associated complication of KFS is scoliosis (NORD, 2016a; Thomsen et al., 1997; Xue et al., 2014).

According to the National Organization of Rare Disorders (2016a), KFS presents with great variability between individuals depending on the severity of their condition and associated conditions and is related to other documented disorders, such as Wildervanck Syndrome, Duane Syndrome, and juvenile rheumatoid arthritis. Patients are typically diagnosed by a medical professional employing magnetic resonance imaging or x-rays, with additional testing dependent on associated problems. Due to the impact on multiple body systems, a multidisciplinary team of medical professionals may typically be needed to plan treatment. Since KFS patients may suffer spinal cord injury due to minor spinal trauma, it is recommended that KFS patients be monitored by a physician regularly and avoid activities that increase the risk of injury to the neck. Treatment may

involve the use of cervical collars, braces, traction, and medications to reduce inflammation and pain to more invasive surgical procedures that address cervical compression or instability (NORD, 2016a).

Although it has been more than 100 years since KFS was first described (Klippel & Feil, 1912), much of the current research on KFS is based on case report studies. One case report underscored the use of a multidisciplinary team as integral in treating a Type III KFS parturient, specifically, the team planned for anesthetic treatment of the patient during delivery (Hsu et al., 2011). Another case study cautioned medical professionals about the difficulty of intubating KFS patients, indicating that successful intubation at one time will not predict repeated success even with the same medical equipment after cervical surgery (Serdiuk & Bosek, 2012). A study of 11 pediatric KFS patients reported on a variety of ways to manage airways during general anesthesia, indicating not all intubations are difficult (Stallmer, Vanaharam, & Mashour, 2008). Central cord syndrome (National Institute of Neurologic Disorders and Stroke [NINDS], 2015) has been reported by O'Donnel and Seupaul (2008) in a case of a 19-year-old man following his fall down ten steps, and by Matsumoto et al. (2006) concerning three men after they suffered injuries while either snowboarding or snow skiing. O'Donnel and Seupaul (2008) emphasized the need to educate KFS patients about the increased risk of neurologic impairment caused by minor trauma. On a different note, Bae (2014) showed that the use of cervical deep muscle strengthening was a promising alternative to surgical intervention for neck pain in a patient with KFS.

As a disorder, KFS is acknowledged to occur most often sporadically or without a known cause, as well as in a dominant and recessive pattern of inheritance (NORD, 2016a). For example, Patel et al. (2014) reported a sporadic and fatal case in which a neonate was born with KFS and additional complications to a family with no history of the disorder. While some cases are reported in isolation of other affected family members, scientists have recently discovered genes that may further elucidate information about KFS. For example, the GDF6 gene has been implicated in dominant patterns of inheritance for KFS (Tassabehji et al., 2008), while the gene MEOX1 has been implicated in recessive inheritance of KFS (Bayrakli et al., 2013). However, KFS is thought to be multifactorial and the disorder may occur as a result of the combination of genetic and environmental factors which contribute to a heterogeneous disease presentation (NORD, 2016a). Essentially, no single gene is the sole cause of KFS and the disease etiology is not completely understood (NORD, 2016a).

Psychosocial Factors

Scoliosis is continually highlighted in research about KFS and there is a high correlation between the two diseases (NORD, 2016a; Samartzis et al., 2011; Thomsen et al., 1997; Xue et al., 2014). Thomsen et al. (1997) reported scoliosis in 70% of the 57 KFS patients evaluated for their study. While Xue et al. (2014) evaluated a population of patients with congenital scoliosis to determine the presence of KFS, reporting that 28 out of 516 patients had KFS. Theiss et al. (1997) evaluated the incidence of cervical symptoms in a follow up study of 32 patients with KFS and congenital scoliosis. Thiess

et al. found a low incidence of cervical related symptoms, with 7 (22%) of the patients reporting a history of symptoms; however, the participants were young when they first presented and the follow up period may not adequately reflect the evolution of cervical symptoms over the life span. No studies could be found that examined the psychosocial impact of scoliosis related to a KFS diagnosis. Additionally, this research did not seek to include studies on the extensive anomalous features associated with KFS; however, the psychosocial impact of scoliosis was examined including family communication about genetics, interactions with healthcare professionals, family impact, and sources of support.

Studies on scoliosis have tended to focus on measuring health related quality of life, although the Health Related Quality of Life measurement has tended to overlook some psychosocial issues experienced by scoliotic patients (Tones, Moss, & Polly, 2006). According to Tones et al. (2006), studies have examined the following issues: bracing in scoliosis treatment, surgical intervention, and psychosocial issues in scoliotic individuals. Payne et al. (1997) found that scoliosis was an independent risk factor for adolescents having suicidal thoughts, concerns about the abnormal development of their body, and concerns about their interactions with peers. Payne et. al.'s sample was collected from 685 students, ages 12-18, who completed the Adolescent Health Survey, a reliable and validated survey measuring health concerns. Reichel and Schanz (2003) reported that psychological implications for adolescents with scoliosis include physical restrictions, necessary adjustment to treatment regimens, uncertainty about disease progression that

negatively impact self-worth and self-esteem, and decreased contact with peers due to treatment measures. Reichel and Schanz discuss the psychological difficulty of adolescent scoliotics who feel they are not normal, attempt to hide their deformity, and experience feelings of shame and inferiority. Furthermore, Reichel and Schanz highlight the importance of recognizing that a scoliotic's perception of their deformity depends more on the patient's subjective experience of their curve rather than their actual impairment. Merenda et al. (2011) found that appearance, feelings, bracing, and discomfort were important themes for girls with idiopathic scoliosis, in a sample of 76 females between the ages of 8 and 16.

Two scoliosis studies (Danielsson & Nachemson, 2001; Freidel et al., 2002) reviewed for this current study examined health related quality of life measures and reported psychosocial impairment. In the first study, a 22 year follow-up study of 283 scoliotic females with a mean age of 40, the researchers showed that 17% of patients with children reported that fear of back problems for the patient or for the patient's child was the second major reason participants ceased having children (Danielsson & Nachemson, 2001). Furthermore, scoliosis participants were twice as likely as the control group to report that feeling physically limited negatively impacted sexual function (Danielsson & Nachemson, 2001). In the second study, Freidel et al. (2002) reported that female scoliotics in a sample of 226 participants under age 17, when compared to a norm sample had less positive attitudes about their lives, reported more bodily complaints, lower self-esteem, and had more feelings of sadness. Additionally, participants over age 21 reported

even lower health related quality of life than the juvenile group (Freidel et al., 2002). Although the psychosocial impact noted in these two studies highlight difficulties related to a scoliotic presentation, scoliosis may represent only one of several physical deformities experienced by KFS patients. For example, a hypothetical KFS patient as proposed by this researcher could have scoliosis, kyphosis, and a very short or almost absent neck, along with ear deformities and facial asymmetry. On the other end of the spectrum, this researcher proposes a KFS patient may look like an average person never knowing of the disorder until cervical symptoms or incidental radiologic findings suggest a problem. While some scoliotics may use clothing to hide a deformity, for the most severely afflicted KFS patient there is no hiding their physical deformity.

An additional complication for many rare disease patients and families is discovering that their rare disease also carries genetic implications. The National Organization of Rare Disorders reports that 80% of rare diseases are genetic (NORD, 2016b). Genetic information and transmission is not always discussed with patients at the time of diagnosis; genetic counseling is not always offered to rare disease patients, and health providers do not always make clear when genetic information should be shared with other family members (Kole & Faurisson, 2009). A study that focused on family communication about genetic inheritance found that 80% of 139 parents did not have any assistance from health professionals about how to communicate genetic inheritance to their children (Gallo et al., 2005).

Research on communication about genetic diseases highlight some of the psychosocial challenges faced by families. McAllister et al. (2007) used focus groups to discern the effects of genetic diseases from health professionals, patients, and patient representatives. Patient focus groups reported that parent-child communication about genetic inheritance was a challenge. Specifically, patients were concerned about how much information to share without overburdening a child, as well as, telling them at the right time (McAllister et al., 2007). A qualitative study of 33 families by Metcalfe et al. (2011) showed that parental discussion about genetic risk depended more on disease morbidity than a child's development. Children between the ages of 8-11 and up were unlikely to openly discuss a condition unless the parents had openly acknowledged it, while parents experienced great worry and concern about discussing genetic inheritance. Additionally, parents and children did not always comprehend patterns of inheritance and would instead rely on similarities in visual likeness and personality as indications of possible inheritance (Metcalfe et al., 2011). Not only is discussing genetic inheritance fraught with concerns about what to say when and how much information should be given, but KFS patients must understand the variability within the disorder's presentation carries a great risk for associated complications (NORD, 2016a).

Patient-provider relationships are significant to disease treatment and management; however, rare disease treatment has its own challenges. When rare diseases do have clinical practice guidelines for medical professionals to follow, efforts to obtain them for a specific disease can be time consuming and many of the existing clinical

practice guidelines for rare diseases fail to meet quality standards (Pavan et al., 2017). In Huyard's (2009) qualitative study of 29 patients with a rare disease and 15 parents of children diagnosed with rare disease, Huyard found rarity in and of itself was not a main concern for patients. Patients described the need for more psychosocial support from physicians during diagnosis and guidance in planning for treatment. Most poignantly, patients expected physicians to acknowledge their own inexperience treating a rare disease. Patients seemed to understand the physicians' limited knowledge and desired for physicians to respond to their worries, listen to their concerns, provide guidance, and explain information in a respectful manner (Huyard, 2009). Additionally, 87% of the participants in the Eurordis Survey reported a need for psychological care during the time of diagnosis (Kole & Faurisson, 2009). Additionally, patients reported that rare conditions often involved care by multiple specialists who have never seen a particular rare condition in their career and long distance travel for specialty care that is increasingly difficult to manage between multiple specialists in different institutions (McAllister et al., 2007).

It is not difficult to deduce that the difficulties faced by rare disease patients impact the family. A study of parents of children with chronic rare disease revealed significantly higher scores on the Hamilton Rating Scale for Anxiety for fears, insomnia, depressed mood, somatic sensory, respiratory symptoms, and autonomic symptoms compared to parents of children with common chronic conditions a year after diagnosis (Picci et al., 2015). Overall, these studies highlight the need to understand psychosocial

issues in rare disease patients and families. As with many other rare diseases, KFS patient needs and expectations in healthcare use have not been specifically evaluated, nor has the impact on the family.

Although much of the research on rare disease has identified major deficits, supportive strategies cannot be overlooked if future researchers aim to develop effective interventions and improve health service delivery. Garrino et al.'s (2015) qualitative research yielded the finding that support from their own family was frequently discussed by rare disease patients as playing a significant role in facilitating disease adjustment and acceptance. Additionally, Garrino et al.'s (2015) study described positive aspects of interactions that patients with rare disease reported having with health professionals who used empathy and humanity when providing care. Picci et al. (2015) compared chronic illnesses between a group of parents with children who have a chronic common illness and a group of parents with children who have a chronic rare disease. Picci et al. (2015) found that chronic rare disease parents were more likely to use religion as a coping strategy, whereas common chronic illness parents used more strategies to actively cope by planning and seeking instrumental social support. Although Picci et al. (2015) determined that religious coping was not useful in reducing the burden for parents with children diagnosed with a chronic rare disease, the study highlights the need to further examine and elaborate coping strategies in the rare disease community. It is highly likely that coping, in and of itself, may look different for a population that is by no means common.

In today's society, the use of technology cannot be overlooked as a way for rare disease patients and families to access information and connect with other rare disease patients. Garrino et al. (2015) report patients commonly used the internet to build their disease knowledge and satisfy their own need to understand their condition. There is no research on the use of an online support group for KFS patients and families, however, some research has evaluated the use of online groups for other rare diseases. Coulson, Buchannan, and Aubeeluck (2007), in a content analysis, found informational and emotional support specifically unique to Huntington's disease were important factors in an online support group. Lasker, Sogolow, and Sharim (2005) evaluated an online community of rare disease participants with primary biliary cirrhosis and found biomedical and socioemotional topics were significantly correlated in participants' posts, that people who were diagnosed for a longer period of time posted more often, age was not a factor in participant posts, and socioemotional topics did not follow topics typically expressed with other chronic illnesses. These studies highlight the importance of understanding the use of online support groups for rare disease patients. Furthermore, it would be important to ascertain from participants themselves how the use of online support is beneficial.

Summary

Rare disease imposes many healthcare barriers, including difficulty in securing an accurate and timely diagnosis (Kole & Faurisson, 2009; Shire, 2013). For KFS patients, there is no published data to elaborate healthcare barriers or experiences of diagnosis in

healthcare access and disease treatment. Although psychological care during diagnosis (Kole & Faurisson, 2009) and psychosocial support in disease treatment (Huyard, 2009) have been highlighted as areas in need of improvement by rare disease patients, no research on KFS has examined or elaborated these issues. Scoliosis is correlated with KFS (NORD, 2016a; Samartzis et al., 2011; Thomsen et al., 1997; Xue et al., 2014) and scoliosis itself carries psychosocial consequences (Danielsson & Nachemson, 2001; Freidel et al., 2002; Payne et al., 1997; Reichel & Schanz, 2003), yet no research elaborates psychosocial issues for the KFS population related to visual deformity of the spine or other affected areas of the body. Current KFS research represents medical case reports or small scale medical studies, which benefit medical professionals and do not elaborate issues specific to the KFS population. As a population KFS can include individuals who not only have scoliosis and face barriers related to rare disease, but KFS individuals may also be diagnosed with additional complications or anomalies (NORD, 2016a). KFS can be inherited genetically (NORD, 2016a) forcing families to discern how information about the condition will be discussed and understood. Yet, research shows communication about inheritance can be difficult for families (McAllister et al., 2007). In a technological era, however, families do not have to remain isolated in their illness experience. Online support groups for rare diseases not only differ in topic content compared to chronic illnesses (Lasker et al., 2005), but may serve a purpose significant to disease treatment and management that is not comprehensively understood. Overall, the context of rare disease and psychosocial issues related to scoliosis allude to a

need for research that defines issues for this population in a manner that can lead to improved disease treatment and management.

CHAPTER III

METHODOLOGY

The purpose of this qualitative study was to explore the experience of diagnosis, health service use, and online support group use by KFS parents using an online survey. The use of qualitative research methodology allowed this researcher to ask parents to share their experiences related to caring for a KFS child (Creswell & Plano Clark, 2007). Facebook support groups as a medium for research are suited for small qualitative studies (Valdez et al., 2014). The sample was recruited through an online support group for parents of children diagnosed with KFS; during the time of the study there were 216 parent members. The recruiting flyer was posted on the website and was, therefore, available to the entire population of members. A final sample of 15 parents responded to the online questionnaire. The open-ended questions supported by the qualitative research design (Creswell & Plano Clark, 2007) allowed the researcher to explore the parental experiences of obtaining a medical diagnosis for their child along with associated health service use; finally, the online questionnaire offered parents the opportunity to discuss their beliefs about the effectiveness of belonging to an online support group. Frequency data was gathered for the purpose of replicating selected questions from the EurordisCare study completed in Europe (Kole & Faurisson, 2009). The findings yielded from this

study provide further understanding of the experiences of diagnosis for families raising a child diagnosed with KFS, including use of health services and online support.

Sample Selection and Recruitment

The purposive sample for this research study included 16 parents who have a child diagnosed with KFS and reported that the child was diagnosed by a medical professional. As living in the U.S. was a criterion for this study, the one participant who lived outside of the United States was not included in the final sample of 15 parents of children diagnosed with KFS. Participation in the study was open to adults irregardless of race, ethnicity, sexual orientation or gender. Participants had to be at least 18 years old, have access to the Internet, and reside in the United States of America.

Participants were recruited from the *Klippel-Feil Syndrome Freedom* closed parent support group. The *Klippel-Feil Syndrome Freedom* support group was selected because the group administrator plays an active role in facilitating and providing information to members. Members must be granted permission by the group administrator to join the group due to their personal connection to KFS. *Klippel-Feil Syndrome Freedom* is a closed Facebook support group with three separate support groups; a teen patient group, an adult patient group, and a parent group. The administrator for the group was contacted through Facebook messenger (see Appendix B) and asked to grant permission for the research study to be posted in the closed parent support group. The group administrator granted permission to post the survey (see Appendix C). A description of the study (see Appendix C) and a link to the online

PsychData survey was posted in the closed parent support group by the group administrator only once. The researcher collaborated with the group administrator to post the recruitment flyer and survey link were open for 11 days, after which the administrator removed the post and the researcher turned off the link to PsychData.

Although the administrator's permission was not required because the survey researcher is a member of the closed group, the administrator is active in facilitating information and providing support. Additionally, the administrator retains privileges, actively monitors the group, and enforces guidelines for appropriate and respectful group interaction. The researcher sought to collaborate with the administrator because her active role in facilitating information within the group was considered beneficial to the current study. Valdez et al. (2014) report that building rapport with gatekeepers is an important prerequisite for recruiting participants.

Research Questions

The research questions are:

1. According to Rolland's (1994) Family Systems Illness Model, with respect to onset, course, and degree of uncertainty what is the experience of diagnosis for KFS patients and families?
2. According to Rolland's (1994) Family Systems Illness Model, with respect to disease treatment and management what barriers do KFS patients and families face in health service use?

3. Based on Walsh's (2003) concept of resiliency what role do online support groups play in KFS disease treatment and management?

Survey Design

The researcher designed a qualitative study that included a number of quantitative questions that were not designed for statistical analysis, but for summary of frequency data. The purpose of gathering the quantitative data in this manner was to enable the researcher to compare the study findings with those obtained by the EurordisCare surveys that served as a guide for developing the questionnaire for this study (Kole & Faurisson, 2009).

The use of an online survey in this research is based on guidelines developed by Dillman, Smyth, and Christian (2014). One guideline states that online surveys should be designed to reflect the capabilities of the survey population, the target population is accustomed to the use of online communication regarding their child's disorder as members of an online support group. Dillman et al. (2014) suggested responses not be required unless the response is significant to the study, most questions in this study do not require responses with the exception of questions used to screen participants. Another guideline encourages researchers to allow participants to back up in their survey and review information; participants in this study were allowed to review their responses and make corrections before submitting the survey which contributes to improved data quality. Finally, Dillman et al. (2014) guidelines proposed the flexibility of online data collection. In this study PsychData had mobile-optimized device displays so participants

that access their Facebook account and link to the survey from a mobile device had a similar experience to participants accessing the survey from a computer (Dillman et al., 2014). Using Facebook to collect study samples for small qualitative studies has been shown to be effective (Valdez et al., 2014).

Development of the Study Questionnaire

EurordisCare Surveys

As this researcher selected for her questionnaire a number of quantitative questions from the EurordisCare 2 Survey and the EurordisCare 3 Survey (see Appendices F and G), permission to use the questionnaires was requested from the Eurordis Organization. The EurordisCare Surveys were developed in Europe by the European Organization for Rare Disorders or Eurordis (2016), an organization that represents 724 rare disease patient organizations. The EurordisCare 2 and 3 Surveys were designed with input from patient organizations to more accurately represent patients' voices following the EurordisCare 1 survey, which sampled patient organizations only. The purpose of the EurordisCare 2 and 3 Surveys were to provide quantifiable information about difficulties faced by rare disease patients and to assist organizations in their advocacy efforts. Although the survey presents quantitative data, its purpose was to provide a descriptive valid synthesis of the experiences of rare disease patients in disease treatment and management. Because the EurordisCare 2 and 3 surveys were developed with input from many stakeholders in the rare disease community, it was used in the current research study as a guide for elaborating the issues and experiences of

KFS parents related to disease treatment and management of their children (Kole & Faurisson, 2009).

The EurordisCare 2 Survey was designed to elaborate factors associated with delays in diagnosis, impact on treatment and disease progression, and identify consequences of delayed diagnosis to increase public awareness of difficulties faced by rare disease patients (Kole & Faurisson, 2009). The Eurordis 3 survey sought to represent patient voices regarding the use of specialized care centers and patients' experiences and expectations of health service access and utilization (Kole & Faurisson, 2009). The EurordisCare 2 and 3 Surveys were created specifically for rare disease patients, and were developed out of collaboration between Eurordis staff and patient organization representatives whose input substantively expanded the survey questions (Kole & Faurisson, 2009). Information from the EurordisCare 2 and 3 surveys are a valid representation of patient opinions primarily explored and quantified for stakeholder debate; reliability was not discussed nor were studies establishing reliability or validity available (Kole & Faurisson, 2009).

This study sought to use part of the EurordisCare 2 and 3 Surveys to evaluate experiences of diagnosis and health service use in KFS patients; for example, the number of doctors seen before getting a KFS diagnosis and types of diagnoses preceding KFS. The EurordisCare 2 and 3 Surveys were formatted to be completed manually and then mailed to Eurordis (Kole & Faurisson, 2009); questions for this research were modified to reduce survey and question lengths, fit the PsychData format, to reflect the use of U.S.

healthcare, and eliminate questions pertaining only to European healthcare and demographic data that was not useful to this research study. The survey for this research study was altered to reflect cultural changes in language, use of the term Klippel Feil Syndrome, including changes pertinent to KFS disease specific information. The EurordisCare 2 and 3 surveys were translated from English to 12 and 15 languages respectively in a process where one volunteer would translate, then a second volunteer would valid the translation (Kole & Faurisson, 2009). The survey developed for this research study was reviewed by the researcher's advisor.

From the EurordisCare 2 Survey, participants were asked to quantify the number of doctors seen before a final diagnosis, types of examinations performed, name misdiagnoses, and identify consequences of delayed diagnosis. Additionally, participants identified who raised the possibility of rare disease, who diagnosed KFS, if a second opinion was sought, and which examinations were used to diagnose. Other questions included information about the type of facility used, including location. Finally, participants were asked to answer a question about moving due to diagnosis, identify the most helpful resources during diagnosis, and if they have had face-to-face contact with another KFS individual. Regarding questions from EurordisCare 3, participants were asked to rate medical professionals in terms of cost, travel, accessibility, and wait times for consultations.

Quantitative questions. Parents in this study were asked to identify their child's gender, current age and the child's age at KFS diagnosis, along with information about

other diagnoses and symptoms preceding KFS including the number and types of physicians used, facilities visited and their proximity to the family, and treatments following these diagnoses. Additionally, parents were asked about health insurance coverage at the time of diagnosis and currently, and whether they have met another KFS person face-to-face. Regarding diagnosis, parents were asked about delays in diagnosis, seeking a second opinion, and changing living arrangements due to KFS. Furthermore, parents were asked to evaluate physicians in terms of personal cost, travel, appointment wait time, and accessibility. Finally, parents were asked about beneficial resources in living with a KFS child and asked to rank order issues related to disease treatment and management.

Qualitative questions. This researcher used qualitative questions regarding participant experiences of diagnosis and health service use, and use of online support groups to fully describe parameters of disease experience that carry psychosocial impact in an attempt to identify issues quantitative questions may overlook. There are a total of five qualitative questions, listed below. Qualitative questions allowed participants to add additional comments about their experience of diagnosis, discuss their experiences of accessing and utilizing health services, explain their reasons for joining and using an online support group for KFS parents, and provide advice to other KFS parents.

The qualitative questions are:

1. What else would you like to say about your family's experience of diagnosis?
2. Please, describe your reasons for using an online KFS support group.

3. What else would you like to say about utilizing or interacting with physicians or health services?
4. What else would you like to say about your ability to access physicians or health services or get the services you need?
5. Please consider all of your experience with KFS, including information that has not been asked about. If you could give another parent of a child with KFS advice about any aspect of managing the disorder (physicians, insurance or even their own decision-making) please share any advice not previously discussed in this survey in the space below.

Procedure

Sandra Courbier, Surveys and Social Research Manager for Eurordis was contacted via email by this researcher and asked to grant copyright permission for use of questions in the EurordisCare 2 and 3 surveys (Appendix A); copyright permission was granted (Appendix B). The study was approved by the Texas Woman's University (TWU) Institutional Review Board (IRB) (Appendix K). A recruitment flyer for the study (Appendix C) with an online link was posted by the Facebook Administrator of *Klippel-Feil Syndrome Freedom* in the closed parent support group. Participants voluntarily accessed the study link online. This researcher followed TWU IRB protocol for including the consent to participate information in the PsychData survey (Appendix F). The criteria on page one of the survey described the study requirements for participation; participants were instructed not to continue if they did not meet the criteria

for the study. Participants were informed that they could withdraw from the study at any time. The survey was posted for 11 days and had a total of 16 participants. The group administrator removed the link to the survey, and the researcher turned off the link to the PsychData survey.

Data Analysis

The summary of the quantitative data consisted of frequencies (summed numbers, percentages, ranges, and one median) drawn from the demographic questions and computed via a PsychData report. Additional calculations were computed manually by this researcher.

Qualitative data were coded independently by the main researcher and academic advisor (Creswell & Plano Clark, 2007). The main researcher downloaded the responses to the qualitative questions into an Excel spreadsheet. The main researcher read through all qualitative responses and searched for key words in each response. Key words were identified and listed for each participant's response; key words represented the participant's actual language and reflected the overall nature of the response. The academic advisor also coded the qualitative data for key words. The two coders compared their key words for agreement and a simple 90% interrater reliability was reached.

For the second round of coding, the main researcher and co-coder reread all responses and compared key word responses for similarities and differences in order to develop broader categorical codes. When both researchers completed the second round

of categorical coding, the two researchers met to compare key words and categories until a simple 90% agreement was achieved. Categorical codes were based on key words with respect to the overall connotation of each response.

The third round of coding involved both coders and the third coder at the end of that process for the purpose of increasing trustworthiness through triangulated coding (Patton, 1999). First, the two coders discussed the categorical codes and the themes that each had generated separately. The two coders reread all responses, key words, and categories to develop and compare themes. When both researchers completed the third round of coding, a simple 90% agreement was achieved. The third coder, who is a doctoral student from Texas Woman's University's Family Therapy Program, was asked to review the coding. The student has previous experience coding her own research; the student was asked to ensure that the identified themes accurately reflected the data. Because the first two coders had achieved a simple agreement of 90%, the third coder was used primarily to provide triangulation (Patton, 1999) to offset the main researcher's bias as a KFS parent.

Additionally, the overall process of clearly and specifically delineating keywords and ensure the connection between each participant response, the keywords, and subsequent categorical codes, and themes was an added protocol to ensure the trustworthiness (Creswell & Plano Clark, 2007) of the data. In essence, every code developed had to clearly tie back to the original response with all data represented.

Study Impact

The study contributes a parent/caregiver perspective to the psychosocial impact of KFS in families through the lens of both resilience (Walsh, 2003) and biopsychosocial theory (Rolland, 1994). In addition, the study elaborates the role of online support in KFS disease treatment and management from a parent/caregiver perspective, providing implications for online support in the rare disease community. The study identified disease barriers and beneficial resources of KFS patients and families, as well as carry implications for rare disease barriers as a whole. Additionally, the study elaborated factors that contribute to KFS disease treatment and management using Walsh's (2003) concept of resiliency. Finally, the study adds context to aspects of disease treatment and management of KFS families by allowing patients to qualitatively discuss their lived experience.

Summary

This study used qualitative research methodology (Creswell & Plano Clark, 2007) to gather information about the impact of KFS diagnosis, access and utilization of physicians and health services, and use of beneficial resources for KFS parents. This survey used questions from the EurordisCare 2 and EurordisCare 3 surveys, along with additional quantitative and qualitative research questions designed by this researcher. The fifteen participants in this study were parents of children diagnosed with KFS. Participants answered questions via an online survey posted in a closed Facebook support group for parents, *Klippel Feil Syndrome Freedom*. Frequencies were computed on

quantitative data and the qualitative data was coded by two coders with a third coder involved to establish trustworthiness.

CHAPTER IV

RESULTS

The descriptive survey and qualitative findings discussed below were gathered from an online study of 15 parents raising children diagnosed with Klippel-Feil Syndrome who were participating in a closed online support group for parents. Descriptive survey findings are reported which provide quantitative data regarding parents' experiences of KFS diagnosis. Qualitative data are reported by providing information about the categories and themes evidenced in participant responses.

Descriptive Survey Findings

The online survey was posted on the Klippel-Feil Syndrome Freedom closed parent support group for 11 days. There was a total of 16 parents who reported that they were the parent of a biological or adopted child with KFS; one participant was excluded because the participant reported living in a country other than the United States of America. Of the children discussed in the survey, 7 were girls and 8 were boys. The children ranged in age from 2 to 20 years of age, and the median age was 12; one child's age was excluded because the parent reported his/her age instead of the age of the child. Of the parents, 35.7% (n = 5) reported that their child was under 10 years of age and 64.3% (n = 9) reported that their child was 11-20 years of age. Parents reported that at the

time of diagnosis and during the time the research survey was completed, all children had health insurance that covered KFS.

Number of Doctors Consulted before Final KFS Diagnosis

A survey question asked parents to share the number of doctors they consulted before their child's final diagnosis of KFS. The mostly frequently reported number of doctors that were consulted from the time the first symptom began until final diagnosis (see Table 2) was 3 to 5, reported by 46.7% (n = 7) of parents; 20% (n = 3) of parents reported consulting 1 to 2 doctors; 13.3% (n = 2) of parents reported consulting 6 to 10 doctors; 13.3% (n = 2) of parents reported seeing between 11 to 20 doctors; 6.7% (n = 1) reported that the diagnosis was an incidental finding following a football injury.

Table 2

Number of Doctors Consulted Before Final KFS Diagnosis

Number of Doctors Consulted	Percentage of Parents	Number of Parents
1 to 2	20.0%	3
3 to 5	46.7%	7
6 to 10	13.3%	2
11 to 20	13.3%	2
More than 20	0.0%	0
Other Response	6.7%	1
Total	100.0%	15

Initial KFS Symptom

A survey question asked parents to report up to three symptoms that initially manifested as a result of their child's KFS. Table 3 shows initial symptoms reported by more than one parent. A complete list of reported symptoms can be found in Appendix K. Of the symptoms reported by more than one parent for their child's KFS, 8.3% (n = 3)

of parents reported torticollis; 8.3% (n = 3) of parents reported breathing problems or lung problem; 8.3% (n = 3) of parents reported craniofacial abnormalities; 5.6% (n = 2) of parents reported back pain; 5.6% (n = 2) of parents reported limited cervical movement; 5.6% (n = 2) of parents reported short neck; 5.6% (n = 2) of parents reported neck pain; 5.6% (n = 2) of parents reported a kidney problem; 5.6% (n = 2) of parents reported cervical fusion; and 5.6% (n = 2) of parents reported a shoulder problem. The remaining symptoms were each reported by 2.8% (n = 1) or one parent and include a hand problem, numbness, swollen neck, hearing loss, unspecified neck problem, dizziness, rib cage deformity, swallowing problem, knee pain, chest pain, low muscle tone, scoliosis, and delayed milestones.

Table 3

Initial KFS Symptom

Symptoms[a]	Percentage of Parents	Number of Responses
Torticollis	8.3%	3
Breathing Problem/ Lungs (example: reactive airway disease, difficulty breathing)	8.3%	3
Craniofacial Abnormality (example: odd shape head, cleft palate, facial asymmetry)	8.3%	3
Back Pain	5.6%	2
Limited Cervical Movement	5.6%	2
Short Neck	5.6%	2
Neck Pain	5.6%	2
Kidney Problem (example: one kidney, multicystic kidney)	5.6%	2
Cervical Fusion	5.6%	2
Shoulder Problem (example: pain, lump on shoulder)	5.6%	2
Total	64.1%	23

Note. Parents reported up to three symptoms.
[a]Only Symptoms with multiple parent responses shown, full table in Appendix K.

Table 4 provides a summary of initial KFS symptoms related to the neck that parents reported for their children. Symptoms related to the neck were 36.3% of overall responses; 8.3% (n = 3) of parents reported torticollis; 5.6% (n = 2) of parents reported limited cervical movement; 5.6% (n = 2) of parents reported short neck; 5.6% (n = 2) of parents reported neck pain; 5.6% (n = 2) of parents reported cervical fusion; 2.8% (n = 1) reported swollen neck; and 2.8% (n = 1) reported an unspecified neck problem.

Table 4

Initial KFS Symptoms Related to the Neck

Symptom	Percentage of Parents	Number of Responses
Torticollis	8.3%	3
Limited Cervical Movement	5.6%	2
Short Neck	5.6%	2
Neck Pain	5.6%	2
Cervical Fusion	5.6%	2
Swollen Neck	2.8%	1
Unspecified Neck Problem	2.8%	1
Total	36.3%	13

Note. Parents reported up to three symptoms.

Treatments Performed based on Symptoms: Time from First Symptom Until the Final Diagnosis

Parents were asked to consider the time symptoms first began until final diagnosis of their child's KFS and report the type of examinations that were carried out during this period. From the time symptoms first began until final diagnosis of KFS for children (see Table 5), 93.3% (n=14) of parents reported radiological or imaging examinations such as x-ray or ultrasound were performed; 40% (n=6) of parents reported genetic testing was performed; 33.3% (n=5) of parents reported functional explorations (respiratory,

muscular, electroencephalogram); 20% (n = 3) of parents reported biological tests (blood test, urine test, biopsy) were performed; and 6.7% (n = 1) reported a sleep study was performed.

Table 5

Treatments Performed based on Symptoms: Time from First Symptom Until the Final Diagnosis

Type of Test Performed	Percentage of Parents	Number of Parents	Percentage of Total Responses
Radiological or Imaging Examinations	93.3%	14	48.3%
Genetic Testing	40.0%	6	20.7%
Functional Explorations	33.3%	5	17.2%
Biological Tests	20.0%	3	10.3%
Sleep Study	6.7%	1	3.5%
Total	-	29	100.0%

Note. Parents selected multiple answers.

Results of Delay in Diagnosis

Parents were asked to report the results of any delays in diagnosis for their children. Regarding diagnosis (see Table 6), 60.0% (n = 9) of parents reported there was no delay in diagnosis. Of the parents that did report issues with diagnosis, 13.3% (n = 2) of parents reported physical consequences such as pain; 13.3% (n = 2) of parents reported a loss of confidence in healthcare; 13.3% (n = 2) of parents did not specify the result of the delay in diagnosis; 6.7% (n = 1) reported physical therapy caused unnecessary pain; and 6.7% (n = 1) reported that diagnosis was only confirmed after the child suffered a concussion.

Table 6

Results of Delay in Diagnosis

Result	Percentage of Parents	Number of Parents	Percentage of Total Responses
No delay in diagnosis	60.0%	9	52.9%
Physical consequences (example: pain)	13.3%	2	11.8%
Loss of confidence in healthcare	13.3%	2	11.8%
Unspecified	13.3%	2	11.8%
Physical therapy caused unnecessary pain	6.7%	1	5.9%
Concussion	6.7%	1	5.9%
Total	-	17	100.1%

Note. Parents selected multiple responses.

Diagnoses, Patient Age, and Diagnosing Physician

Parents were asked to report up to three diagnoses given before the diagnosis of their child's KFS, along with the child's age at the time of the diagnosis and the type of physicians that made the diagnosis (see Table 7-9). The initial diagnosis, patient's age at diagnosis, and diagnosing physician are reported in Table 7. A total of 11 parents reported an initial diagnosis; 3 parents reported their child was diagnosed with torticollis by a pediatrician when the child was less than 1 year of age; 1 parent reported their child was diagnosed with torticollis by neurologist when the child was less than 1 year of age; 1 parent reported their child was diagnosed with a vertebral fusion by a geneticist when the child was less than one year of age; 1 parent reported the child was diagnosed with renal reflux when the child was less than 1 year of age by a radiologist; 1 parent reported their child was diagnosed with KFS by an unspecified specialist when the child was 1 year of age; 1 parent reported their child was diagnosed with scoliosis by a pediatrician

when the child was 1 year of age; 1 parent reported their child was diagnosed with spina bifida by a radiologist when the child was 1 year of age; 1 parent reported their child was diagnosed as having a single kidney by a nephrologist when the child was 1 year of age; and 1 parent reported that an urgent care physician was unable to determine a diagnosis when the child was 11 years of age. One parent reported she was diagnosed with a lack of fluid in the placenta during her third trimester by her gynecologist, but the response was excluded because the child/patient was not diagnosed.

Table 7

Initial Diagnosis, Patient Age, Diagnosing Physician

Diagnosis 1	Patient's Age by Year at Diagnosis	Diagnosing Physician
Torticollis	<1	Pediatrician
Torticollis	<1	Neurologist
Torticollis	<1	Pediatrician
Torticollis	<1	Pediatrician
Vertebral Fusion	<1	Geneticist
Renal Reflux	<1	Radiologist
KFS	1	Unspecified Specialist
Scoliosis	1	Pediatrician
Spina bifida	1	Radiologist
One kidney	1	Nephrologist
Unable to Determine	11	Urgent Care Physician

Note. Only 11 parents reported an initial diagnosis

A total of 6 parents reported a second diagnosis for their child, the child's age at diagnosis, and diagnosing physician (see Table 8); 2 parents reported their child was diagnosed with KFS when the child was less than 1 year of age, with one child diagnosed by an orthopedist and the other by an orthopedic surgeon; 1 parent reported their child

was diagnosed with mild hearing loss when the child was less than 1 year of age by an otolaryngologist; 2 parents reported their child was diagnosed with scoliosis when the child was 1 year of age, one child by an orthopedist and the other by an unspecified specialist; and 1 parent reported their child was diagnosed with idiopathic short stature by an endocrinologist when the child was 4 years of age;

Table 8

Second Diagnosis, Patient Age, Diagnosing Physician

Diagnosis 2	Patient's Age by Year at Diagnosis	Diagnosing Physician
KFS	<1	Orthopedist
KFS	<1	Orthopedic Surgeon
Mild Hearing Loss	<1	Otolaryngologist
Scoliosis	1	Orthopedist
Scoliosis	1	Unspecified Specialist
Idiopathic Short Stature	4	Endocrinologist

Note. Only 6 parents reported a second diagnosis.

A total of 2 parents reported a third diagnosis, patient's age at diagnosis, and diagnosing physician (see Table 9); 1 parent reported the child was diagnosed with amblyopia or lazy eye by an ophthalmologist when the child was 3 years of age, and 1 parent reported their child was diagnosed with Sprengel's deformity by an unspecified specialist when the child was 2 years of age.

Table 9

Third Diagnosis, Patient Age, Diagnosing Physician

Diagnosis 3	Patient's Age by Year at Diagnosis	Diagnosing Physician
Amblyopia (lazy eye)	3	Ophthalmologist
Sprengel's Deformity	2	Unspecified Specialist

Note. Only 2 parents reported a third diagnosis.

Treatments Following Diagnoses Before the KFS Diagnosis

Parents were asked to report any treatments that followed diagnoses that were given prior to the KFS diagnosis for their child. Following the diagnoses given before that of KFS (see Table 10), 40% (n = 6) of parents reported that no treatments were performed; 26.7% (n=4) of parents reported medicinal treatments; 26.7% (n = 4) of parents reported surgical treatment; 13.3% (n=2) of parents reported physical therapy; 6.7% (n = 1) reported psychological treatment; 6.7% (n = 1) reported having weekly ultrasounds during her pregnancy with the child possibly to monitor kidney development; and 6.7% (n = 1) reported the child received growth hormones from age 5 to 16.

Table 10

Treatments Following Diagnoses Before the KFS Diagnosis

Type of Treatment	Percentage of Parents	Number of Parents	Percentage of Total Responses
No Treatment	40.0%	6	31.6%
Medicinal Treatment	26.7%	4	21.0%
Surgical Treatment	26.7%	4	21.0%
Physical Therapy	13.3%	2	10.5%
Psychological Treatment	6.7%	1	5.3%
Ultrasound during Pregnancy of KFS Child	6.7%	1	5.3%
Growth Hormones	6.7%	1	5.3%
Total	-	19	100.0%

Note. Parents selected multiple answers.

Age of the Child when KFS was Considered and Diagnosed

Parents were asked to report the age of the child when KFS was first considered as a possible diagnosis. Only 14 parents reported the age of the child when the possibility of KFS was first raised (see Table 11), 42.9% (n = 6) of parents reported the

child was less than 1 year of age; 35.7% (n = 5) of parents reported the child was age 1 to 2; 21.4% (n = 3) of parents reported the child was an adolescent age 11-16. In a separate question parents were asked to report the age of the child when the KFS diagnosis was made. The same 14 parents reported the age of the child when the diagnosis of KFS was made, 35.7% (n = 5) of parents reported the diagnosis was made when the child was less than 1 year of age; 35.7% (n = 5) of parents reported the child was age 1 to 2; 7.1% (n = 1) of parent reported the child was age 4; and 21.4% (n = 3) of parents reported the child was an adolescent age 12-16. In a separate question, concerning who raised the possibility of KFS, 80% (n = 12) of parents reported a physician, 13.3% (n = 2) of parents reported that it was the parent that raised the possibility of KFS, and 6.7% (n = 1) the possibility was raised by a healthcare professional other than a physician.

Table 11

Age of the Child when KFS was Considered and Diagnosed

Age Range	Percentage of Parents Reporting Child Age when KFS was Considered	Number of Parents	Percentage of Parents Reporting Child Age when KFS was Diagnosed	Number of Parents
Under 1 year of age	42.9%	6	35.7%	5
1 to 2 years of age	35.7%	5	35.7%	5
3 to 5 years of age	0.0%	0	7.1%	1
6 to 10 years of age	0.0%	0	0.00%	0
11 to 17 years of age	21.4%	3	21.4%	3
Total	100.0%	14	99.9%	14

Note. Only 14 parents reported child age.

Type of Physician and Data Used to Diagnose KFS

Parents were asked to specify the type of doctor that made the KFS diagnosis for their child (see Table 12), 60% (n = 9) of parents reported the diagnosis was made by an orthopedist or pediatric orthopedist; 20% (n = 3) of parents reported that the diagnosis was made by a geneticist, 13.3% (n = 2) of parents reported it was an orthopedist or pediatric orthopedist and neurosurgeon; 6.7% (n = 1) reported an unspecified physician. In a separate question, parents were asked to specify the type of data used to support the KFS diagnosis. Data supporting KFS diagnosis was reported by 73.3% (n = 11) of parents to be based on radiological or imaging tests (MRI, x-ray, or other imaging); 20% (n = 3) of parents reported it was based on combined clinical and radiological/imaging; and 6.7% (n = 1) reported it was based on radiological and blood testing.

Table 12

Type of Physician Diagnosing KFS

Type of Physician	Percentage of Parents	Number of Parents
Orthopedist or Pediatric Orthopedist	60.0%	9
Geneticist	20.0%	3
Orthopedist or Pediatric Orthopedist and Neurosurgeon	13.3%	2
Unspecified Physician	6.7%	1
Total	100.0%	15

Facility Type and Facility Proximity

Parents were asked to report the type of facility where the KFS diagnosis was made. Regarding the facility where the KFS diagnosis was given (see Table 13) 40% (n = 6) of parents reported the diagnosis was given in a specialized care facility; 33.3% (n = 5) of parents reported the diagnosis was given in a hospital; 20% (n = 3) of parents

reported the diagnosis was made in a private practice; and 6.7% (*n* = 1) reported the diagnosis was made in a clinic. Parents were also asked to report how they found the facility. Regarding how the facility was found, 73.3% (*n* = 11) of parents reported the facility was recommended by a physician; 6.7% (*n* = 1) reported the facility was found through the internet; 6.7% (*n* = 1) reported the facility was associated with the child's school football program; 6.7 % (*n* = 1) reported the facility was found due to an emergency room visit; and 6.7% (*n* = 1) reported the facility was recommended by family.

Table 13

Facility Where KFS Diagnosis was Given

Type of Facility	Percentage of Parents	Number of Parents
Specialized Care Facility	40.0%	6
Hospital	33.3%	5
Private Practice	20.0%	3
Clinic	6.7%	1
Total	100.0%	15

Parents were asked to report the location of the facility in proximity to their home at the time of diagnosis (see Table 14). Of the responses, 33.3% (*n* = 5) of parents reported the facility was in the same region, 33.3% (*n* = 5) of parents reported the facility was in another region of the same state; 26.7% (*n* = 4) of parents reported the facility was in the same city; and 6.7% (*n* = 1) reported the facility was in another state.

Table 14

Facility Proximity to Parents' Home

Proximity	Percentage of Parents	Number of Parents
In the Same Region	33.3%	5
In another Region of the Same State	33.3%	5
In the same City	26.7%	4
In another State	6.7%	1
Total	100.0%	15

Second Opinion and Changes in Living Arrangements

Parents were asked if they sought a second opinion to confirm the KFS diagnosis for their child. Regarding a second opinion, 60% (n = 9) of parents reported they sought a second opinion to confirm the diagnosis, and 40% (n = 6) of parents reported they did not seek a second opinion.

Parents were asked if knowledge of the KFS diagnosis led them to change their living arrangements by moving from one place to another. Regarding living arrangements, 86.7% (n = 13) of parents reported no changes; 6.7% (n = 1) reported the family moved to get closer to a physician or treatment facility; and 6.7% (n = 1) reported being unable to pay their mortgage and care for the KFS child.

Resources Beneficial for Helping the Family Live with KFS

Parents were asked to select multiple answers from a provided list regarding resources that had been beneficial in helping their family to live with KFS (see Table 15). Of these responses, 60% (n = 9) of parents reported that a support group was beneficial; 53.3% (n = 8) of parents reported physicians, nurses, and other health professionals were

beneficial; 46.7% (*n* = 7) of parents reported health insurance was beneficial; 40% (*n* = 6) of parents reported family discussion/ problem solving was beneficial; 33.3% (*n* = 5) of parents reported family emotional expression/ mutual support was beneficial; 33.3% (*n* = 5) of parents reported religion/faith/spirituality/positive outlook were beneficial; and 33.3% (*n* = 5) of parents reported extended family/friends were beneficial. In addition, other responses included: 26.7% (*n* = 4) of parents reported income was beneficial; 26.7% (*n* = 4) of parents reported work schedule was beneficial; 13.3% (*n* = 2) of parents reported a family therapist was beneficial; 6.7% (*n* = 1) reported geographical location was beneficial; 6.7% (*n* = 1) reported being a stay at home parent was beneficial; and 6.7% (*n* = 1) reported a psychologist/counselor was beneficial (no parent reported an integrated healthcare facility was beneficial).

Table 15

Resources Beneficial for Living with a KFS Child

Resource/Benefit	Percentage of Parents Per Resource/Benefit	Number of Parents	Percentage of Total Responses
Support Group	60.0%	9	15.5%
Physicians, Nurses, and Other Health Professionals	53.3%	8	13.8%
Health Insurance	46.7%	7	12.1%
Family Discussion/Problem Solving	40.0%	6	10.3%
Family Emotional Expression/Mutual Support	33.3%	5	8.6%
Religion/Faith/Spirituality/Positive Outlook	33.3%	5	8.6%
Extended Family/Friends	33.3%	5	8.6%
Income	26.7%	4	6.9%
Work Schedule	26.7%	4	6.9%
Family Therapist	13.3%	2	3.5%
Geographical Location	6.7%	1	1.7%
Stay at Home Parenting	6.7%	1	1.7%
Psychologist/Counselor	6.7%	1	1.7%
Integrated Healthcare Facility	0.0%	0	0.0%
Total	-	58	99.9%

Note. Parents selected multiple responses.

In a separate question, parents were asked to report if they had ever met another person with KFS face-to-face with 14 parents responding, 85.7% ($n = 12$) of parents reported they had not met another person with KFS face-to-face; 7.1% ($n = 1$) reported meeting another KFS parent; and 7.1% ($n = 1$) reported that there were multiple family members with KFS.

Parents Evaluations of Physicians and Health Services Over the Previous 12 Months

Parents were asked to consider their interaction with specific physicians and health services over the previous 12 month period (see Tables 16-19). Overall, parents were asked to consider the following: Accessibility, personal cost, amount of travel, and appointment wait time.

Accessibility. Parents were asked to evaluate physicians and health services based on accessibility (see Table 16). A total of 7 parents reported that they were unable to access physicians and health services over the previous 12 months, as follows: 42.9% (n = 3) of parents reporting pain control specialist; 14.3% (n = 1) reported an orthopedist; 14.3% (n = 1) reported a neurologist; 14.3% (n = 1) reported a nephrologist; 14.3% (n = 1) reported a surgical operation; and no parent reported an audiologist, mental health provider, or family therapist. A total of 21 responses reported physicians and health services that were the easiest to access over the previous 12 months (see Table 15); 38.1% (n = 8) of parents reported an orthopedist was easiest to access; 19.1% (n = 4) of parents reported a neurologist; 14.3% (n = 3) of parents reported an audiologist; 14.3% (n = 3) of parents reported a family therapist; 9.5% (n = 2) of parents reported a mental health provider; 4.8% (n = 1) reported a pain control specialist. A total of 6 responses reported physicians and health services that were difficult to access over the previous 12 months (see Table 15); 50.0% (n = 3) of parents reported a pain control specialist was difficult to access; 16.7% (n = 1) reported an orthopedist; 16.7% (n = 1) reported a

neurologist; 16.7% (n = 1) reported a nephrologist; and no parents reported an audiologist, surgical operation, mental health provider, or family therapist were difficult to access.

Table 16

Parents' Evaluations of Accessibility for Physicians and Health Services Over the Previous 12 Months

Type of Physician or Health Service	Evaluation and Percentage of Parents					
	Unable to Access	Percentage of Parents	Easiest to Access	Percentage of Parents	Difficult to Access	Percentage of Parents
Orthopedist	1	14.3%	8	38.1%	1	16.7%
Neurologist	1	14.3%	4	19.1%	1	16.7%
Audiologist	0	0.0%	3	14.3%	0	0.0%
Nephrologist	1	14.3%	0	0.0%	1	16.7%
Pain Control Specialist	3	42.9%	1	4.8%	3	50.0%
Surgical Operation	1	14.3%	0	0.0%	0	0.0%
Mental Health Provider	0	0.0%	2	9.5%	0	0.0%
Family Therapist	0	0.0%	3	14.3%	0	0.0%
Total Responses	7	100.1%	21	100.1%	6	100.0%

Note. Parents were allowed to select multiple responses or not respond.

Personal cost. Parents were asked to evaluate physicians and health services based on personal cost over the previous 12 months (see Table 17). A total of 14 responses reported physicians and health services that were incurred at the least personal cost as follows: 35.7% (n = 5) of parents reported orthopedist; 21.4% (n = 3) of parents

reported audiologist; 14.3% (*n* = 2) of parents reported neurologist; 7.1% (*n* = 1) reported nephrologist; 7.1% (*n* = 1) reported pain control specialist; 7.1% (*n* = 1) reported mental health provider; 7.1% (*n* = 1) reported family therapist; and no parents reported surgical operation. A total of 10 responses reported physicians and health services that incurred the most personal cost over the previous 12 months; 40.0% (*n* = 4) of parents reported orthopedist; 40.0% (*n* = 4) of parents reported neurologist; 10.0% (*n* = 1) reported audiologist; 10.0% (*n* = 1) reported family therapist; and no parents reported nephrologist, pain control specialist, surgical operation, or mental health provider.

Table 17

Parents Evaluations of Personal Cost for Physicians and Health Services Over the Previous 12 Months

Type of Physician or Health Service	Evaluation and Percentage of Parents			
	Least Personal Cost	Percentage of Parents	Most Personal Cost	Percentage of Parents
Orthopedist	5	35.7%	4	40.0%
Neurologist	2	14.3%	4	40.0%
Audiologist	3	21.4%	1	10.0%
Nephrologist	1	7.1%	0	0.0%
Pain Control Specialist	1	7.1%	0	0.0%
Surgical Operation	0	0.0%	0	0.0%
Mental Health Provider	1	7.1%	0	0.0%
Family Therapist	1	7.1%	1	10.0%
Total Responses	14	99.8%	10	100.0%

Note. Parents were allowed to select multiple responses or not respond.

Amount of travel. Parents were asked to evaluate physicians and health services based on amount of travel over the previous 12 months (see Table 18). A total of 13

responses reported the least amount of travel for physicians and health services over the previous 12 months; 38.5% (n = 5) of parents reported orthopedist; 23.1% (n = 3) of parents reported family therapist; 15.4% (n = 2) of parents reported neurologist; 15.4% (n = 2) of parents reported audiologist; 7.7% (n = 1) reported a surgical operation; and no parents reported nephrologist, pain control specialist, or mental health provider. A total of 16 responses reported physicians and health services incurring the most amount of travel over the previous 12 months; 31.3% (n = 5) of parents reported orthopedist; 31.3% (n = 5) of parents reported neurologist; 12.5% (n = 2) of parents reported audiologist; 6.3% (n = 1) reported nephrologist; 6.3% (n = 1) reported pain control specialist; 6.3% (n = 1) reported mental health provider; 6.3% (n = 1) reported family therapist; and no parent reported surgical operation.

Table 18

Parents Evaluations of Amount of Travel for Physicians and Health Services Over the Previous 12 Months

Type of Physician or Health Service	Evaluation and Percentage of Parents			
	Least Amount of Travel	Percentage of Parents	Most Amount of Travel	Percentage of Parents
Orthopedist	5	38.5%	5	31.3%
Neurologist	2	15.4%	5	31.3%
Audiologist	2	15.4%	2	12.5%
Nephrologist	0	0.0%	1	6.3%
Pain Control Specialist	0	0.0%	1	6.3%
Surgical Operation	1	7.7%	0	0.0%
Mental Health Provider	0	0.0%	1	6.3%
Family Therapist	3	23.1%	1	6.3%
Total Responses	13	100.1%	16	100.3%

Note. Parents were allowed to select multiple responses or not respond.

Appointment wait time. Parents were asked to evaluate physicians and health services based on appointment wait time over the previous 12 months (see Table 19). A total of 15 responses reported the least appointment wait time over the previous 12 months; 46.7% (n = 7) of parents reported orthopedist; 26.7% (n = 4) of parents reported neurologist; 13.3% (n = 2) of parents reported audiologist; 6.7% (n = 1) reported pain control specialist; and no parents reported nephrologist, surgical operation, or mental health provider. A total of 7 responses reported the most appointment wait time over the previous 12 months; 28.6% (n = 2) of parents reported orthopedist; 14.3% (n = 1) reported neurologist; 14.3% (n = 1) reported audiologist; 14.3% (n = 1) reported nephrologist; 14.3% (n = 1) reported pain control specialist; 14.3% (n = 1) reported surgical operation; and no parent reported mental health provider or family therapist.

Table 19

Parents Evaluations of Appointment Wait Time for Physicians and Health Services Over the Previous 12 Months

Type of Physician or Health Service	Evaluation and Percentage of Parents			
	Least Appointment Wait Time	Percentage of Parents	Most Appointment Wait Time	Percentage of Parents
Orthopedist	7	46.7%	2	28.6%
Neurologist	4	26.7%	1	14.3%
Audiologist	2	13.3%	1	14.3%
Nephrologist	0	0.0%	1	14.3%
Pain Control Specialist	1	6.7%	1	14.3%
Surgical Operation	0	0.0%	1	14.3%
Mental Health Provider	0	0.0%	0	0.0%
Family Therapist	1	6.7%	0	0.0%
Total Responses	15	100.1%	7	100.1%

Note. Parents were allowed to select multiple responses or not respond.

Parents Rank Order Issues in KFS Treatment

Parents were asked to rank a series of five issues related to KFS treatment in order from 1 to 5, with 1 being the most difficult and 5 being the least difficult. Table 20 shows the number of parents who ranked each issue, and table 21 shows the overall percentage of parents who chose each rank. All 15 parents responded and ranked the following issues from 1 to 5: Medical expenses and health insurance; travel time to physicians or health services; finding physicians with KFS experience; managing appointments, physicians, and care; time involved in scheduling and waiting for an appointment. Regarding finding physicians with KFS experience, 60.0% (n = 9) of parents ranked it as 1, no parents ranked it as 2, 6.7% (n = 1) ranked it as 3, 13.3% (n = 2) of parents ranked it as 4, and 20.0% (n = 3) of parents ranked it as 5. Regarding travel time to physicians or health services, 13.3% (n = 2) of parents ranked it as 1, 46.7% (n = 7) of parents ranked it as 2, 6.7% (n = 1) ranked it as 3, 13.3% (n = 2) of parents ranked it as 4, and 20.0% (n=3) of parents ranked it as 5. Regarding managing appointments, physicians, and care, no parents ranked the issue as 1, 26.7% (n=4) of parents ranked it as 2, 46.7% (n = 7) of parents ranked it 3, 13.3% (n = 2) of parents ranked it 4, and 13.3% (n = 2) of parents ranked it as 5. Regarding time involved in scheduling and waiting for an appointment, 6.7% (n = 1) or 1 parent ranked it as 1, 26.7% (n = 4) of parents ranked it as 2, 13.3% (n = 2) of parents ranked it 3, 33.3% (n = 5) of parents ranked it as 4, and 20.0% (n = 3) of parents ranked it 5. Regarding medical expenses and health insurances, 20.0% (n = 3) of parents ranked it as 1, no parents ranked it as 2, and 26.7% (n = 4) of parents

ranked it as 3, 26.7% (n = 4) of parents ranked it as 4, and 26.7% (n = 4) of parents ranked it as 5.

Table 20

Parents Rank Order of Issues in KFS Treatment

Issue	Number of Parents Who Ranked Each Issue (ranked from 1 to 5 with 1 being the most difficult and 5 being the least difficult).				
	Rank 1 (most difficult)	Rank 2	Rank 3	Rank 4	Rank 5 (least difficult)
Finding Physicians with KFS Experience	9	0	1	2	3
Travel Time to Physicians or Health Services	2	7	1	2	3
Managing Appointments, Physicians, and Care	0	4	7	2	2
Time Involved in Scheduling and Waiting for an Appointment	1	4	2	5	3
Medical Expenses and Health Insurance	3	0	4	4	4
Total	15	15	15	15	15

Table 21

Percentage of Parents in Rank Order Issues of KFS Treatment

Issue	Percentage of Parents Who Ranked Each Issue (ranked from 1 to 5 with 1 being the most difficult and 5 being the least difficult).				
	Rank 1 (most difficult)	Rank 2	Rank 3	Rank 4	Rank 5 (least difficult)
Finding Physicians with KFS Experience	60.0%	0.0%	6.7%	13.3%	20.0%
Travel Time to Physicians or Health Services	13.3%	46.7%	6.7%	13.3%	20.0%
Managing Appointments, Physicians, and Care	0.0%	26.7%	46.7%	13.3%	13.3%
Time Involved in Scheduling and Waiting for an Appointment	6.7%	26.7%	13.3%	33.3%	20.0%
Medical Expenses and Health Insurance	20.0%	0.0%	26.7%	26.7%	26.7%

Qualitative Findings

Experience of Diagnosis

A total of nine parents responded to the survey question: What else would you like to say about your family's experience of diagnosis of KFS? Two major themes and several subthemes emerged from these responses: Difficulties faced by parents in their child's KFS diagnosis and parental assets in managing a child's KFS diagnosis are reported below.

Difficulties faced by parents in their child's KFS diagnosis. The themes that represent parental difficulties were the following: Parents facing a lack of experienced or

accessible KFS physicians; Parents' experiences of invalidation with physicians; and Parental adjustments or experiences related to living with a child diagnosed with KFS.

Parents facing a lack of experienced or accessible KFS physicians. Several parents discussed the difficulty they experienced in their efforts to find a physician with KFS experience. Parents described the lack of available doctors in their geographical location or interactions with physicians who did not have KFS experience. This emerged as a theme of parents facing a lack of experienced or accessible KFS physicians. One parent stated, "We do not have physicians in our area equipped to deal with KFS." Another parent stated, "orthopedic department doctor confirmed KFS but said he was not knowledgable…." Parents provided accounts of seeing multiple specialty physicians while acknowledging they saw multiple physicians with no KFS experience as one parent stated, "Its been hard to not find many doctors who know about it[KFS]."

Parents' experiences of invalidation with physicians. Parents' experiences of being invalidated by physicians represented parent descriptions of negative interactions with physicians. For example, one parent reported "He acted as if we were only trying to figure out if he could continue to play football or not." Another parent reported, "…Neurosurgeon who seemed uninterested if not a surgical case." One parent discussed how doctors focused more on addressing the child's head shape and torticollis stating, "At first they brushed off his fusion in his neck…." This theme emerged from responses that highlighted parent's negative interactions with physicians.

Parental adjustments or experiences related to living with a child diagnosed with KFS. Parents mentioned many adjustments or experiences related to living with a child diagnosed with KFS. These adjustments and experiences represented many factors regarding KFS, including emotional discomfort, awareness of marginalization, need for understanding genetics, chronicity, co-occurring diagnoses, and financial impact.

Emotional discomfort emerged as a theme in two ways; parents discussed emotionally difficult conversations with their children or referenced emotional discomfort in their overall KFS experience. A couple of parents discussed having conversations with their children that were emotionally difficult. For example, one parent reported "My son has expressed that he does not want children because he doesn't want to pass the gene on. That breaks my heart." Another parent described the difficulty in explaining to the child that she could no longer do gymnastics noting "…she wants to be a wrestler but I told her a lot of things have to change now all the things she used to do she cannot do no more…." Additionally, parents made reference to their overall KFS experience as one parent described, "This whole experience has been tough …." Another parent went further stating, "KFS has turned our world upside down and continues to do so and will forever." One parent discussed the invalidation involved in living with KFS that underscored emotional discomfort stating, "…the pain no one can see or understand."

An additional adjustment was that of awareness of marginalization. One parent highlighted the need for research for KFS stating, "I think it's more than important that

we continue to push for research, understanding and for health professionals to take us seriously." Although the parent acknowledges invalidation, it is within the context of living with a disease that does have enough research to advance understanding.

One parent reported that two of her children and her husband were diagnosed with KFS. This was the only report of a family with multiple KFS members, as such the case draws attention to the family's need to understand the genetic impact of KFS as they adjusted to the reality of living with KFS. With multiple affected members, the family displayed an obvious desire to learn more about the genetic aspects and origin of KFS. Specifically, the parent described how the family explored their own genealogy in order to learn more about the occurrence of KFS in their family. It is important to note that the parent reports two other children were not diagnosed with KFS. The parent referencing the father, daughter, and son with KFS stated "…all 3 with KFS have short stature…Husband adopted but recently found bioMom (deceased) and family describes her as having skeletal issues. Another son of hers has short stature KFS features."

Some parents described experiences of managing chronicity in relation to KFS. For example, one parent discussed a child's ongoing issues with pain stating "…the pain is so bad at times or she don't want to move on the couch or her head will hurt…." The parent describes attempts to help the child stating, "I will massage… from back of her neck and her shoulder in her lower back…." Another parent referenced the overall experience with KFS stating, "It has been a long journey and will continue with this condition as we have learned over the last 13 years…."

Several parents discussed their experience with KFS as involving other co-occurring diagnoses. For example, in reference to the experience one parent stated, "…[KFS] almost monthly brings in new diagnoses and issues that we have to figure out." Other parents specifically named additional diagnoses, such as "…he also has PUF60" and "… Scheuermann's disease."

One parent discussed a negative financial impact in their experience with KFS. The parent stated, "…I have missed a lot of work we have so many bills and we struggle right now I'm still trying to look for work but we don't know what tomorrow brings…." The parent further explained anticipating a call for a specialist who would inform the family of the need for surgery, which could extend the already difficult financial situation.

Parental assets in managing a child's KFS diagnosis. Themes that represent parental assets in managing a child's KFS diagnosis are as follows: Parent's use of KFS knowledge; Utilizing social support; and the Importance of selecting physicians with KFS experience.

Parent's use of KFS knowledge. Parent's use of KFS knowledge was evident as a theme in several ways. Parents spoke of learning about KFS as a way to improve their child's situation, as one parent stated, "We are trying learn more about KFS and who he will adapt when he becomes An[*sic*] adult." KFS knowledge was also used by parents to inform treatment and bolster parental advocacy in interactions with physicians. For example, one parent stated "…lack of information was obtained by them[physicians]."

This parent clearly questioned the legitimacy of information used by physicians because the parent was knowledgeable about KFS. Another parent referencing an experience of following through with referrals for testing from a pediatrician stated "…but a test for his entire spine hasn't been completed." This parent's knowledge of KFS shows that the parent expected the test to be done. Based on the fact that it had not occurred, KFS knowledge influenced the parent's expectations in interactions with physicians. One parent completely disagreed with physicians stating, "They pushed for a helmet[treating the child's head shape]. When I said I knew something else was wrong I found my own doctors in my area and pushed for testing." For some of these parents, KFS knowledge served to put the parent in the expert role in interactions with physicians.

Utilizing social support. Parents' utilization of social support emerged as a theme in their experience of KFS diagnosis. One parent reported, "The KFS group has been amazing to be a part of and I am thankful for the support systems I have found through this jorney[*sic*]." Another parent stated, "The Facebook support group has also been a life savor." Yet another displays a sense of trust and safety in recommendations of a Facebook friend stating, "We flew from Nashville, TN to Philadelphia to see an Orthopedic Surgeon there that came highly recommended by a FB friend that has a son with KFS in Chattanooga."

Importance of selecting physicians with KFS experience. A final theme that emerged as beneficial to parents in their experience of KFS diagnosis was the importance of selecting physicians with KFS experience. Parents emphasized the importance of

finding a physician with KFS experience. One parent described this as "Find the right docotor[*sic*]." Another parent discusses the benefit of access to experienced physicians stating, "I was lucky and they[hospitals] have great doctors in my area who have a strong background in this." Another parent stated, "[The] Optamologist[*sic*] gives us the most info & her geneticist is full of info as well."

Reasons for Using an Online Support Group

A total of 14 parents responded to the survey question: Please, describe your reasons for using an online KFS support group. Parent responses represented three themes: Information about KFS; Communion with others; and Concerns about their child's future.

Information about KFS. Information about KFS was represented in a majority of parent descriptions of seeking online support in order to gain information about KFS and physicians. Some parents simply stated using the support group for information: "To find out more information about physicians and research the condition." For some parents, the information was used to assist in planning their own child's treatment. For example, one parent reported "It has been the best guide to getting my child everything he needs. I would have never known where to start." Another parent stated, "Information! All of us are seeing different specialists, with different opinions and I find it critical to consider different options and directions when getting treatment for associated conditions especially[*sic*]."

Communion with others. Communion was represented in many parent descriptions of seeking online support to relate to others in similar circumstances which helped participants to feel validated and gain perspective about one's own experience with KFS. One parent underscored the value of relating to others by stating, "To have others to vent to, share accomplishments and concerns with. They are people that know exactly what we go through without doubt…" Other descriptions explained how parents used the group to gain perspective about their situation. For example, "To understand how other children cope…" and "To meet other parents of kfs[*sic*] children." Another parent reported, "Since it is so rare we want to hear from others like us in our situations."

Concerns about the child's future. Concerns about the child's future were represented in a few parent descriptions. Parents' described using the online support group to gain information and insight about outcomes for KFS children as they age. One parent stated, "…we were scared for our child and what this means for him and his future." Other parents spoke of preparing and planning in relation to their child's KFS. For example, "…it gives me the support I need to know what to look for in the future" and "… what we will face when he gets older."

Utilizing Physicians and Health Services

A total of seven parents responded to the question: What else would you like to say about utilizing or interacting with physicians or health services? One parent response was excluded because the parent reported not understanding the question. Parent responses represented two overall categories: needs in treatment and care along with

factors that impact utilization. Some parents described a need for knowledgeable physicians in treatment. For example, "Physicians need more education about KFS [and] about other health issues attributed to KFS…" The same parent described a need for integrated care stating, "…so the proper doctors evaluate the whole child and not just the spine." Other factors attributed to healthcare utilization were parent reports of physicians with KFS knowledge, collaborative parent-physician interaction, positive facility experiences, and one parent who reported utilizing care based on the affected family member's need for care. A parent who described utilizing a physician with KFS knowledge and collaborative parent-physician interaction reported, "Most of them that we encountered knew or had heard about KFS, the ones that had not were open too and willing to learn and understand it more." Some parents described positive experiences using hospitals. One parent stated, "Our son is a patient at Children's Hospital Los Angeles and they are AWESOME!" The parent who described utilizing physicians and services based on the affected family member's need was the only case in the sample of parents reporting multiple family member's with KFS. The parent reported her husband, adult son, and adolescent daughter had all been diagnosed with KFS. The parent reported that although the daughter consulted a neurosurgeon annually, the father had never seen a specialist for KFS, and the college aged son had not seen a specialist for KFS in years.

Accessing Physicians and Health Services

A total of four parents responded to the survey question: What else would you like to say about your ability to access physicians or health services or get the services

you need? One parent response was excluded because the parent reported not understanding the question. Parent responses represented two overall categories: barriers in accessing care and factors that benefit access. Barriers in care represented factors that made the parent's ability to access physicians and health services more difficult, while factors that benefited access made a positive contribution to care access. Of the barriers identified in accessing care, parents reported travel and disease barriers. Regarding travel, one parent reported, "More availability of doctors familiar with KFS are needed so travel doesn't become the main issue." One parent response identified a lack of collaborative care and disease information as barriers, along with KFS as a pre-existing condition acting as a possible future barrier. The parent stated, "Honestly, I don't know who my son should be seeing…. I worry about him losing access to health care due to [a] pre-existing condition." Factors that benefit access were geographical location and agency. Regarding geographical location, one parent responded, "I think we were fortunate because we live in Atlanta and are surrounded by amazing physicians who were eager and willing to help our son and us." Regarding agency, one parent stated, "He[adult child] felt that chiropractic care has so far been the most helpful for him."

Advice to Other Parents

A total of nine parents responded to the survey question: Please consider all of your experience with KFS, including information that has not been asked about. If you could give another parent of a child with KFS advice about any aspect of managing the disorder (physicians, insurance or even their own decision-making), please share any

advice not previously discussed in this survey in the space below. Parent responses represented the following categories: Parent education, parent intuition, parent organization, parent advocacy, physician selection, respecting the child's illness experience, and educating others.

Responses that described parent education reinforced the need for parents to learn about KFS. For example, "You have to be more educated than the physicians" and "…research and learn all you can on your own and bring it with you." Parents spoke of following and trusting their own instincts as important. For example, "…listen to their gut instinct" and "My best advice is to trust your instincts! … Don't let inexperienced doctor's[*sic*] overrule your instincts." Regarding parent organization, parents discussed the importance of using organizational strategies to keep track of all health related information related to KFS. For example, "…start a binder. Bring that binder with you to every appointment. It's also good to keep medical records in as well." Parent advocacy was represented by several parents who gave advice encouraging parents to take an active role in KFS treatment. For example, "I would encourage them to keep pushing ahead and meeting with doctors as money and travel allows…" and "If you feel you are getting bad information or recommendations, get a second opinion." Physician selection was reported by a few parents who discussed the importance of selecting a physician. For example, "…try to find a doctor that can understand this disease…" and "…to find the right physician for you and their family." Respecting the child's illness experience was represented by several parents who gave advice recommending that KFS parents remember the KFS child may also face difficulties, and therefore implored KFS

parents to acknowledge the child's situation. For example, "…remember that your child needs your help and that they are dealing with this each day and feel completely alone" and "…try to also have care for your child because they're dealing with a lot too give them lots of hugs." One parent wrote about educating others as a way to help them understand and respond to her child more positively. The parent discussed writing about her child's illness in a story and having the child take the story to school at the beginning of each school year to help other children understand KFS and prevent teasing.

Summary

A main finding from the results of this study is that parents signify their difficulty in finding physicians experienced in the treatment of KFS. An additional finding was that parents emphasize the importance of online support as a positive influence in the treatment and management of their child's KFS. The descriptive survey results provide information regarding testing, diagnoses, and symptomology, along with information about accessing and utilizing physicians and health services. Parents' qualitative responses outline difficulties they face along with beneficial strategies and resources for managing their child's KFS diagnosis.

CHAPTER V

DISCUSSION

The survey data generated by the 15 parents who participated in this qualitative study yielded valuable information that will be useful in building a foundation for understanding the experience of diagnosis of Klippel-Feil Syndrome in the United States; use of health services for children diagnosed with KFS; and, usefulness of online support groups for parents raising children diagnosed with KFS. The discussion below will follow the three major themes: experience of diagnosis; barriers in KFS treatment, and the role of online support.

Experience of Diagnosis

Parent reports about their child's KFS diagnosis led to the identification of two types of disease onset in children with KFS, indications of a progressive illness course, and evidence of psychosocial issues related to parents' experiences of KFS diagnosis. Additionally, this research highlights ways in which the degree of uncertainty in KFS treatment was evidenced in parents' descriptions and how that uncertainty manifested in different aspects of treatment. Resiliency is discussed in relation to support group use and parental knowledge of KFS. KFS barriers are discussed in relation to overall and specific issues in KFS treatment and management.

Rolland's Family Systems Illness Model

Rolland's (1994) family systems illness model (FSIM) is used as the lens to view the information provided by parents in this research. Because Rolland's model aims to clarify the reality of an illness to a family, it was useful in this research for understanding the parameters of KFS and discussing psychosocial issues that impact family adjustment. Specifically, this researcher found evidence of KFS information regarding onset, course, and degree of uncertainty. Despite discussing the FSIM as categorical, Rolland asserts that the variables within his model occur on a continuum, with each category representing "key anchor points along the continuum" (Rolland, 1994, p. 23). Thus, this researcher asserts that the information discussed here simply occurs on a continuum, with the goal of elaborating some of the possibilities of KFS onset and course. Rolland described the degree of uncertainty in illness as including variability in the nature of onset, course, outcome, or incapacitation, and variability in the rate at which an illness progresses. The flexibility in Rolland's interpretation is useful for understanding the variability found between participants in this research and for overall differences in KFS patients. Overall, Rolland's model is useful in this research for understanding the multitude of systemic implications for parents who have a child diagnosed with KFS (Rolland, 1994).

Variability in KFS onset. As described by Rolland's (1994) FSIM, this study found cases of KFS with an acute presentation in early childhood and acute presentation in adolescence with the variation in presentation explained by Rolland's conceptualization of onset as a continuum, which can also exhibit unpredictable characteristics. Overall, the vast majority of parents in this study had a

child diagnosed with KFS before the age of 2, with a majority of parents reporting there was no delay in their obtaining a diagnosis of KFS for their child. While this information shows a positive trend toward early diagnosis in the United States, many rare disease patients in the EurordisCare 2 study experienced delays in diagnosis, which was viewed as a major barrier by that study (Kole & Faurisson, 2009). Likewise, many KFS parents did report issues related to delays in diagnosis from physical pain to loss of confidence in healthcare; most notable was a child who concussed before getting a KFS diagnosis at age 16.

The second group represented parents of adolescents who described their experience of the diagnosis as acute, with two of the three parents discovering KFS incidentally after their child was injured. In addition to the adolescent who experienced a concussion (described in the paragraph above), another parent of a child diagnosed at age 16 discovered KFS following an accident the child had while playing football, with a third adolescent diagnosed at age 11 who initially presented with a swollen neck. Although the parent of the 11year-old did not report that the symptom was associated with an injury, KFS patients are typically urged to avoid physical activities that could increase the risk of trauma (NORD, 2016a). It is possible that the child suffered an injury that prompted treatment. Findings that indicate some children may not present with symptomology that leads physicians to discover KFS supports research by Samartzis et al. (2016) that shows not all children present with the clinical triad (short neck, limited range of cervical movement, and low posterior hairline) for KFS, and the triad does not

have to be present for the patient to have KFS. Unfortunately, this research study shows delays in diagnosis may lead to increased risk of injury for children who go undiagnosed.

The disparity in diagnosis for those in early childhood versus adolescence is a finding in this research that is concerning. Rolland (1994) affirms that illnesses can be unpredictable in the nature of their onset; although KFS is a congenital condition, the fact that some children in this research were undiagnosed until adolescence highlights the complexity of this rare disease. There were children in this study who presented with notable symptomology related to their neck in early childhood. Specifically, parents reported young children who were diagnosed with torticollis, scoliosis, or other conditions related to the spine that represented visual symptomology which occurred in the first year of the child's life. The second group represented adolescents who were not diagnosed with specific spinal conditions (one child was diagnosed with idiopathic short stature) in early childhood whose parents discovered KFS following an injury or problem with the child's neck. None of the KFS children diagnosed as adolescents in the study reported visual symptomology in early childhood, such as torticollis or scoliosis.

Furthermore, it is important to consider possible differences in psychosocial adjustment related to the disparity in KFS onset. Family adjustment to illness with KFS children diagnosed in adolescence may evolve differently compared to young children. An adolescent may have to sacrifice a beloved sport or activity reflective of identity in order to prevent injury for a disorder that is newly diagnosed, yet cannot be physically observed by the child or others. Families with children diagnosed in early childhood have the capacity to adjust to the child's physical limitations and focus on nonrestrictive

activities as the family unit learns to become a family living with KFS before the child even knows what the disease means. Although the initial adjustment may be different, this is not meant to assume that diagnosis at one age is more difficult than the other, just that families may have differing issues to resolve with respect to the child's age. The young children in this study presented with symptomology that underscores visual characteristics, such as scoliosis which will likely progress with the child's growth. Rolland (1994) points out that although visual symptoms allow others to be more objective and discerning in their interactions with respect to illness, the disadvantage is the possibility of stigma. On the other hand, invisible symptoms such as those identified earlier with the adolescent group could increase the potential for experiences of invalidation and may even serve to escalate any existing relational dysfunction when family members disagree about the true impact of the illness (Rolland, 1994).

KFS chronicity. Although this current research cannot elaborate a specific illness course for KFS, parents described difficulties they face in their child's diagnosis of KFS that indicate a progressive illness course. Specifically, several parents discussed chronic issues related to their children being in physical pain. One mother discussed how her now 20-year-old son uses medical marijuana to manage pain. Another mother reported that even after 13 years since the KFS diagnoses, new diagnoses related to KFS continue to emerge. The same parent stated, "It has been a long journey and will continue with this condition…." Although this current research supports the idea that some of the parents' children in the study experience a progressive illness course, it is

possible that due to the variability in KFS presentation (NORD, 2016a) that others may experience the illness as relapsing or constant; hence, more research is needed.

Psychosocial issues. A majority of parents reported their children were currently adolescents. Not surprisingly, several of these parents discussed psychosocial issues related to their child's current developmental stage along with issues related to the family's experience of living with KFS. These psychosocial issues emerged in themes related to parental adjustment in living with KFS; these themes are discussed below.

Physical restriction and physical presentation. One parent discussed the difficulty of communicating to her 12-year-old that she was unable to continue gymnastics and could not pursue wrestling as a sport. This is similar to research on scoliotic adolescents that has shown physical restriction and adjusting to treatment routines are important psychosocial issues (Reichel & Schanz, 2003). These issues may not fully represent the spectrum of difficulties that KFS adolescents may face, especially if their physical presentation is a manifestation of multiple deformities, which are not easily hidden. Many of the parents in this study reported additional diagnoses or conditions that occurred in conjunction with KFS and denoted additional visual symptomology, such as Sprengel's deformity. Many of these visual symptoms are not easy, if not impossible, to obscure. It is possible that adolescents in this research face similar psychosocial concerns as scoliotic adolescents who attempt to hide their disfiguring condition (Reichel & Schanz, 2003).

Emotional discomfort. Several parents discussed emotional discomfort in their experience of KFS diagnosis, mostly in relation to their overall KFS experience. One parent stated "KFS has turned our world upside down…" and another stated "This whole experience has been tough…." The presence of KFS in the life of a family indicates increased psychosocial demand and adjustment that both impact the family system. One parent broadly stated, "…the pain no one can see or understand." This parent highlights the uncertainty that colors the reality of rare disease as though no one has or could possibly understand the parent's experience. Uncertainty in KFS is discussed later in this research.

Genetics. Only one parent reported a case of multiple family members diagnosed with KFS. The parent stated that "…doctors may say KFS is not genetic, but clearly in our family, it is [genetic] and a dominant mutation." In fact, Rolland (1994) asserts that a family's understanding of genetics is a significant psychosocial issue. Procreation and emotional adaptation are impacted by a family's ability to adequately and accurately communicate the possibilities of genetic transmission (Rolland, 1994).

Although KFS can be inherited genetically, most cases are thought to occur spontaneously (NORD, 2016a). The family in this study had two children and one parent diagnosed with KFS. The family linked the genetic connection themselves by synthesizing symptomology and investigating their genealogy, despite their interaction with physicians who did not indicate their case was genetically linked. There is no

definitive genetic test for KFS, and this family had no assistance from medical professionals in mapping genetic inheritance.

One parent of an 18-year-old reported that it was difficult for her when her son expressed he does not want to have children for fear of passing on the gene that causes KFS stating "That breaks my heart." This finding supports previous research that shows parents have difficulty communicating with their children about genetics (Gallo et al., 2005; McAllister et al., 2007; Metcalfe et al., 2011). Similarly, some scoliotics reported that their condition impacted decisions regarding procreation (Danielsson & Nachemson, 2001).

Multiple diagnoses. Several parents in the study who had children with other additional co-occurring diagnosis or diagnoses discussed the added complexity of having a child with multiple diagnoses. Most significantly, some of the conditions mentioned were additional rare diagnoses, such as Scheuermann's disease (National Institutes of Health, 2017) and PUF60 (Unique: Rare Chromosome Disorder Support Group, 2017), adding to the complexity of disease treatment and management. One parent acknowledged that their KFS child continually has new diagnoses to manage. While KFS denotes fusions of the cervical spine, it also has a high correlation with scoliosis (NORD, 2016a; Samartzis et al., 2011; Thomsen et al., 1997; Xue et al., 2014), not to mention a plethora of other related diagnoses that are, like scoliosis, more common (NORD, 2016a). Diagnoses included a range of other conditions from Sprengel's deformity to hearing loss, scoliosis, and conditions of the kidney. Clearly, families in this

study had additional, and sometimes complex, conditions to treat and manage along with KFS.

Uncertainty in KFS. Rolland (1994) states the uncertainty of an illness is an overarching concept that influences a family's response. Illnesses with highly predictable courses and outcomes allow families to plan and make decisions knowing the issues and limitations they may face; while other illnesses may be more unpredictable in terms of unexpected changes in course over time and lesser known time frames for anticipated changes. Knowing the parameters of an illness allows families to adapt to necessary changes and effectively reduce ambiguity, yet some families must learn to accept the uncertainty that accompanies a diagnosis (Rolland, 1994). For KFS families, uncertainty seems to plague their efforts to definitively determine treatment with physicians who have experience with KFS and can provide answers about the short and long term implications of KFS. The ambiguity and uncertainty of living with a KFS diagnosis was evident in this research in several ways; uncertainty in treatment; uncertainty regarding the implications of KFS over the child's lifespan; and uncertainty in relation to accessing and utilizing physicians.

Uncertainty in treatment. Parents expressed uncertainty about the current treatment of their child's KFS. One parent of a 16-year-old reported discussing trying to gain more knowledge of KFS in order to plan for the child's adaptation to adulthood, especially since the child had a co-occurring diagnosis. One parent discussed witnessing the child's pain, trying to massage the child to reduce pain, while simultaneously waiting

for a call from a neurosurgeon regarding surgery stating "… we don't know what tomorrow brings…." Interestingly, one parent not only displayed an attitude of agency, but simultaneously drew attention to the plight of patients impacted by rare diseases stating, "I think it's more than important that we continue to push for research, understanding…." This highlights the parent's awareness of living in the context of rare disease, even an awareness of marginalized status. This parent is fully aware that research is needed, and that research could possibly provide definitive answers about KFS. A parent who is imploring the need for research about their child's disease is a parent living in the presence of uncertainty. Systemically, it is likely that the uncertainty experienced by the KFS parents in this study does not exist in isolation. Although this research underscores the difficulty of parents living in uncertainty, Reichel and Schanz (2003) reported that uncertainty regarding disease progression is an important psychosocial concern for adolescent scoliotics. It is highly probable that the children and adolescents in this study experience their own uncertainty regarding KFS progression.

Uncertainty about the child's future. An additional theme that draws attention to parents living with uncertainty is supported by several parents wanting to know what KFS meant for their child's future. As one parent stated regarding the benefits of using the online support group, "To understand how other children cope and what to possibly expect in the future." With very limited studies on long term health outcomes for KFS (Theiss et al. 1997), it is not surprising that parents seek alternative means to supplement their knowledge of KFS, reduce ambiguity, and plan for future possibilities. Rolland

(1994) asserts illnesses accompanied by a high degree of uncertainty force families to plan their lives in ways that allow for increased flexibility.

Difficulty finding physicians. Parents that discussed difficulty finding physicians with KFS experience connected the experience to uncertainty about KFS treatment. For example, one parent stated "Its been hard to not find many doctors who know about it or [*sic*] what's next to come and all [the] different doctors we see." Another parent discussed being disillusioned about what to do next in treatment, while noting that there were no physicians in the parent's geographical area with KFS experience. The lack of experienced physicians seemed to amplify parental uncertainty surrounding KFS and complicate the parent's ability to make decisions regarding treatment. One parent stated "Honestly, I don't know who my son should be seeing." Although difficulty accessing doctors with rare disease experience is documented in research literature (Kole & Faurisson, 2009; Shire, 2013), a rare diagnosis may be a parent's awakening to the complex issues involved in rare disease care. It is plausible that these parents experience cognitive dissonance when help is not accessible, or when it is accessible, physicians lack experience with KFS. As one parent stated, "That doctor confirmed KFS and Scheuermann's but said there was not much to be done other than pain management if needed." Parents must adapt to the difficulties involved in the context of rare disease, along with the realization that treatment will focus on alleviating symptoms or addressing associated conditions, not curing the disorder (NORD, 2016a).

Physician invalidation. Parents discussed experiencing invalidation in their child's KFS diagnosis, in reference to emotional discomfort and particularly with physician interactions which underscore uncertainty. Regarding invalidation in physician interactions, one parent stated, "He acted as if we were only trying to figure out if he could continue to play football or not." Another parent stated, "At first they [physicians] brushed off his fusion in his neck…." Considering the difficulty parents face in finding physicians with KFS experience, negative physician interactions only serve to complicate treatment and further invalidate the parent's attempts to intervene on their child's behalf. Even when parents can find experienced physicians, what happens when they do not like the physician? How does the relationship impact KFS management and treatment? Research shows that rare disease patients prefer to interact with physicians who are empathetic and will help them determine a course of treatment (Huyard, 2009). It is possible that some of these invalidating experiences contribute to parents feeling rejected by health professionals in their efforts to understand KFS, seek treatment, and determine an appropriate treatment course, which further contributes to parental uncertainty about KFS treatment. The Eurordis surveys reported experiences of rejection with patients who felt physicians were hesitant to treat them because of the complexity of their diagnosis (Kole & Faurisson, 2009).

Physicians with KFS experience. Conversely, some parents were grateful for being able to find a physician with KFS experience. One parent described physician selection in terms of "Find the right docotor[*sic*]." Finding a physician with experience

was a key piece of advice one parent offered to other KFS parents. As another parent stated, "…try to find a doctor that can understand this disease…." A parent who discussed having access to knowledgeable doctors had a more favorable opinion of physician experiences. For example, "I was lucky and they[hospital] have great doctors in my area who have a strong background in this." Other parents with favorable interactions described "amazing" physicians and a hospital that was "AWESOME!" It stands to reason that physicians with KFS experience play a significant and pivotal role in providing knowledge and guidance to parents in their child's KFS treatment.

Illustrated areas of uncertainty in KFS. It is obvious that KFS parents experience uncertainty around many issues related to KFS treatment and management. Rolland (1994) denotes the overall nature of uncertainty and its significant impact. In an effort to illustrate the manner in which KFS parents expressed uncertainty (previously discussed), Figure 1 is provided below. Parental questions in Figure 1 are derived from the research to represent questions pertinent to KFS parents.

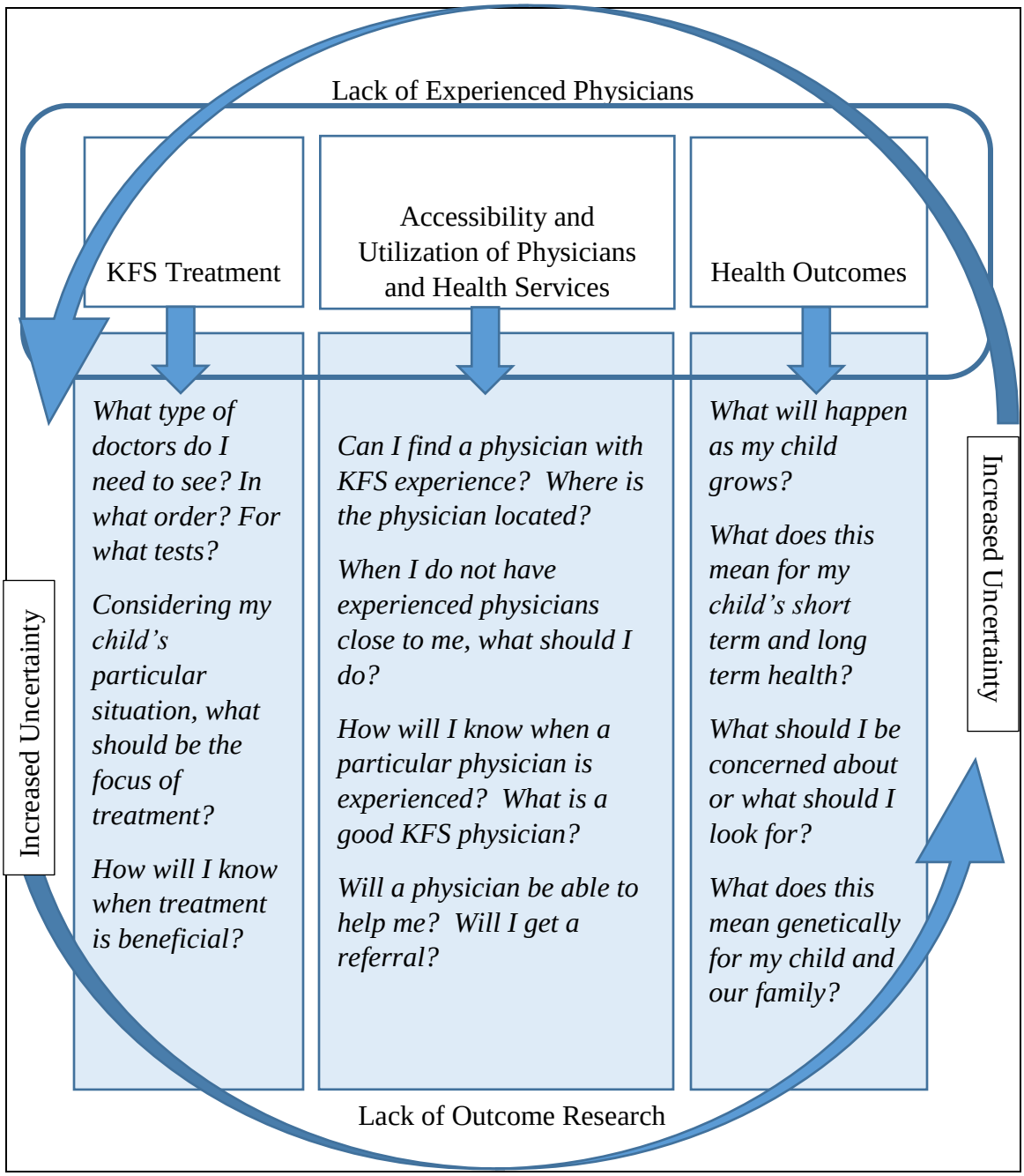

Figure 1. Areas of Uncertainty for KFS Parents

Integrating Models

Rolland's (1994) Family Systems Illness Model provides a comprehensive foundation for evaluating the findings in this research study. Specifically, Rolland appreciates the significance of psychosocial issues in the treatment of illness; he draws attention to family systems and ways in which family members are inherently intertwined and take positions when facing illness. Although Rolland acknowledges contextual family factors that impact illness such as a family dynamics and beliefs, larger sociological issues that exist when multiple systems intersect to impact families are not comprehensively addressed; however, these systemic interactions are important for elaborating psychosocial issues for the participants in this research. Walsh's (2003, 2016) attention to contexts, especially multisystemic contexts, provides greater clarity for elaborating issues specific to rare disease and the participants in this study. Furthermore, while the context of rare disease as observed by a person unversed in the difficulties it imposes could risk overly focusing on deficits, Walsh's model helps to elaborate known resources and processes that support resiliency and provide direction for establishing interventions that respect the strengths of this population (Walsh, 2003, 2016).

Walsh's Concept of Resiliency

Walsh's concept of resiliency (2003, 2016) can be applied to the findings in this study to extend the illness perspective of KFS to include processes that are beneficial to adjusting to KFS. Rolland (1994) asserts that research should focus more on drawing attention to adjustment processes that are shared between illnesses, instead of describing

family adjustment for a specific illness and highlighting differences. This research seeks to use Walsh's concept of resiliency as a way to highlight processes that build resiliency, which have been evidenced in a plethora of family situations and have the potential to be more globally shared in families (Walsh, 2003, 2016). Furthermore, as clinicians may see a family at any point in the life cycle of illness (Rolland, 1994), understanding processes that capitalize on or mobilize a family's existing strengths (Walsh, 2003) have the potential to increase positive therapeutic outcomes.

Parental Use of KFS Knowledge. Several parents who referenced interactions with physicians often provided evidence of how their own KFS knowledge was used for treatment purposes; as one parent stated "…learn all you can on your own and bring it with you." Parents indicated that treatment was so complex, in relation to KFS knowledge and interactions with specialists, that taking information with them often meant the parent also needed to be very organized. As one parent stated, "Ask for all documents from the physicians & start a binder." Bring that binder with you to every appointment." Parents spoke of KFS knowledge as though obtaining and learning it were not an option. One parent stated, "You have to be more educated than the physicians." This shows that parents plan for their interactions with physicians by educating themselves, determining expectations for treatment, and waiting for referrals or appointments that might provide some answers within an ambiguous disease context. Physician interactions were preceded by parents planning and allotting time for purposes of finding the "right" physician. One parent stated, "Our pediatrician hasn't followed up

with other testing such as cardiac and only took a urine sample to test his kidneys. She referred us to a spine surgeon…but a test for his entire spine hasn't been completed." This parent expresses expectations of treatment based on the parent's own knowledge of KFS, which was in line with information provided about testing posted by *Klippel-Feil Syndrome Freedom.*

Expert role of parents. Some parents' interactions with physicians went beyond a parent's basic use of disease information and seemed to indicate that some parents' assumed more of an expert role. One parent stated, "When I said I knew something else was wrong, I found my own doctors in my area and pushed for testing." Some parents urged others to use their own power and intuition, as one parent stated "Don't let inexperienced doctor's overrule your instincts." It is plausible that a traditional patient-provider relationship is characterized as one in which the physician retains the expert knowledge and thus a higher degree of influence and power. In the context of rare disease and specifically KFS, when physicians do not have greater knowledge, parents seem to assume the role of gaining knowledge and acting as the expert. It is possible that this is evidence of a key process for rare disease parents in building resiliency (Walsh, 2003), perhaps an outcome that hopes to reduce ambiguity. Walsh (2003) discusses the key process of collaborative problem solving as one where families set and focus on goals, learn from failures, and assume a proactive stance. In this research, parents not only acknowledged the difficulties they face in KFS treatment but did so while calling attention to their position as advocates for their children. One parent stated, "I would

encourage them[other KFS parents] to keep pushing ahead…" and another parent stated "Be their[KFS children] advocate and fight for them."

Although parents certainly seem to demonstrate problem solving in their expert role positions, it is important to note that open emotional expression where members can share mutual empathy is also an important process for building resiliency (Walsh, 2003). With respect to illness, physicians essentially become a member of the family (Rolland, 1994), so it stands to reason that open emotional expression with physicians is significant for building resilience in KFS families. As one parent described an interaction with physicians stating, "… physicians who were eager and willing to help our son and us." This may provide partial support for research that shows rare disease patients actually do not expect their physicians to be experts, and that patients truly desire physicians who are empathetic to their situation and collaborate to plan treatment (Huyard, 2009). In essence, it is the combination of parental advocacy and physician empathy that builds collaboration, and by extension resiliency.

Resources for KFS Parents. Parents ranked support group, health professionals, health insurance, and family discussion and problem solving respectively as the top four most important resources in living with a KFS child. Walsh (2016) emphasizes the significance of multilevel systemic processes that occur over time with respect to the particular circumstances and resources available to a family, which are dependent on particular conditions or contexts. Interestingly, the parents in this study emphasize the significance of multilevel resources that contribute to KFS family adjustment. In

essence, parents defined specific family, community, societal, and economic resources underscoring Walsh's notion that it is the interplay between systems that impact and foster resiliency (see Figure 2).

Furthermore, as health insurance was noted by KFS parents as being beneficial, it stands to reason that a loss of coverage for KFS would greatly impact KFS families. While the state of healthcare in the United States remains a highly debated political topic, the KFS population would be immensely impacted by legislation that could preclude KFS treatment. KFS patients would be at risk for a loss of health insurance coverage if current protections for pre-existing conditions under the Affordable Care Act were absolved. As one parent stated, "I worry about him losing access to health care due to [having a] pre-existing condition." All the parents in this study reported their children had health insurance at the time of KFS diagnosis and had health insurance at the time the study was completed. In fact, parents ranked medical expenses and health insurance as the least difficult issue in KFS treatment; however, it is imperative to consider that a loss of this beneficial resource could negatively impact family resilience and contribute to increased uncertainty about KFS treatment and accessibility of care.

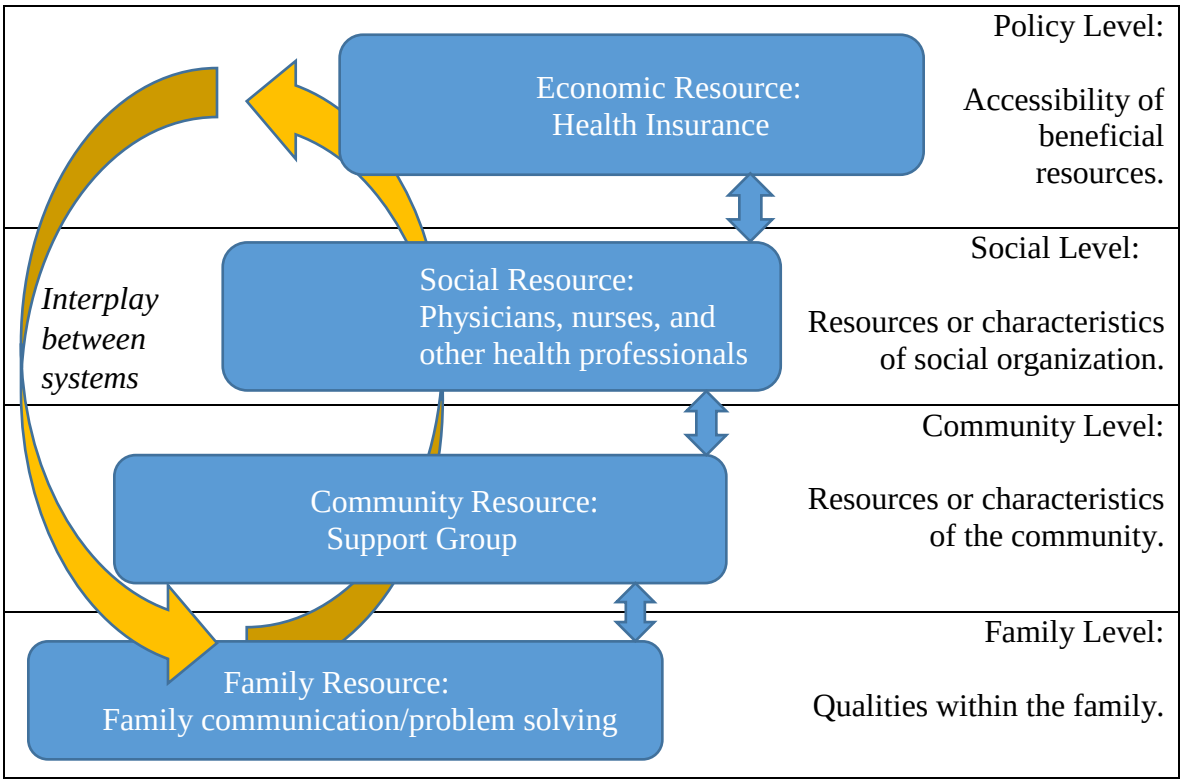

Figure 2. Multisystemic KFS Resources based on Walsh's (2016) Concept of Resiliency

It is significant that a majority of parents in this study rated their support group as the most beneficial resource to their adaptation to KFS, lending credit to the significance of interventions that involve communities and foster families to mobilize their own strengths in difficult times (Walsh, 2016). Walsh emphasizes the importance of a relational perspective because relationships are significant for helping families to adjust to adverse situations. Online support group use in the context of rare disease is an obvious strength that underscores the resiliency of the study population in establishing and maintaining beneficial communal connectedness (Walsh, 2016), despite facing difficulties in their experiences of diagnosis. The role of online support group use is discussed later.

Barriers in KFS Treatment

This research shows evidence of barriers faced by KFS parents. Specifically, parents faced difficulties accessing physicians with KFS experience, with some parents reporting experiences of rejection. Parents expressed accessibility and utilization difficulties regarding cost and travel for orthopedists, along with cost, travel, and appointment wait time for neurologists. Parents experienced barriers in accessibility with pain control specialist, but reported family therapists were easily accessible. Furthermore, parents did not report significant changes in living arrangements.

Finding Knowledgeable Physicians

Finding physicians with KFS experience was ranked as the most difficult issue in KFS treatment by a majority of parents, this supports previous research that shows a need for more physicians with rare disease expertise (Shire, 2013) and research showing patients face difficulties accessing treatment (Kole & Faurisson, 2009). More importantly, just over half of KFS parents reported that physicians, nurses, and other health professionals were a beneficial resource in living with a KFS child. This shows that KFS parents value the role physicians play, it also underscores the relational aspect of disease treatment outlined by Rolland (1994) as the therapeutic quadrangle.

The quadrangle envisions the patient, family, illness, and health-care team as individual family members, with fluctuating roles during the course of an illness. While Rolland's illness typology aims to provide some clarity about the identity of the illness as a member in the family (Rolland, 1994), the course of KFS is not well understood

because the illness is under researched and there are a lack of health outcome studies. It is possible that parental expectations for definitive and clarifying answers are high, directly impacting the relationship between the parent and physician. As previously discussed, parents reported experiences of invalidation in their interactions with physicians. These experiences are easily likened to patients that feel rejected by physicians (Kole & Faurisson, 2009). Rejection is a barrier in accessing the care KFS parents seek for their children.

Orthopedists, neurologists, pain control specialists, and family therapists. Most parents did not report major barriers in their ability to access orthopedists. In fact, some parents reported orthopedists were the easiest to access, and required the least amount of travel. Overall, orthopedists show some difficulty regarding cost and travel barriers, but orthopedists were accessible and utilized by the KFS parents in this study.

Neurologists, on the other hand, were shown by parents to have some difficulties in utilization. Neurologists were reported by a few parents as incurring the least personal cost and travel, while many others reported they incurred the most personal cost and travel, indicating that neurologists are difficult for some parents to utilize. Compared to orthopedists, fewer parents reported that neurologists had the least appointment wait time. Overall, neurologists were somewhat difficult for KFS families to utilize regarding cost, travel, and appointment wait time.

Parents showed difficulty in their ability to access a pain control specialists, many were unable to gain access while others reported difficulty accessing a pain control

specialist. A parent in the study reported being informed by a physician that pain management was not only an option, but that there were no other major medical interventions that could be done for the parent's child. It is not surprising that families in this study attempted to access a pain management specialist, but parents appear to face difficulty in accessing these physicians.

Family therapists were reported by several of the parents as requiring the least amount of travel, which shows accessibility for a population that has highlighted a lack of psychological care as a deficit in diagnosis (Kole & Faurisson, 2009). Furthermore, several parents claimed family therapists were easy to access along with mental health providers.

Though changes in living arrangements requiring rare disease patients to move have been shown in other research (Kole & Faurisson, 2009), a majority of KFS parents did not report significant changes in living arrangements. One parent reported moving to get closer to a physician or treatment facility.

Travel

Travel time to physicians or other health services was ranked as the second most difficult issue in KFS treatment by parents. A clear majority of parents reported traveling outside of the city they reside in when their child was diagnosed with KFS. A large majority of parents reported traveling to a specialized care facility or hospital when their child was diagnosed. This shows the complexity involved in KFS treatment and supports previous research that shows parents report difficulty managing care related to travel to

consult specialty physicians (McAllister et al., 2007). Overall, travel was a barrier for parents to obtaining care for KFS patients.

Usefulness of EurordisCare 2 and 3

The EurordisCare 2 and 3 surveys were useful in this research for delineating issues relevant to the study population. Use of the surveys as a guide for research inquiry was substantive and encompassed a majority of issues and areas of concern that the study sample affirmed as representative and reflective of their experiences. The surveys omitted qualitative responses in lieu of manageable efficiency using a substantially large research population, which the current study addressed with the inclusion of qualitative responses. The EurordisCare 2 and 3 surveys substantially influenced the structure and direction of this research study, providing invaluable insight about issues pertinent to rare disease patients. The surveys and the research they produced were an immense support to this research study, which was made possible because of the diligent efforts of the Eurordis organization to represent the voices of rare disease patients (Kole & Faurisson, 2009). Overall, the Eurordis surveys were useful for outlining the experiences of diagnosis for parents with a KFS child and directly contributed to the creation of the research survey.

Role of Online Support

Walsh (2003) emphasizes the vital role of sociocultural context and the importance of time across generations when evaluating risk and resiliency. As a support resource, *Klippel-Feil Syndrome Freedom* was first established on Facebook in October

2014. A majority of the children represented in this study were diagnosed with KFS before their parents had access to the Facebook support group. A majority of parents also reported that they have not met another KFS person face-to-face. The only exceptions were the family case, and one other parent who reported talking with another KFS parent. Organizationally, the administrator of the group who is a KFS advocate socially and is an active member of the closed parent group, frequently answers questions and provides information about KFS treatment and management within the group. Additionally, it is imperative to remember the overall context of rare disease and the significance of parents reporting emotional discomfort in their experiences of diagnosis and invalidation in their experiences with physicians.

Parents overwhelming shared that their use of the online support group enabled them to commune with others. Parents discussed ways in which the group validated their experience, as one parent reported "…it's always good to be in a tight group so we can have understanding for one another." Without access to others face-to-face and in a context laden with barriers, the online support group offered validation for many of the parents in this study. This supports previous research that shows online support groups serve to validate emotional experiences that can be invalidated in other social environments (Varga & Paulus, 2014).

The opportunity to have online communication not only validated the parents' experience but enabled them to access information from others they could trust. As one parent stated, "To have others to vent to, share accomplishments and concerns with. They

are people that know exactly what we go through without doubt and questions everyone is able to support and help with answers if needed." This supports other research about online support groups showing individuals prefer to have information from people with firsthand experience (Mo & Coulson, 2014). Parental trust within the group and value for lived experience framed parents' overall use of information. Parents discussed using the group as a way to compare their child's symptoms to other KFS children or inquire about specific issues. As each person with KFS can present with a myriad of symptoms and associated conditions (NORD, 2016a), disease presentation played an important role in driving parents to use the group to compare or inquire about similar symptoms or conditions. For example, parents expressed "…to see if they have the similar symptoms…," "To learn more about others with it…," and "I find it critical to consider different options and directions when getting treatment for associated conditions especially." The online support provided parents with a trusted avenue for gaining insight about the KFS experience of other children, and allowing parents to converse about specific symptoms and conditions with an audience that understands and lives with KFS. The findings in this qualitative study may provide partial support for research that showed disease specific informational and emotional support were important factors in posts of an online support group for a different rare disease (Coulson et al., 2007).

Parents' use of the online support group to foster understanding supports key processes in Walsh's (2003) concept of family resilience. Of Walsh's key processes that underscore the ability to make meaning of adversity, resilience as relational instead of

individual and contextualizing adversity are the most pertinent for the use of online support in this study. First, the online support group served to help parents engage with one another and share their experience; it was not the fact that the online group existed, but the fact that the parents in the group joined the group, engaged within the group, and retained the perspective of sharing with other KFS parents as significant for fostering their understanding of a difficult experience. Second, parents discussed ways in which they used the group to procure information and gauge their own situation, which served not only to legitimize the reality of rare disease, but fostered parents to normalize one another's reality. In essence, within the group the difficulties they face as KFS parents are normal, other parents experience the same distress and other parents understand.

Furthermore, some parents drew attention to the Facebook support group when discussing their experience of diagnosis. These parents underscored an additional process according to Walsh (2003), which is establishing a sense of coherence or viewing adversity as a manageable challenge. These parents mentioned the significant impact of membership in the *Klippel-Feil Syndrome Freedom* support group. One parent stated, "The KFS group has been amazing to be a part of and I am thankful for the support systems I have found through this jorney[*sic*]." Another parent stated, "The Facebook support group has also been a life savor." These parents show that the online support helped them gain perspective and find direction in their uncertainty, moving toward a position of manageability in an adverse circumstance.

Overall, the online support group played an important role in supporting parents to make meaning out of their adverse experiences with KFS. In the absence of access to other KFS parents or individuals face-to-face, this online support group provides a home for parents that fosters resiliency and normalizes the reality of parenting a KFS child.

Self of the Researcher

The main researcher in this study acknowledges that her own biases as a parent of two KFS children may have influenced her perspective so she added a third coder to the study (Patton, 1999). Specifically, this researcher retains a strengths based perspective that she finds more conducive to her clinical work and more helpful in her personal KFS experiences. This researcher acknowledges that some of the participants' descriptions mirrored her personal experiences, including emotionally difficult experiences. On more than one occasion the faculty advisor played a pivotal role in guiding the main researcher to more critically evaluate the qualitative research. The third coder affirmed the themes in the research and further elucidated emotional content, which forced additional evaluation by the main researcher and faculty advisor. The main researcher and faculty advisor reevaluated their coding to verify the connection between the key words and themes with respect to the connotation of the overall response of each participant. Ultimately, this researcher and the faculty advisor agreed that the original coding was substantive and adequately represented participant's key words and overall meaning with the third coder's information reflecting the overall difficult context faced by rare disease patients.

Limitations

This study has several limitations. The sample size is small and may not adequately reflect all parents who have a child diagnosed with KFS, especially economically disadvantaged parents who do not have the ability to access social media or who do not have access to some of the resources highlighted in this research. This study may disproportionally reflect persons who display a particular set of characteristics in the treatment and management of KFS, specifically parents who utilize social support, demonstrate agency, and who are active in accessing information and advocating on their child's behalf in the treatment and management of KFS. Furthermore, this research includes information about parents' experiences of KFS diagnosis without including information from the children themselves. Therefore, the perspective of the KFS child is not represented in this research.

Application

The information obtained in this study is useful for mental health professionals, physicians, nurses, and other related professionals working with KFS families and possibly other rare disease families. This study provides insights into the needs of KFS parents in seeking treatment for their children, including barriers and resources that could be used to advocate on behalf of or develop interventions for this population, including interventions or advocacy at the policy level. This study is useful for clinicians in understanding the unique role of online support, particularly with rare disease where there may not be nationally funded, networked, and established advocacy groups or

support resources. The study is useful for clinicians in recommending or searching for a Facebook support group for a rare disease client or family. Furthermore, the study provides evidence of resiliency that is useful for clinicians in assessing family strengths and developing treatment plans.

Future Research

The significance of online support in this study population shows that it would be important to explore variables that are significant to positive and supportive online interaction, including the use of tools to evaluate the effectiveness of online support groups. This is important because the Facebook administrator in the online support group for this study plays an active and encouraging role in furthering the experience of parents in the closed support group. Furthermore, it would be important to understand how particular characteristics of online support groups influence healthcare perceptions, and to further understand the relationship between the two and their contribution to disease treatment and management. Additionally, as the parents in this study demonstrated resilience it would be important to explore resilience in the lived experience of KFS and other rare disease families using qualitative inquiry.

Conclusion

There are several key findings from this research. First, this research elucidates beneficial multisystemic resources for KFS families that highlight the significant impact policies, health providers, communities, and families themselves have in fostering resilience. Multisystemic resources as discussed in this research also highlight the many

contexts that are implicated in rare disease care. In this research, a closed online support group was repeatedly stressed as a beneficial resource for parents. The support group was revered for providing an informative, safe, and open exchange that parents could trust when making decisions or asking questions about their KFS child. Finally, this research clearly delineates areas of uncertainty for KFS parents. In a difficult disease context when uncertainty permeates questions about the course of KFS, parents placed great emphasis on physicians in terms of their experience and their ability to assist in planning treatment. Physicians and the research that informs their practice seem to act as gatekeepers of reduced uncertainty for KFS families, which lead parents in this study to focus on KFS treatment, access and utilization of physicians, and health outcomes. As KFS research knowledge may be slow to evolve, positive physician-patient interactions should be emphasized as an intervention to improve collaboration. Overall, this study provides insight about the experiences of families living with a child diagnosed with KFS.

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APPENDIX A

Letter to Surveys and Social Research Manager of Eurordis

The following message was emailed to Sandra Courbier at sandra.courbier@eurordis.org on November 11, 2016:

Dear Ms. Courbier:

I would like to use the Eurordis Care Survey 2 and 3 to research a population of Klippel Feil Syndrome patients. The surveys will be adapted to the population being studied with some questions presented in entirety with other questions changed or deleted. Qualitative and quantitative questions will be added. As the surveys in *The Voice of 12,000 Patients* have been copyrighted by Eurordis, I would like to secure permission from Eurordis to use the surveys. I need to provide information to the Institutional Review Board at Texas Woman's University in Denton, Texas in the United States of America that I have been granted permission to use the copyrighted surveys. The printed surveys are on pages 314-321 of *The Voice of 12,000 Patients.* In addition, I would also use the Eurordis Care 3 Survey questions 7-11 that were missing from the book but provided to me via email from you, Sandra Courbier, Surveys and Social Research Manager for Eurordis. My research should be completed during 2017. If you cannot grant permission to use copyrighted material, please provide the appropriate contact. I appreciate your assistance and look forward to hearing from you.

Respectfully,

Shirley Shropshire
Family Therapy Graduate Student
Texas Woman's University

APPENDIX B

Response from Surveys and Social Research Manager of Eurordis

Dear Shirley,

Thank you for these documents, we grant full copyright permission for the use of EURORDIS previous work,

I wish you all the best for the continuation of your project,

Kind regards,

Sandra.

APPENDIX C

Permission Request to Klippel Feil Freedom Support Group Administrator

Sharon Nissley is the creator and administrator of Klippel Feil Freedom. The following communication was posted through Facebook messenger on December 2, 2016:

Dear Ms. Nissley,

I am a member of the Klippel Feil Freedom parent group that you created. I am also a graduate student in the Masters of Family Therapy Program at Texas Woman's University. I am beginning a research study about the experience of diagnosis in KFS patients and families, barriers faced by KFS patients and families in health service use, and the role of online support group in KFS disease treatment and management. I am interested in getting your permission to post a link to the survey in the parent support group, and possibly the adult group. Do you have any information about the geographical location of the members or any additional information about the diversity of the group? I look forward to hearing from you and I am happy to answer any questions you might have. My email address is sshropshire@twu.edu.

Sincerely,

Shirley Shropshire

APPENDIX D

Permission Request Response from Klippel Feil Syndrome Freedom Administrator

April 20, 2017

To Whom It may Concern,

Klippel-Feil Syndrome Freedom will gladly allow your survey to be posted in our communities three peer support groups for your research study about the about the experience of diagnosis in Klippel-Feil syndrome patients and families, barriers faced by Klippel-Feil syndrome patients and families in health service use, and the role of online support group in KFS disease treatment and management. We understand you are providing the survey to collect data from 30 participants as a graduate student in the Masters of Family Therapy Program at Texas Woman's University.

Sincerely,

Sharon Rose Nissley Founder of Klippel-Feil Syndrome Freedom

APPENDIX E

KFS Support Group Study Recruitment Flyer

Dear Klippel Feil Syndrome Patient or Parent,

Are you interested in participating in research about Klippel Feil Syndrome? Shirley Shropshire, a Master of Science in Family Therapy student at Texas Woman's University is conducting a research study focused on learning more about the experience of diagnosis, health service use, and online support group use for Klippel Feil Syndrome patients. Participation in the study requires the completion of an online survey that will take between 30 to 60 minutes to complete. Participants should be at least 18 years of age, read and write English at a 3rd grade level, have access to the internet, and reside within the United States of America. Participants should be an adult with Klippel Feil Syndrome or parent of a child with a medical diagnosis of Klippel Feil Syndrome. Participants will be asked to disclose health related information. Participants may experience emotional distress while participating in the study, and participants will be provided with information to contact local mental health service providers. There is a risk of loss of confidentiality with the use of online surveys. Questions about the study may be directed to Shirley Shropshire at sshropshire@twu.edu. Participants may click on the link provided to learn more about the study. There is a potential risk of loss of confidentiality in all email, downloading, electronic meetings, and internet transactions. Participation in the study is voluntary and participants may withdraw from the study at any time without penalty. Participants may click on the link provided to access the study.

To ask questions about the study you may contact the following:

Investigator: Shirley Shropshire, BS…… sshropshire@twu.edu, (469)-383-0874

Advisor: Linda Ladd, PhD, PsyD……lladd@mail.twu.edu, (940)-898-2694

APPENDIX F

Consent to Participate in Research

TEXAS WOMAN'S UNIVERSITY

CONSENT TO PARTICIPATE IN RESEARCH

KLIPPEL-FEIL SYNDROME: A STUDY OF PARENT AND CAREGIVERS' EXPERIENCES OF DIAGNOSIS, HEALTH SERVICE USE, AND ONLINE SUPPORT IN A RARE DISEASE POPULATION

Investigator: Shirley Shropshire, BS…… sshropshire@twu.edu, (469)383-0874

Advisor: Linda Ladd, PhD, PsyD……lladd@mail.twu.edu, (940)898-2694

EXPLANATION AND PURPOSE OF THE RESEARCH

The purpose of this study is to describe the experience of diagnosis, health service use, and online support group use for Klippel Feil Syndrome (KFS) parents and caregivers. This study is being conducted by Shirley Shropshire to complete requirements for her master's thesis in Family Therapy at Texas Woman's University. As a participant in this study, you will be contributing to research about the impact of living with KFS, a rare disease. Specifically, you will be helping to establish social and healthcare research about KFS.

Questions in this survey have been adapted from the EurordisCare 2 and EurordisCare 3 Surveys developed by EURORDIS-Rare Diseases Europe, an alliance of rare disease patient organizations that works to improve the lives of people living with a

rare disease in Europe. The EurordisCare 2 and 3 Surveys represent a tremendous effort to identify and address the needs of rare disease patients in Europe. The researchers are appreciative of the knowledge gained in the Eurordis Surveys and their influence in developing the research survey for this study.

The researchers appreciate the assistance of Klippel-Feil Syndrome Freedom as a resource for gathering patient data. KFS Freedom is a patient-run organization that empowers and unites patients and their families through peer support, education, research and advocacy for a lifetime of improved health care.

You may begin completing the survey when you are ready and have read this document. Your agreement to participate in this study, on a computer at a site of your choosing, will be assumed if you click the 'Continue' button below. Once you have clicked to continue the survey, you have given your consent to participate in this survey. The survey may take between 30 to 60 minutes to complete.

Participants will be asked to provide information regarding their child's KFS diagnosis including ages of diagnosis, misdiagnosis, medical tests, consulted physicians, use of mental health support, and expenses related to medical treatment. Participants will also be asked to answer multiple choice questions and provide written descriptions about their experience of diagnosis, health service use, and online support group use. Participants are **not required** to provide the name of the child referenced in the survey.

Participation in this study is voluntary. Participants may stop the survey at any time and withdraw from the study at any time without penalty. Participants will not be able to save or return to the survey. Please review the following requirements for study participation:

- Participants must be at least 18 years of age or older, must be able to read and write English at a 3rd grade level, and have internet access.
- Participants should be a parent or caregiver of a child with a medical diagnosis of Klippel Feil Syndrome.
- Participation in the study is limited to U.S. residents only.
- Participation in this study is anonymous.
- Participants will not be compensated for participation.
- Participants should complete the survey only once.

If you do not meet the criteria described for this study, please do not complete the survey.

POTENTIAL RISKS

- There is potential risk of loss of confidentiality in all e-mail, downloading, internet transactions, and use of online surveys. PsychData always provides end-to-end encryption of all account data, and all participant survey data is encrypted for security. Please complete the survey in a private setting to reduce the risk of loss of confidentiality.

- You may experience emotional distress or discomfort while answering questions about your personal experiences related to Klippel Feil Syndrome. Please complete the survey in a private setting where you can accommodate your personal needs for comfort or emotional distress.
- You will be provided an online link to access local mental health service providers at the end of the study. If you contact a mental health service provider, you will do so at your own expense.
- You may experience fatigue while completing this survey. Please allow yourself to take needed breaks or gather information before beginning the survey to expedite completion. Remember you will not be allowed to save and return to the survey at a later time.
- The principal investigator is also a parent member of Klippel-Feil Syndrome Freedom. Participating or not participating in the study will not impact a person's member relationship or status in the Klippel-Feil Syndrome Freedom Support Group.

The researchers will try to prevent any problem that could happen because of this research. You should let the researchers know at once if there is a problem and they will help you. However, TWU does not provide medical services or financial assistance for injuries that might happen because you are taking part in this research.

Participants may provide an email address via a link at the end of the study to obtain a summary of the information learned from this study.

Resources for participants who need to talk to a professional about their discomfort:

- American Association of Marriage and Family Therapy – Therapist Locator: http://www.therapistlocator.net/imis15/therapistlocator/
- Christian Association for Psychological Studies (after clicking the link select the 'online directory'): https://caps.net/
- You may also find a therapist near you via Psychology Today (after clicking the link select 'find a therapist') https://www.psychologytoday.com

APPENDIX G

KFS Research Survey

KLIPPEL-FEIL SYNDROME: A STUDY OF PARENT AND CAREGIVERS' EXPERIENCES OF DIAGNOSIS, HEALTH SERVICE USE, AND ONLINE SUPPORT IN A RARE DISEASE POPULATION

Please note that a majority of questions in this survey have been adapted from the EurordisCare 2 and EurordisCare 3 Surveys developed by the Eurordis Organization. Klippel Feil Syndrome Freedom has been a resource in collecting data in this survey.

Please select one answer for the following questions. KFS is Klippel-Feil Syndrome.

1. Please identify your relationship to the person diagnosed with KFS.

    a) I am the parent of a child with KFS (child is biological or adopted)
    b) I am a relative/caregiver of an adult or child with KFS
    c) I am the parent of a child with KFS who is also an adult with KFS

2. Patient's age at last birthday:____

3. Patient's gender:

    a) female
    b) male
    c) Other (please specify):_____

The following two questions ask about your ability to access insurance that covers KFS:

4. When your child was diagnosed with KFS, did your child have health insurance that covers KFS?

a) yes

b) no

5. Does your child diagnosed with KFS currently have health insurance that covers KFS?

a) yes

b) no

Please think back to the beginning of your child's or your own experience with KFS and how KFS first presented in terms of symptoms. You may provide up to three symptoms for the following question.

6. What were the first manifestations or symptoms of the disease?

a) symptom 1:_____

b) symptom 2:_____

c) symptom 3:_____

7. How many doctors did you consult between the first symptom and the final diagnosis?

a) 1 to 2

b) 3 to 5

c) 6 to 10

d) 11 to 20

e) more than 20

f) Other (please specify):_____

8. Considering the time symptoms first began until final diagnosis of KFS, what type of examinations were carried out during this period? (Multiple answers may be selected)

a) none
b) biological examinations (blood test, urine test, biopsy)
c) genetic testing
d) radiological or imaging examinations (ultrasound, x-ray, MRI)
e) functional explorations (respiratory, muscular, electroencephalogram)
f) Other (please specify):_____

Please consider other diagnoses your child was given in relation to KFS symptoms. For the following questions, please list up to three other diagnoses you were given before that of KFS and provide the patient's age and the type of physician who diagnosed KFS.

9. Diagnosis 1

Diagnosis:_____

Patient's age at diagnosis:_____

Specify the type of physician who made the diagnosis:_____

10. Diagnosis 2

Diagnosis:_____

Patient's age at diagnosis:_____

Specify the type of physician who made the diagnosis:_____

11. Diagnosis 3

Diagnosis:_____

Patient's age at diagnosis:_____

Specify the type of physician who made the diagnosis:_____

12. Following these diagnoses, were treatments performed?

a) no
b) yes, medicinal treatment
c) yes, psychological treatment
d) yes, surgical treatment
e) no, the treating physician requested test(s) that the patient or patient's parent chose not to complete
f) Other (please specify):_____

13. What were the results of the delay in diagnosis? (Multiple answers may be selected)

a) none
b) physical consequences (for example, pain)
c) birth of other children with KFS
d) inability to consider genetic implications in family planning
e) behavioral health consequences (depression or other mental health concerns)
f) family behavior was not adapted to functioning with KFS
g) loss of confidence in healthcare

h) Other (please specify):_____

14. What was the age of the patient when the possibility of KFS was first raised?_____

15. Who raised the possibility of KFS?

a) a physician

b) a health professional other than a physician

c) a rare disease patient

d) an internet search

e) a relative

f) Other (please specify):_____

16. What was the age of the patient when the final diagnosis of KFS was made?_____

17. Which type of doctor made the diagnosis?

a) primary care physician

b) geneticist

c) orthopedist or pediatric orthopedist

d) Other (please specify):_____

18. On which type of data was the diagnosis based?

a) clinical (examination of the patient and symptoms)

b) radiological or imaging (MRI, x-ray, or other imaging)

c) Other (please specify):_____

19. In which type of facility was the diagnosis carried out?

a) private practice

b) specialized care center

c) hospital consultation

d) Other (please specify):_____

20. How did you find the facility?

a) recommended by a physician

b) recommended by a health professional other than a physician

c) recommended by a patient

d) internet, website

e) media, press

f) Other (please specify):_____

21. In relation to your home at the time, where was the facility located?

a) in the same city

b) in the same region

c) in another region of the same state

d) in another state

e) in another country

22. Did you seek a second opinion to confirm the diagnosis?

a) yes

b) no

23. Did knowledge of the diagnosis lead you to change your living arrangements by moving from one place to another?

a) no

b) yes, to get closer to a physician or treatment facility

c) yes, to get closer to a relative or friend for physical support or emotional support

d) yes, to move to a more adapted house or living quarters

e) yes, for educational or other opportunities for yourself or a KFS patient unrelated to medical treatment.

f) Other (please specify):_____

24. Which of the following have been the most helpful to you or your family in living with KFS? (Multiple answers may be selected)

a) family discussion/ problem solving

b) family emotional expression/ mutual support

c) religion/faith/ spirituality/ positive outlook

d) extended family/ friends

e) physicians/ nurses/ other health professionals

f) income

g) health insurance

h) psychologist/ counselor

i) family therapist

j) work schedule

k) geographical location

l) integrated healthcare facilities

m) support group

n) Other (please explain):_____

25. What else would you like to say about your family's experience of diagnosis?

26. Have you ever met another person with KFS face-to-face?

a) no

b) yes

c) Please explain what has been meaningful or helpful about meeting another KFS individual._____

27. Please, describe your reasons for using an online KFS support group.

The following questions ask you to evaluate types of physicians or health services, healthcare costs, and travel to see medical professionals or seek treatment that may be used by KFS patients. Please respond to the questions by considering your interactions with these physicians, health services, healthcare costs, and travel over the last twelve months. It is possible to select more than one answer for each question or if none of the answers apply then skip the answers and move to the next question in the series.

28. Which of the following physicians or health services did you need but were UNABLE to access over the past 12 months?

a) orthopedist

b) neurologist

c) audiologist

d) nephrologist

e) pain control specialist

f) surgical operation

g) mental health provider

h) family therapist

29. Which of the following physicians or health services were the EASIEST to access over the past 12 months?

a) orthopedist

b) neurologist

c) audiologist

d) nephrologist

e) pain control specialist

f) surgical operation

g) mental health provider

h) family therapist

30. Which of the following physicians or health services were DIFFICULT to access over the past 12 months?

a) orthopedist

b) neurologist

c) audiologist

d) nephrologist

e) pain control specialist

f) surgical operation

g) mental health provider

h) family therapist

31. Which of the following physicians or health services incurred the LEAST personal cost over the past 12 months?

a) orthopedist

b) neurologist

c) audiologist

d) nephrologist

e) pain control specialist

f) surgical operation

g) mental health provider

h) family therapist

32. Which of the following physicians or health services incurred the MOST personal cost over the past 12 months?

a) orthopedist

b) neurologist

c) audiologist

d) nephrologist

e) pain control specialist

f) surgical operation

g) mental health provider

h) family therapist

33. Which of the following physicians or health services required the LEAST amount of travel over the past 12 months?

a) orthopedist

b) neurologist

c) audiologist

d) nephrologist

e) pain control specialist

f) surgical operation

g) mental health provider

h) family therapist

34. Which of the following physicians or health services required the MOST amount of travel over the past 12 months?

a) orthopedist

b) neurologist

c) audiologist

d) nephrologist

e) pain control specialist

f) surgical operation

g) mental health provider

h) family therapist

35. Which of the following physicians or health services required the LEAST amount of wait time to obtain an appointment over the past 12 months?

a) orthopedist

b) neurologist

c) audiologist

d) nephrologist

e) pain control specialist

f) surgical operation

g) mental health provider

h) family therapist

36. Which of the following physicians or health services required the MOST wait time to obtain an appointment over the past 12 months?

a) orthopedist

b) neurologist

c) audiologist

d) nephrologist

e) pain control specialist

f) surgical operation

g) mental health provider

h) family therapist

37. Please consider how the following answer selections have impacted your family in managing KFS. Overall, please rank the following in order for having the most to least impact your family in managing KFS. In other words item 1 will have the most impact and item 5 will have the least.

___medical expenses and health insurance

___travel time to physicians or health services

___finding physicians with KFS experience

___managing appointments, physicians, and care

___time involved in scheduling and waiting for an appointment

38. What else would you like to say about utilizing or interacting with physicians or health services?

39. What else would you like to say about your ability to access physicians or health services or get the services you need?

40. Please consider all of your experience with KFS, including information that has not been asked about. If you could give another parent of a child with KFS advice about any

aspect of managing the disorder (physicians, insurance or even their own decision-making) please share any advice not previously discussed in this survey in the space below.

APPENCIX H

EurordisCare 2 Survey

EurordisCare 2
Survey of the delay in diagnosis for 10 rare diseases in Europe

Dear friends, we are contacting you as a member of a patient organisation collaborating in this survey.
As you know, Rare Diseases are still poorly known both by the general public and by most health professionals. This lack of information often leads to late diagnoses which delay the beginning of adapted treatments and can be responsible for the disease's progression or to severe consequences. While rare disease patients often face this delay in diagnosis, it remains badly documented and its consequences are insufficiently taken into account by the health authorities.
In collaboration with ***75*** European associations including yours, EURORDIS * is undertaking this survey to study the delay in diagnosis for ***10*** rare diseases (Crohn's disease, Cystic fibrosis, Duchenne muscular dystrophy, tuberous sclerosis, Ehlers-Danlos syndrome, Marfan syndrome, Prader Willi syndrome, X fragile syndrome, retinitis pigmentosa, Williams syndrome). This survey, named EurordisCare 2, will cover all European countries (as far as possible!!).
This study's objective is to identify the main causes of delay in diagnosis and to find solutions to reduce this delay by appropriate measures such as: medical training, raising public awareness, systematic screening, etc. Delay in diagnosis can vary greatly depending on the disease, and on the country, but also on individual factors: each patient has his own history. **For this reason, this questionnaire is addressed to the only information holders: the patients and families concerned. Each answer is significant. The quality and the repercussions of this study depend on the number of answers and on their widespread origin. If you have any problems answering this questionnaire, please contact us at eurordiscare@eurordis.org**
To save time, your members should return the completed questionnaire directly to Eurordis using the pre-paid envelope herewith. Each participating association will of course have access to the anonymous data concerning its own members.
The number of answers received per country and per disease, will be regularly updated and available at www.eurordis.org in the section "EurordisCare 2".

We hope to be able to present the synthesis of this investigation in the summer of 2004 and would therefore thank you in advance for answering very quickly, as the questionnaires received after June 15th may not be taken into account.
** Eurordis is a European alliance of rare disease patient associations which brings together more than 200 associations in 17 countries. A prior survey, "EurordisCare 1", of access to care for six rare diseases. 50 patient organisations participated - the results can be consulted on our site* www.eurordis.org

1

• Questionnaire(s)** completed by

□ the patient	□ a relative	□ another member of the association	□ other: _____

*** If several people suffer from the disease in your family, please complete a questionnaire per person and return them to us in the same envelope. You can photocopy this form or ask your association or eurordiscare@eurordis.org for extra copies*

2

•Patient's date of birth: . . /. . /. . . . (day/month/year) • Sex: □ Male □ Female

•When the disease first manifested itself, what was the patient's professional category or that of the patient's parents if symptoms occurred during childhood?

	□ Student	□ Teacher
□ Middle or senior management	□ Unemployed	□ Farmer
□ Bluecollar worker	□ Retired	□ Employee
□ Craftsman, tradesman	□ Liberal professions	□ Other

• When the disease first manifested itself, where did the patient live, or his parents if symptoms occurred during childhood?

□ country or village	□ town of 1000 to 10 000 inhabitants	□ town of 10 000 to 50 000 inhabitants
□ town of 50 000 to 100 000 inh.	□ town of 100 000 to 500 000 inhabitants	□ city of more than 500 000 inhabitants

• Was the diagnosis already known at birth, or within the first 3 months ? **3**

□ No	Yes, specify the circumstances		
	□ Other cases in the family	□ Disorders during the pregnancy or at the birth	□ Systematic neonatal screening
	If yes go directly to: **7.** (bottom of page 3)		

• What were the first manifestations of the disease? **4**

Nature of the symptoms	When did these symptoms start?	How many time did these symptoms occur before the final diagnosis?			
	age: ..... or date: . . /. . . . (month/year)	□ 1 to 5 times	□ 6 to 20 times	□ more than 20 times	□ persistent disorders
	age: ..... or date: . . /. . . . (month/year)	□ 1 to 5 times	□ 6 to 20 times	□ more than 20 times	□ persistent disorders
	age: ..... or date: . . /. . . . (month/year)	□ 1 to 5 times	□ 6 to 20 times	□ more than 20 times	□ persistent disorders

• How many doctors did you consult between the first manifestations and the final diagnosis?

□ 1 to 2 □ 3 to 5 □ 6 to 10 □ 11 to 20 □ more than 20: _____

• Which types of examination were carried out during this period? ***(several answers possible)***

□ none	□ biological examinations (blood test, urine test, CSF test, biopsy)	□ genetic testing
□ radiological examinations, (ultrasound, NMR, scanner, other imagery…)	□ functional explorations (respiratory, muscular, electroencephalogram)	□ others: _____

• In view of these disorders, were you given other diagnosis before that of your disease?

□ No □ Yes, specify:

Diagnosis:	When?	By whom?	
1	age: ..... or date: . . /. . . . (month/year)	□ a general practitioner	□ a specialist (specify): _____
2	age: ..... or date: . . /. . . . (month/year)	□ a general practitioner	□ a specialist (specify): _____
3	age: ..... or date: . . /. . . . (month/year)	□ a general practitioner	□ a specialist (specify): _____

• Following these diagnosis, were treatments undertaken? ***(several answers possible)***

□ No	Yes, specify:	□ medicinal	□ surgical
		□ psychological	□ others.....

• What were the consequences of the delay in diagnosis? ***(several answers possible)***

□ none	□ physical consequences	□ birth of other children suffering from the disease
	□ psychiatric consequences	□ family behaviour not adapted
	□ intellectual consequences	□ loss of confidence in medicine
	□ death	□ other: .....

5

• When was the possibility of a rare disease first raised?

Age of the patient: ..... or Date: . . /. . . . (month/year)

• Who raised this possibility?

□ a physician	□ another health professional	□ the media/ press	□ Internet
□ a teacher	□ a rare disease patient	□ a close relative	□ other.....

6

• When was the final diagnosis carried out?

Age of the patient: ..... or Date . . /. . . . (month/year)

• By whom was the diagnosis given?

□ a general practitioner | □ a specialist specify the speciality _____

• On which type of data was the diagnosis based? ***(several answers possible)***

□ clinical (examination of the patient, symptoms, evolution...)	□ biological (blood or urine test, biopsy...)	□ functional (respiratory, muscular, EEG..)
□ radiological (scanner, ultrasound, scintigraphy, NMR...)	□ genetics	□ other: _____

• In which type of facility was the diagnosis carried out?

□ private practice	□ hospital consultation
□ specialised centre	□ other: .....

• How did you find this facility?

□ recommended by a physician	□ by another health professional	□ media, press
□ recommended by a patient	□ Internet, Web site	□ other: _____

• In relation to your home at that time, where was this facility located?

□ in the same city	□ in the same region	□ in another region	□ in another country

• Did you seek a second opinion to confirm this diagnosis?

□ No | □ Yes, when ?

• Age of the patient: ..... or Date: . . /. . . . (month/year)

• Who provided this confir matory diagnosis?

□ a general practitioner □ a specialist specify: _____

• Where was this confirmatory diagnosis carried out?

□ private practice □ hospital consultation

□ specialised centre □ other: _____

• Did seeking diagnosis require any personal expenditure?

□ no	□ low	□ moderate	□ high	□ very high

• Do you think that the delay in diagnosis depends on the level of personal expenditure?

□ not at all	□ slightly	□ partially	□ primarily

7

• Did knowledge of this diagnosis lead you to move?

□ No | □ Yes

□ within the same region	□ to another area	□ to another country

8

•Who announced the disease diagnosis to you?

	□ a general practitioner	□ a specialist
□ an analysis laboratory	□ a geneticist	□ other:

• How?

□ orally in consultation	□orally, elsewhere (corridor…)	□ by phone
□ in writing with explanations	□ in writing without explanations	□ other:

• Was the announcement of the diagnosis accompanied by psychological support?

□ No

Yes, specify by whom:

□ by a general practitioner	□ by a specialist	□ by a psychologist
□ by an association member	□ other:	

• Should this support be systematically offered? □ No □ Yes

• Did you then receive complete information about the disease? ***(several answers possible)***

□ No

Yes, specify by whom:

□ by a general practitioner	□ by a specialist	□ by a geneticist
□ by an association member	□ other:	

• Should this information be systematically provided at that time? □ No □ Yes

• Globally, the conditions in which the diagnosis was announced of the diagnosis seemed:

□ well-adapted	□ acceptable	□ poor	□ unacceptable

9

• Was the genetic nature of the disease identified?

□ No

Yes

- □ not inherited (new mutation or isolated case)
- □ transmitted with possibility of other carriers in the family (recessive gene)
- □ transmitted with possibility of other people suffering in the family (dominant gene)

• When? Age of the patient: ..... or Date: . . /. . . . (month/year)

• Following the diagnosis of the disease did you obtain genetic advice?

□ No □ Yes

• Once diagnosed did you contact other members of your family to inform them of the genetic nature of the disease? ***(several answers possible)***

□ No

Yes, specify which:

□ father, mother	□ brothers, sisters
□ grandparents, grandchildren	□ uncles, aunts, cousins
□ more distant relative	

•Was this step suggested by doctors or health professionals?

□ No

Yes, specify on which occasion:

□ during the diagnosis □ during genetic counselling □ later

• Did this step lead to diagnoses among other members of your family?

□ No

Yes, specify	Family link:
□ Suffering from the disease and already having symptoms	
□ Suffering from the disease while not yet having symptoms	
□ Not suffering from the disease but liable to transmit it	

Thank you for your collaboration…

APPENDIX I

EurordisCare 3 Survey

Logo

Eurordis survey on patients' experience and expectations concerning access to health services in Europe.

EURORDIS
Rare Diseases Europe

Dear friends,

We are writing to you, patient or patient's relative, who is member of ***<name of the organisation>*** which is collaborating with Eurordis on this survey.

Access to health services is part of the difficulties associated with rare diseases that are encountered daily by patients and their families. Access to health services faces various obstacles: identification of skilled professionals, access to structures that are sometimes far away, acceptance of financial liability or reimbursement of the medical services and journey, etc. The difficulties encountered can vary a lot according to the disease, the country, and individual factors: each patient has his/her own history.

We are leading this survey in order to assess the current situation and define expectations of patients affected by Chromosome 11q disorders and 15 other rare diseases in Europe (Alternating hemiplegia, Aniridia, Ataxias, Cystic fibrosis, Ehlers-Danlos syndrome, Epidermolysis bullosa, Fragile X syndrome, Huntington disease, Marfan syndrome, Myasthenia, Osteogenesis imperfecta, Prader-Willi syndrome, Pulmonary arterial hypertension, Tuberous sclerosis, and Williams syndrome). Our aim is to make the patient voice heard, while several European countries are leading a reflection on the reorganisation of the offer for care for rare diseases. The quality of this survey and the attention that it will get from health policy makers depends on the number of responses we receive. **Patients' opinion is the priority.**

Questions concerning your personal experience were defined in collaboration with organisations of every disease to better adapt them to specific situations. Questions concerning your expectations, and those that help to identify the origin of the disparities of access to health services due to individual situations are common to all diseases. **It is essential to answer all questions.** If a section is not relevant (sections 1 to 8, 11 and 12), please select "no" in the first line of the section, before moving on to the next one. Unless mentioned otherwise, each question requires a single answer.

The distribution of the questionnaire by your organisations, who have exclusive access to your contact details, allows us to specifically survey the patients of the diseases studied. Once completed, this anonymous questionnaire can be returned directly to Eurordis (using the included prepaid envelope), which will ensure the complete confidentiality of the information and responses. Eurordis will complete the analysis of this data and commits not to share your personal data with anyone. Every organisation will receive the statistical results of the analysis corresponding to the responses of their members.

The first presentation of preliminary results is planned for 13-14th July 2007 during the "European Workshop on Centres of Expertise" in Prague.

(contact : mherasse@eurordis.org)

Thank you for replying quickly to ensure all responses are taken into account in the analysis (before … 2007).

1 Have you needed to consult a **cardiologist** over the last 12 months ? ☐ YES ☐ NO → go to **2**

Did you have access to these consultations ? ☐ YES

- Access to these consultations was :
  ☐ very easy ☐ easy ☐ difficult ☐ very difficult
- Overall, the number of consultations was :
  ☐ well adapted ☐ sufficient ☐ insufficient ☐ very insufficient ☐ no opinion
- The personal cost incurred was :
  ☐ nil ☐ partial ☐ total specify £ ☐ / consultation
  You found this amount : ☐ acceptable ☐ excessive
- Most of the consultations took place :
  ☐ at home ☐ private practice ☐ hospital ☐ clinic ☐ specialised centre for your disease
  specify where : ☐ same region ☐ other region ☐ other country ☐ miles from home
  You found this distance : ☐ acceptable ☐ excessive
  journey completed : ☐ alone ☐ with a relative / friend ☐ with professional assistance or transport
- You found the time to obtain the first consultation :
  ☐ very short ☐ short ☐ long ☐ very long specify how long ☐ month
- These consultations responded to your expectations :
  ☐ fully ☐ partially ☐ poorly ☐ not at all

☐ NO

Why ? (several responses possible)

- Specialist could not be found or unavailable … ☐ yes ☐ no
- Lack of referral … ☐ yes ☐ no
- Excessive waiting time … ☐ yes ☐ no
  specify ☐ month
- Personal cost incurred too high … ☐ yes ☐ no
  specify £ ☐ / consultation
- Location of the consultations too far … ☐ yes ☐ no
  specify ☐ miles from home
  specify where ☐ same region ☐ another region ☐ another country
- Excessive time of journey … ☐ yes ☐ no
- Nobody was available to accompany you … ☐ yes ☐ no
- Cost of journey too high … ☐ yes ☐ no
- Lack of suitable transport … ☐ yes ☐ no
- Travelling too difficult (pain, fatigue, injuries) ☐ yes ☐ no

2 Have you needed to consult a **neurologist** over the last 12 months ? ☐ YES ☐ NO → go to **3**

Did you have access to these consultations ? ☐ YES

- Access to these consultations was :
  ☐ very easy ☐ easy ☐ difficult ☐ very difficult
- Overall, the number of consultations was :
  ☐ well adapted ☐ sufficient ☐ insufficient ☐ very insufficient ☐ no opinion
- The personal cost incurred was :
  ☐ nil ☐ partial ☐ total specify £ ☐ / consultation
  You found this amount : ☐ acceptable ☐ excessive
- Most of the consultations took place :
  ☐ at home ☐ private practice ☐ hospital ☐ clinic ☐ specialised centre for your disease
  specify where : ☐ same region ☐ other region ☐ other country ☐ miles from home
  You found this distance : ☐ acceptable ☐ excessive
  journey completed : ☐ alone ☐ with a relative / friend ☐ with professional assistance or transport
- You found the time to obtain the first consultation :
  ☐ very short ☐ short ☐ long ☐ very long specify how long ☐ month
- These consultations responded to your expectations :
  ☐ fully ☐ partially ☐ poorly ☐ not at all

☐ NO

Why ? (several responses possible)

- Specialist could not be found or unavailable … ☐ yes ☐ no
- Lack of referral … ☐ yes ☐ no
- Excessive waiting time … ☐ yes ☐ no
  specify ☐ month
- Personal cost incurred too high … ☐ yes ☐ no
  specify £ ☐ / consultation
- Location of the consultations too far … ☐ yes ☐ no
  specify ☐ miles from home
  specify where ☐ same region ☐ another region ☐ another country
- Excessive time of journey … ☐ yes ☐ no
- Nobody was available to accompany you … ☐ yes ☐ no
- Cost of journey too high … ☐ yes ☐ no
- Lack of suitable transport … ☐ yes ☐ no
- Travelling too difficult (pain, fatigue, injuries) ☐ yes ☐ no

3 Have you needed to consult an **ear, nose and throat medicine specialist** over the last 12 months ? ☐ YES ☐ NO → go to 4

Did you have access to these consultations ? ☐ YES

- Access to these consultations was :
  ☐ very easy ☐ easy ☐ difficult ☐ very difficult
- Overall, the number of consultations was :
  ☐ well adapted ☐ sufficient ☐ insufficient ☐ very insufficient ☐ no opinion
- The personal cost incurred was :
  ☐ nil ☐ partial ☐ total specify £ ☐ / consultation
  You found this amount : ☐ acceptable ☐ excessive
- Most of the consultations took place :
  ☐ at home ☐ private practice ☐ hospital ☐ clinic ☐ specialised centre for your disease
- specify where :
  ☐ same region ☐ other region ☐ other country ☐ miles from home
  You found this distance : ☐ acceptable ☐ excessive
- journey completed : ☐ alone ☐ with a relative / friend ☐ with professional assistance or transport
- You found the time to obtain the first consultation :
  ☐ very short ☐ short ☐ long ☐ very long specify how long ☐ month
- These consultations responded to your expectations :
  ☐ fully ☐ partially ☐ poorly ☐ not at all

☐ NO

Why ? (several responses possible)

- Specialist could not be found or unavailable ... ☐ yes ☐ no
- Lack of referral ... ☐ yes ☐ no
- Excessive waiting time ... ☐ yes ☐ no
  specify ☐ month
- Personal cost incurred too high ... ☐ yes ☐ no
  specify £ ☐ / consultation
- Location of the consultations too far ... ☐ yes ☐ no
  specify ☐ miles from home
  specify where ☐ same region ☐ another region ☐ another country
- Excessive time of journey ... ☐ yes ☐ no
- Nobody was available to accompany you ... ☐ yes ☐ no
- Cost of journey too high ... ☐ yes ☐ no
- Lack of suitable transport ... ☐ yes ☐ no
- Travelling too difficult (pain, fatigue, injuries) ☐ yes ☐ no

4 Have you needed to consult a **dietician** or **nutritionist** over the last 12 months ? ☐ YES ☐ NO → go to 5

Did you have access to these consultations ? ☐ YES

- Access to these consultations was :
  ☐ very easy ☐ easy ☐ difficult ☐ very difficult
- Overall, the number of consultations was :
  ☐ well adapted ☐ sufficient ☐ insufficient ☐ very insufficient ☐ no opinion
- The personal cost incurred was :
  ☐ nil ☐ partial ☐ total specify £ ☐ / consultation
  You found this amount : ☐ acceptable ☐ excessive
- Most of the consultations took place :
  ☐ at home ☐ private practice ☐ hospital ☐ clinic ☐ specialised centre for your disease
- specify where :
  ☐ same region ☐ other region ☐ other country ☐ miles from home
  You found this distance : ☐ acceptable ☐ excessive
- journey completed : ☐ alone ☐ with a relative / friend ☐ with professional assistance or transport
- You found the time to obtain the first consultation :
  ☐ very short ☐ short ☐ long ☐ very long specify how long ☐ month
- These consultations responded to your expectations :
  ☐ fully ☐ partially ☐ poorly ☐ not at all

☐ NO

Why ? (several responses possible)

- Specialist could not be found or unavailable ... ☐ yes ☐ no
- Lack of referral ... ☐ yes ☐ no
- Excessive waiting time ... ☐ yes ☐ no
  specify ☐ month
- Personal cost incurred too high ... ☐ yes ☐ no
  specify £ ☐ / consultation
- Location of the consultations too far ... ☐ yes ☐ no
  specify ☐ miles from home
  specify where ☐ same region ☐ another region ☐ another country
- Excessive time of journey ... ☐ yes ☐ no
- Nobody was available to accompany you ... ☐ yes ☐ no
- Cost of journey too high ... ☐ yes ☐ no
- Lack of suitable transport ... ☐ yes ☐ no
- Travelling too difficult (pain, fatigue, injuries) ☐ yes ☐ no

5 Have you needed **electroencephalograms (EEG)** over the last 12 months ? ☐ YES ☐ NO → go to 6

Did you have access to these tests ? ☐ YES

- Access to these tests was :
  ☐ very easy ☐ easy ☐ difficult ☐ very difficult
- Overall, the number of tests was :
  ☐ well adapted ☐ sufficient ☐ insufficient ☐ very insufficient ☐ no opinion
- The personal cost incurred was :
  ☐ nil ☐ partial ☐ total specify £ ☐ / examination
  You found this amount : ☐ acceptable ☐ excessive
- Most of the tests took place :
  ☐ at home ☐ private practice ☐ hospital ☐ clinic ☐ specialised centre for your disease
- specify where :
  ☐ same region ☐ other region ☐ other country ☐ miles from home
  You found this distance : ☐ acceptable ☐ excessive
- journey completed : ☐ alone ☐ with a relative / friend ☐ with professional assistance or transport
- You found the time to obtain the first test :
  ☐ very short ☐ short ☐ long ☐ very long specify how long ☐ month
- These tests responded to your expectations :
  ☐ fully ☐ partially ☐ poorly ☐ not at all

☐ NO

Why ? (several responses possible)

- Type of tests could not be found ... ☐ yes ☐ no
- Lack of prescription ... ☐ yes ☐ no
- Excessive waiting time ... ☐ yes ☐ no
  specify ☐ month
- Personal cost incurred too high ... ☐ yes ☐ no
  specify £ ☐ / examination
- Location of the tests too far ... ☐ yes ☐ no
  specify ☐ miles from home
  specify where ☐ same region ☐ another region ☐ another country
- Excessive time of journey ... ☐ yes ☐ no
- Nobody was available to accompany you ... ☐ yes ☐ no
- Cost of journey too high ... ☐ yes ☐ no
- Lack of suitable transport ... ☐ yes ☐ no
- Travelling too difficult (pain, fatigue, injuries) ☐ yes ☐ no

6 Have you needed **psychotherapy** or **psychomotility therapy** over the last 12 months ? ☐ YES ☐ NO → go to 7

Did you have access to this type of care ? ☐ YES

- Access to this type of care was :
  ☐ very easy ☐ easy ☐ difficult ☐ very difficult
- Overall, the number of sessions was :
  ☐ well adapted ☐ sufficient ☐ insufficient ☐ very insufficient ☐ no opinion
- The personal cost incurred was :
  ☐ nil ☐ partial ☐ total specify £ ☐ / session
  You found this amount : ☐ acceptable ☐ excessive
- Most of the sessions took place :
  ☐ at home ☐ private practice ☐ hospital ☐ clinic ☐ specialised centre for your disease
- specify where :
  ☐ same region ☐ other region ☐ other country ☐ miles from home
  You found this distance : ☐ acceptable ☐ excessive
- journey completed : ☐ alone ☐ with a relative / friend ☐ with professional assistance or transport
- You found the time to obtain the first appointment :
  ☐ very short ☐ short ☐ long ☐ very long specify how long ☐ month
- This type of care responded to your expectations :
  ☐ fully ☐ partially ☐ poorly ☐ not at all

☐ NO

Why ? (several responses possible)

- Professional could not be found or unavailable ☐ yes ☐ no
- Lack of prescription ... ☐ yes ☐ no
- Excessive waiting time ... ☐ yes ☐ no
  specify ☐ month
- Personal cost incurred too high ... ☐ yes ☐ no
  specify £ ☐ / session
- Location of the sessions too far ... ☐ yes ☐ no
  specify ☐ miles from home
  specify where ☐ same region ☐ another region ☐ another country
- Excessive time of journey ... ☐ yes ☐ no
- Nobody was available to accompany you ... ☐ yes ☐ no
- Cost of journey too high ... ☐ yes ☐ no
- Lack of suitable transport ... ☐ yes ☐ no
- Travelling too difficult (pain, fatigue, injuries) ☐ yes ☐ no

7 Have you needed **speech and language therapy** over the last 12 months ? ☐ YES ☐ NO → go to 8

Did you have access to this type of care ? ☐ YES

➢ Access to this type of care was :
☐ very easy ☐ easy ☐ difficult ☐ very difficult

➢ Overall, the number of sessions was :
☐ well adapted ☐ sufficient ☐ insufficient ☐ very insufficient ☐ no opinion

➢ The personal cost incurred was :
☐ nil ☐ partial ☐ total specify £ [ ] / session
You found this amount : ☐ acceptable ☐ excessive

➢ Most of the sessions took place :
☐ at home ☐ private practice ☐ hospital ☐ clinic ☐ specialised centre for your disease

specify where :
☐ same region ☐ other region ☐ other country [ ] miles from home
You found this distance : ☐ acceptable ☐ excessive

journey completed : ☐ alone ☐ with a relative / friend ☐ with professional assistance or transport

➢ You found the time to obtain the first appointment :
☐ very short ☐ short ☐ long ☐ very long specify how long [ ] month

➢ This type of care responded to your expectations :
☐ fully ☐ partially ☐ poorly ☐ not at all

☐ NO
Why ? (several responses possible)
- Professional could not be found or unavailable ☐ yes ☐ no
- Lack of prescription ☐ yes ☐ no
- Excessive waiting time ☐ yes ☐ no
  specify [ ] month
- Personal cost incurred too high ☐ yes ☐ no
  specify £ [ ] / session
- Location of the sessions too far ☐ yes ☐ no
  specify [ ] miles from home
  specify where ☐ same region ☐ another region ☐ another country
- Excessive time of journey ☐ yes ☐ no
- Nobody was available to accompany you ☐ yes ☐ no
- Cost of journey too high ☐ yes ☐ no
- Lack of suitable transport ☐ yes ☐ no
- Travelling too difficult (pain, fatigue, injuries) ☐ yes ☐ no

8 Have you needed (a) **surgical operation(s)** over the last 12 months ? ☐ YES ☐ NO → go to 9

Did you have access to this type of care ? ☐ YES

➢ Access to this type of care was :
☐ very easy ☐ easy ☐ difficult ☐ very difficult

➢ Overall, the number of sessions was :
☐ well adapted ☐ sufficient ☐ insufficient ☐ very insufficient ☐ no opinion

➢ The personal cost incurred was :
☐ nil ☐ partial ☐ total specify £ [ ] / session
You found this amount : ☐ acceptable ☐ excessive

➢ Most of the sessions took place :
☐ at home ☐ private practice ☐ hospital ☐ clinic ☐ specialised centre for your disease

specify where :
☐ same region ☐ other region ☐ other country [ ] miles from home
You found this distance : ☐ acceptable ☐ excessive

journey completed : ☐ alone ☐ with a relative / friend ☐ with professional assistance or transport

➢ You found the time to obtain the first appointment :
☐ very short ☐ short ☐ long ☐ very long specify how long [ ] month

➢ This type of care responded to your expectations :
☐ fully ☐ partially ☐ poorly ☐ not at all

☐ NO
Why ? (several responses possible)
- Professional could not be found or unavailable ☐ yes ☐ no
- Lack of prescription ☐ yes ☐ no
- Excessive waiting time ☐ yes ☐ no
  specify [ ] month
- Personal cost incurred too high ☐ yes ☐ no
  specify £ [ ] / session
- Location of the sessions too far ☐ yes ☐ no
  specify [ ] miles from home
  specify where ☐ same region ☐ another region ☐ another country
- Excessive time of journey ☐ yes ☐ no
- Nobody was available to accompany you ☐ yes ☐ no
- Cost of journey too high ☐ yes ☐ no
- Lack of suitable transport ☐ yes ☐ no
- Travelling too difficult (pain, fatigue, injuries) ☐ yes ☐ no

9 What type of care or medical services related to your disease have you needed over the last 24 months ? (several responses possible)

Consultations		Tests / explorations	Care
☐ Cardiology	☐ Gastroenterology	☐ Biological, biochemical analyses (blood, urine, etc.)	☐ Nursing care
☐ Pulmonary medicine	☐ Endocrinology / diabetes	☐ Microbiology (bacterial, viral, parasitic, fungal)	☐ Physiotherapy / rehabilitation
☐ Nephrology	☐ Hepato-pancreato medicine	☐ Genetic, chromosomal testing	☐ Injections / perfusions
☐ Internal medicine	☐ Genito-urinary medicine	☐ Biopsies / smear-test / cytological analyses	☐ Psychotherapy
☐ Haematology	☐ Gynaecology / obstetrics	☐ Standard radiological investigation	☐ Occupational therapy
☐ Oncology	☐ Immunology / allergies	☐ Specialised imagery examinations : PET-scan / MRI	☐ Psychomotility therapy
☐ Rheumatology	☐ Neurology / epilepsies	☐ Echographies	☐ Speech and language therapy
☐ Podology / Foot medicine	☐ Oral and maxillofacial medicine	☐ Doppler test	☐ Vision therapy / orthoptics
☐ Ophthalmology	☐ Ear, nose and throat medicine	☐ Electrocardiograms	☐ Dental care
☐ Dermatology	☐ Pain control services	☐ Electroencephalograms / evoked potential tests	☐ Surgical operations
☐ Psychiatry	☐ Nutrition / dietetics	☐ Electromyograms	
☐ Paediatrics	☐ Emergency services	☐ Functional testing (respiratory, muscular, etc.)	☐ Glasses, lens / visual aids
☐ Geriatrics	☐ Orthopaedics		☐ Hearing aids
☐ Genetics	☐ Rehabilitation medicine		☐ Internal or external prostheses

➢ **Hospitalisation** : over thes last **24 months** did you have to spend time in hospital : ☐ no ☐ yes : how many times ? [ ] for how many days in total ? [ ]

10 Have you been rejected by health professionals because of your disease ? ☐ YES, specify causes : ☐ NO → go to 11

☐ due to physical aspect (weight, height, appearance, etc.) ☐ due to disease-related behaviour (excitement, autistic behaviour, etc.) ☐ due to communicative difficulties (understanding, language, etc.) ☐ due to reluctance of professionals because of complexity of disease or of specialised care

11 Have you needed to meet a **social worker** over the last 12 months ? ☐ YES ☐ NO → go to 12

➢ Access to this assistance was ☐ very easy ☐ easy ☐ difficult ☐ very difficult ☐ impossible

➢ Which structure did the social worker belong to ? (several responses) ☐ medical structure ☐ associative structure (patients organisation, etc.) ☐ community care (administrative) ☐ insurance / mutual insurance company ☐ professional structure (employer, syndicate, etc.) ☐ religious structure

➢ Interviews were : ☐ in the social workers' office ☐ at home ☐ by phone

➢ Based on the following list of needs, did this social assistance meet your expectations :	entirely	good	somewhat	not at all	not concerned
• Information on the social, legal and financial rights	☐	☐	☐	☐	☐
• Referral to other services (psychological support, home care, etc.)	☐	☐	☐	☐	☐
• Information on the specialised technical supports (home or car adaptation, assistance of a dog, etc.)	☐	☐	☐	☐	☐
• Assistance with financial paperwork (acceptance of financial liability or reimbursement, allowance, etc.)	☐	☐	☐	☐	☐
• Assistance for obtaining exceptional financial support (purchase of wheelchair, home adaptation, consultation or medical care abroad, etc.)	☐	☐	☐	☐	☐
• Help in getting personal assistance (help at home, personal care, etc.)	☐	☐	☐	☐	☐
• Assistance with social integration (school, leisure, professional, etc.)	☐	☐	☐	☐	☐

Your expectations regarding specialised centres

The basic mission of a specialised centre for a rare disease is to ensure diagnosis and follow-up of patients by offering - in the same location - multidisciplinary consultations, medical examinations, specialised equipment and genetic advice.

12 According to your needs, how would you qualify the other following functions that could be provided by a specialised centre ?	of no use	of little use	useful	essential	no opinion
Providing occasional care related to the rare disease (surgery, prosthesis, orthopaedics, etc).	☐	☐	☐	☐	☐
Providing frequent care related to the rare disease (physiotherapy, speech therapy, psychotherapy, etc).	☐	☐	☐	☐	☐
Offering patients the option of grouping consultations or tests on the same day in the specialised centre, and organising the appointments.	☐	☐	☐	☐	☐
Coordinating the sharing of medical information on the patient between all professionals who care for him/her in the specialised centre.	☐	☐	☐	☐	☐
Facilitating the follow-up of patients at different stages of their life by easing the passage from paediatric care to adult care, or from adult care to geriatric care.	☐	☐	☐	☐	☐
Informing patients about their rights and guiding them toward social services, schools, leisure activities, or vocational guidance, etc.	☐	☐	☐	☐	☐
Creating material for teachers, employers, social services, insurance companies and the general public to inform them about patients' needs and improve the social integration of patients.	☐	☐	☐	☐	☐
Collaborating with research teams working on the rare disease (in particular for clinical studies)	☐	☐	☐	☐	☐
Monitoring the current needs of the patient community of this rare disease (through surveys or registers of patients)	☐	☐	☐	☐	☐
Training local professionals in responding to the specific needs of patients and supplying their contact information to patients.	☐	☐	☐	☐	☐
Coordinating the sharing of medical information between health professionals of the specialised centre and local health professionals, to facilitate the continuity of the patients' follow-up.	☐	☐	☐	☐	☐
Communicating with other specialised centres and professional networks to harmonise treatments and research at the national and European levels.	☐	☐	☐	☐	☐

13 Could you please rate the following statements regarding the potential implementation of specialised centres?	strongly disagree	partially disagree	partially agree	strongly agree	no opinion
A single, national centre would be preferable because it could gather all the medical skills and competences and the most up-to-date equipment in a same location.	☐	☐	☐	☐	☐
Rather than concentrating all the expertise and competences in a single, national centre, sharing them between several centres would be preferable, because more accessible to patients.	☐	☐	☐	☐	☐
To maintain the skills and experience of its professionals, a specialised centre must follow a high number of patients affected by a specific disease.	☐	☐	☐	☐	☐
Rare diseases are not well known by the majority of health professionals; it is therefore preferable to travel to a specialised centre for consultations and most specialised care.	☐	☐	☐	☐	☐
Quality of relationships is as important as skills and competences, therefore, a local professional is preferable because of freedom of choice.	☐	☐	☐	☐	☐
The main hurdles in travelling to a specialised centre are the cost of transport and/or the need to be accompanied by someone.	☐	☐	☐	☐	☐
The main hurdles in travelling to a specialised centre are the time needed to get there and/or physical difficulties encountered by the patient (pain, fatigue, and injuries).	☐	☐	☐	☐	☐
The role of the general practitioner consists mainly in looking after health problems not related to the rare disease.	☐	☐	☐	☐	☐
A specialised centre should involve patient organisations to benefit from their knowledge of daily life and needs of patients.	☐	☐	☐	☐	☐

14 Questionnaire completed by : ☐ the patient ☐ a relative ☐ personal care assistant ☐ other

age of the patient ☐ years gender ☐ male ☐ female age of the patient at diagnosis ☐ years

Where does the patient live ?

☐ village or city of less then 1000 inhabitants ☐ city of 1000 to 100 000 inhabitants ☐ city of more than 100 000 inhabitants ☐ capital city

Were you forced to move because of the disease ? ☐ no ☐ yes, specify :

☐ to a more adapted house ☐ to a specially adapted care centre ☐ to get nearer to specialists of the disease ☐ to get closer to a relative

15 Structure of the family : total number of adults ☐ total number of children ☐ number of patients ☐

Highest level of education in the family : ☐ primary education ☐ secondary / professional education ☐ university / higher education

Activities of patient or parents : ☐ working ☐ unemployed ☐ never working ☐ retired ☐ student

Occupations of patient or parents (present occupation if working, last occupation if not) : ☐ farmer / fisherman ☐ craftsman / tradesman ☐ senior management ☐ middle management ☐ office work or services ☐ skilled worker ☐ unskilled worker

Specify if sector is : ☐ health / research professional ☐ teacher

Net family income per month, from all sources (salary, allowances, etc.) : ☐ < £ 540 ☐ from £ 540 to £ 1,400 ☐ from £ 1,400 to £ 3,600 ☐ > £ 3,600

Because of the disease, did one member of the family have work less or stop his/her professional activity ?

☐ no ☐ yes, specify : ☐ as patient ☐ to take care of a relative

Thank you for your collaboration

APPENDIX J

Institutional Review Board Approval Letter

TWU
TEXAS WOMAN'S UNIVERSITY
DENTON DALLAS HOUSTON

Institutional Review Board
Office of Research and Sponsored Programs
P.O. Box 425619, Denton, TX 76204-5619
940-898-3378
email: IRB@twu.edu
http://www.twu.edu/irb.html

DATE: June 2, 2017

TO: Ms. Shirley Shropshire
Family Sciences

FROM: Institutional Review Board (IRB) - Denton

Re: Approval for Klippel-Feil Syndrome: A Study of Parent and Caregivers' Experiences of Diagnosis, Health Service Use, and Online Support in a Rare Disease Population (Protocol #: 19559)

The above referenced study has been reviewed and approved by the Denton IRB (operating under FWA00000178) on 6/2/2017 using an expedited review procedure. This approval is valid for one year and expires on 6/2/2018. The IRB will send an email notification 45 days prior to the expiration date with instructions to extend or close the study. It is your responsibility to request an extension for the study if it is not yet complete, to close the protocol file when the study is complete, and to make certain that the study is not conducted beyond the expiration date.

If applicable, agency approval letters must be submitted to the IRB upon receipt prior to any data collection at that agency. A request to close this study must be filed with the Institutional Review Board at the completion of the study. Because you do not utilize a signed consent form for your study, the filing of signatures of subjects with the IRB is not required.

Any modifications to this study must be submitted for review to the IRB using the Modification Request Form. Additionally, the IRB must be notified immediately of any adverse events or unanticipated problems. All forms are located on the IRB website. If you have any questions, please contact the TWU IRB.

cc. Dr. Karen Petty, Family Sciences
Dr. Linda Ladd, Family Sciences
Graduate School

APPENDIX K

Table 3 Initial KFS Symptoms

Table 3

Initial KFS Symptoms

Symptom	Percentage of Parents	Number of Responses
Torticollis	8.3%	3
Breathing Problem/ Lungs (example: reactive airway disease, difficulty breathing)	8.3%	3
Craniofacial Abnormality (example: odd shape head, cleft palate, facial asymmetry)	8.3%	3
Back Pain	5.6%	2
Limited Cervical Movement	5.6%	2
Short Neck	5.6%	2
Neck Pain	5.6%	2
Kidney Problem (example: one kidney, multicycstic kidney)	5.6%	2
Cervical Fusion	5.6%	2
Shoulder Problem (example: pain, lump on shoulder)	5.6%	2
Hand Problem (example: hanging thumb, no webbing)	2.8%	1
Numbness	2.8%	1
Swollen Neck	2.8%	1
Hearing Loss	2.8%	1
Unspecified Neck Problem	2.8%	1
Dizziness	2.8%	1
Rib Cage Deformity	2.8%	1
Swallowing Problem	2.8%	1
Knee Pain	2.8%	1
Chest Pain	2.8%	1
Low Muscle Tone	2.8%	1
Scoliosis	2.8%	1
Delayed Milestones	2.8%	1
Total	100.5%	36

Note. Parents reported up to three symptoms