

FABRY PATIENTS' PERSPECTIVES ON
MULTIDISCIPLINARY CLINICS

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of
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of Master of Science in Genetic Counseling

By
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CERTIFICATION OF APPROVAL

FABRY PATIENT'S PERSPECTIVE ON
MULTIDISCIPLINARY CLINICS

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DEDICATION

To my father, whose accomplishments in the face of having Fabry disease have inspired and ultimately led me to my career as a genetic counselor.

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I would like to acknowledge all who have contributed and advised me throughout every step of this research project. I would like to thank my committee members, Marta Sabbadini, Wendy Packman, and Janey Youngblom for the endless support they have provided.

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ABSTRACT

Fabry Disease (FD) is an X-linked inherited lysosomal storage disorder which leads to progressive tissue damage, particularly in the renal, cardiac, and cerebrovascular systems. Disease presentation can vary among FD patients, males tend to be more severely affected than females. The complexity of disease manifestation means that individuals affected with FD require multispecialty management. A Multidisciplinary Clinic (MDC) is a proposed care delivery model that can provide a more comprehensive approach to clinical management for complex diseases such as FD. This study aims to understand FD patients' opinions towards the use of an MDC for FD management. Thirty-two adult individuals with FD were interviewed over the phone using a semi-structured interview guide. Of the 32 participants, four were being seen at an MDC, and all four felt that they were getting better overall FD care from the MDC compared to a non-MDC setting and preferred the MDC to their previous clinic model. Additionally, all four would recommend the MDC to other individuals with FD. The remaining 28 participants had never been seen in an MDC. When asked about possible advantages to an MDC setting, the majority (26/28) mentioned the convenience of having various appointments in one day and several (16/28) mentioned the FD expertise of providers participating in the MDC. When asked about challenges, 12/28 participants mentioned that the clinic day could be long and time consuming, additionally 11/28 noted that travel to attend the clinic could be

an issue. About half (16/28) said it would not be overwhelming to see multiple providers in one day. Most (20/28) reported that they would be interested in participating in an MDC. These findings suggest that patients see various benefits and are interested in a multidisciplinary approach for managing FD. Evaluating and incorporating the perspectives of patients with FD is essential for the improvement of health outcomes.

CHAPTER I

INTRODUCTION

Fabry Disease

Fabry disease (FD) is an X-linked lysosomal storage disorder (LSD) caused by pathogenic variants in the GLA gene. Pathogenic variants in the GLA gene lead to deficient enzyme activity of encoded alpha-galactosidase A (α -Gal A). This enzyme functions in the lysosome and is responsible for the breakdown of globotriaosylceramide (GL3) in various tissue cells including the vascular endothelium, renal tissue, cardiac tissue, and central nervous system tissue. When α -Gal A is absent or deficient, GL3 accumulates in the lysosomes, leading to progressive tissue damage in various organ systems (Schiffmann, 2009).

The GLA gene is located on Xq22.1 (Bishop, Kornreich, & Desnick, 1988). According to the Human Gene Mutation Database (HGMD®) there are more than 900 known pathogenic variants (Stenson et al., 2017). Pathogenic variants that result in little to no α -Gal A activity lead to the classic form of FD. Pathogenic variants that result in higher residual α -Gal A activity lead to non-classic FD, a form of FD that tends to be less severe with less widespread organ involvement (Desnick, Ioannou, & Eng, 2001; Schiffmann, 2009). The incidence of classic FD is estimated to be 1 in 40,000 – 60,000 males. Newborn screening data seems to suggest that the incidence of non-classic variants may be much higher. A study in Italy found a high incidence

of FD closer to 1 in 3,100 males, with non-classic variants accounting for 92% of the diagnoses (Spada et al., 2006).

In individuals with classic presentation of FD, symptoms typically begin in childhood with a decreased ability to sweat, gastrointestinal issues such as abdominal pain and diarrhea, and episodes of neuropathic pain known as acroparesthesia, primarily in the hands and feet (Ries et al., 2005). While not present in all individuals with FD, angiokeratomas and cornea verticillatae are symptoms that typically do not impact health but can be helpful in making a diagnosis and manifest by adolescence (Sodi et al., 2007).

The cardinal complications of FD include the progressive decline of kidney, cardiac, and central nervous system function that typically present in untreated individuals by the third to fifth decade of life (Schiffmann, 2009). Impaired renal function begins with progressive proteinuria and a decline in glomerular filtration rate (GFR), eventually leading to end stage renal disease (Schiffmann, 2009). Cardiac manifestations can include cardiomyopathy, valvular insufficiency, and conduction abnormalities that can progress to heart failure (Patel et al., 2011). Cerebrovascular involvement presents as ischemic stroke in most severe cases but can also manifest as transient ischemic attacks, chronic white matter lesions, and brain vessel abnormalities (Burlina & Poliei, 2016). Individuals can also experience cochlear dysfunction leading to the development of tinnitus, vertigo and or hearing loss (Ries et al., 2007).

Individuals with FD can have psychological and psychosocial issues related to FD (Mehta, 2009). The presentation of debilitating symptoms such as recurring pain, heat intolerance, and gastrointestinal issues, can significantly inhibit peoples' daily lives while the progressive nature of the disease can engender a feeling of hopelessness (Kolodny & Pastores, 2002). Studies have found that individuals with FD have a lower quality of life compared to healthy individuals (Arends, Hollak, & Biegstraaten, 2015). This has been studied and confirmed in children as well, noting that the disease burden starts taking its toll early on (Ries et al., 2005). A study by Cole et al. (2007) found a high incidence of underdiagnosed depression among individuals with FD. Research comparing psychological functioning among four chronic illness patient groups found that FD patients report a significantly higher burden of psychological distress, similar to what is experienced by patients with chronic pain (Crosbie, Packman & Packman, 2009).

FD can be variable in disease presentation. Genotype-phenotype correlation can aid in determining classic versus non-classic disease presentation but there can still be significant variability even in individuals with the same familial pathogenic variant (Schiffmann, 2009). The disease is fully penetrant in hemizygous males however disease presentation in females is on a much broader spectrum due to X-inactivation and other factors. Women can range from being asymptomatic to developing all of the same complications seen in males. On average presentation in females tends to be less severe with a later onset (Wilcox et al., 2008).

Systemic manifestations of FD are currently managed using two approaches, enzyme replacement therapy (ERT) and more recently chaperone therapy (CT). ERT, the first treatment available for FD, has been commercially available since 2001 and involves the regular intravenous infusion of recombinant α -Gal A enzyme (Schiffmann, 2009). In most cases ERT is able to halt the progression of major organ involvement, particularly when intervention is started early in disease progression, but it cannot revert tissue damage. ERT does not address all symptoms so it should be used in conjunction with adjunctive therapies to manage FD complications. Guidelines stipulate that asymptomatic classic males should be considered for initiation of ERT in childhood. Timing for the initiation of ERT for females and individuals with non-classic FD has been more challenging to determine given the heterogeneity of disease but should be considered once there is biochemical, histological or imaging evidence of injury related to FD (Ortiz et al., 2018) CT is a newer treatment that has become available since 2018 and involves the oral administration of small molecules that facilitates folding of existing enzymes. Given its recent approval, recommendations about the use of CT have not yet been included into published guidelines. Pharmaceutical companies continue to work on the development of alternative therapies for FD. There are currently several new treatment modalities that are in various stages of clinical trials that will eventually provide clinicians and patients with more treatment options.

Multidisciplinary Care for Fabry Disease

Updated guidelines published in 2018 and developed by an international panel of FD experts from seven related subspecialties, emphasize the importance of a multidisciplinary and personalized approach to disease management. The panel determined that, “treatment and follow-up assessments to evaluate treatment responses should ideally be supervised by a physician experienced in the management of patients with FD, with input from sub-specialists who also have FD experience, as part of a multidisciplinary clinical team that includes a neurologist, nephrologist, cardiologist, medical geneticist, genetic counselor, psychologist, and nurse” (Ortiz et al., 2018).

Recent research conducted by Bawa (2018) provides insight into the existing care for FD as well as provider opinions regarding multidisciplinary clinics for the management of FD. Of the 67 participating providers, only 13.4% (N=9) were currently involved in a multidisciplinary clinic for the management of patients with FD. The more commonly used care model included use of a “quarterback” or team leader who oversees management of FD care. Additionally, participants reported the use of clinic coordinators that facilitate communication among specialty providers and patients. Existing multidisciplinary clinics were unique, demonstrating that a multidisciplinary clinic can be adapted to fit the needs of a particular institution and the patient population. Some reported a case conference that formalized the communication among various participating specialty providers, and some reported

seeing patients with related disorders in order to increase the number of participants in the clinic.

Providers reported support for the multidisciplinary clinic model, recognizing the opportunity for improved patient care. However, providers noted several barriers to this model. Providers noted not having enough patients with FD to justify a specialized multidisciplinary clinic, the potential geographical hardship for some patients, and the logistical challenges of scheduling, billing, and obtaining institutional support. And when considering disadvantages to MDCs, providers noted that not all patients will require consultation with all providers, there would not be enough time to accomplish all evaluations and physician clinic visits, patients might be put off by the long clinic day, and patients might feel overwhelmed seeing multiple providers in one day.

Multidisciplinary Clinics

The ongoing management of complex diseases has become more sophisticated and long termed (Gupta, 2007). The traditional care model for complex chronic illness involves seeing independent specialists and can include limited communication and coordination among patient providers (Katon, Von Korff, Lin & Simon, 2001). An alternative integrative care model provides comprehensive medical care by bringing together various specialty providers to work collaboratively with the goal of better managing complex diseases (Erskine et al., 2013; Grosse et al., 2009; Gupta, 2007; Makaray, 2011, Katon et al., 2001).

A multidisciplinary clinic (MDC) is a specialty treatment clinic that aims to provide coordination of care for a specific pleiotropic condition. Typically, a medical center offers focused MDCs for the treatment, management, and long-term care of patients affected by complex disorders (Grosse et al., 2009). These clinics bring together a team of different staff members including specialty physicians, nurses, and other relevant healthcare providers that specialize in the needs of patients with these complex conditions. MDCs allow patients to meet with multiple providers at the same time to receive disease management on a holistic scale (Makaray, 2011).

There are multiple benefits to a multidisciplinary approach to care that result in improved quality of care for patients with complex disorders (Tyler et al., 2015; Baker et al., 2005; Curtis et al., 2005; Erskine et al., 2013; Gupta, 2007; Makary, 2011; Mathes et al., 2004; Traynor et al., 2003). For example, consolidating the collective knowledge of varying specialists makes management more efficient and effective as providers are primed to consider the global impact of disease manifestation (Tyler et al., 2015). Additionally, pooling the experiences of multiple specialty providers within interdisciplinary discussions enables management decisions to be founded on a stronger platform. When referred to an MDC for vascular anomalies, most patients with complex cases that were previously undiagnosed or misdiagnosed, were able to receive accurate diagnoses and clear treatment and management recommendations (Mathes et al., 2004). Providers of a cutaneous lymphoma MDC find that patients experience higher satisfaction from the collaborative environment of the clinic (Tyler et al., 2015).

In addition to the improved quality of care, patients benefit from the convenience of consolidating specialist appointments into one visit. This can cut down on the overall time carved out of their schedule. Participating in an MDC may also provide patients easier access to supportive services with specialized health professionals (Erskine et al., 2013). Additionally, the MDC can provide an easier transition from pediatric to adult care if the MDC is able to care for both patient populations (Grosse et al., 2009).

MDCs have the potential to lower overall healthcare costs. They operate more on a preventative level and can recognize and treat critical problems earlier in their disease course therefore potentially eliminating the need for more costly intervention (Makaray, 2011). In terms of patient preferences, Makaray (2011) describes that patients are less satisfied with fragmented care and are more susceptible to ‘falling through the cracks.’ Additionally, Gupta (2007) noted that a multidisciplinary approach to care nurtures patients and caregiver confidence.

Even though there are many benefits and there have been reported improvements in quality of care for patients seen at MDCs, there are challenges associated with this care model. There are logistical challenges to the establishment and operation of MDCs. Facilities were simply not designed to support an interdisciplinary model, space that can accommodate different specialties on a rotating schedule is often lacking (Erskine et al., 2013). Additionally, there are challenges in determining reimbursement and there can be health insurance restrictions. Some of the most difficult hurdles to manage include the lack of specific

billing codes that appropriately meet the need of the services provided, the restrictions placed on reimbursement for providers outside of network, and the limit on the number of services billed for in an allotted time span (Grosse et al., 2009; Baker et al; 2005; Erskine et al., 2013).

Moreover, if not managed appropriately, an MDC has the potential to create an environment where resources are not used appropriately. Even though providers from multiple specialties are represented in the MDC, not every patient may need to see every single one of them. In order to prevent overutilization of unnecessary services, clinicians must be mindful of guidelines and be critical of the utility of services provided on an individualized patient level. When resources are not managed appropriately, MDCs can result in higher costs for health care systems (Makaray, 2011).

Additionally, a team approach has the potential to reduce an individual physician's sense of responsibility for the decisions made, it can also provide a false sense of reassurance that the correct decision is being made because others express support in it as well (Gupta, 2007). Addressing these challenges will require all participating medical providers to be actively engaged in critical assessment of conversations and decisions made in a team setting.

A challenge for patients includes the need for travel, given that not all patients reside near an institution that provides this specialized service. Some patients may decide that the benefits are not worth the distance and individuals who might benefit from being seen at an MDC may not do so. Also, having multiple appointments in

one sitting makes for a very long day in clinic which can be very draining for patients, enough so that this may not be the ideal care model for everyone. (Grosse et al., 2009; Erskine et al., 2013).

In a clinic for Cornelia de Lange syndrome (CdLS), researchers found that participants had an inclination for multidisciplinary care. Parents whose children had CdLS or a related disorder and who either attended (N=50) or did not attend an MDC (N=61) were surveyed about their preferences. Of those whose child had attended an MDC, 90% reported a preference for the MDC setting, 8% had no preference, and 2% preferred a non-MDC setting. Some parents suggested the opportunity for receiving the best health care and having access to multiple specialists as primary reasons for returning to the MDC. Of those whose child had never attended an MDC, 93% expressed interest in attending an MDC and 7% reported not being interested in attending. Both groups indicated that the major prohibitive factor to attending would be travel distance and expenses (January et al., 2016).

Study Purpose

Multidisciplinary care can take on various forms. The MDC model in which multiple specialty providers convene to see multiple patients in one day, is not frequently adopted. There are few centers in the United States that provide care in this way and MDCs for the treatment of FD have not been described in published literature. Likewise, information regarding FD patient attitudes toward this method of care are just as scarce.

This research study aims to understand how FD patients are being managed across the United States, their opinions regarding the quality of care they are currently receiving, and their attitudes towards MDCs for the management of FD. To the author's knowledge, the current study is the first to report patient opinions regarding their preferences in care models for FD in the United States. It is important for health care professionals to understand if patients are interested in attending an MDC, given that it factors into institutions' decisions to establish an MDC at their facilities.

CHAPTER II

METHODS

Research Approval

The California State University, Stanislaus Institutional Review Board approved the research project #1819-018 on October 10, 2018.

Participants

Eligible participants for this study were English-speaking individuals with FD living in the United States who were 18 years or older. Participants were recruited from the two national FD support groups, the Fabry Support and Information Group (FSIG) and the National Fabry Disease Foundation (NFDF). A letter describing the details of the study was provided to the organization leaders who then distributed it to subscribing members. Participants were incentivized to participate in the study with a five-dollar Target gift card. Those interested in participating were instructed to contact the primary investigator (PI) via email. Further recruitment was done directly from a known MDC at the University of California, Irvine (UC Irvine) in order to include additional perspectives from individuals who had experience being cared for in an MDC.

A total of 37 participants expressed interest in participating. Thirty-four of the participants were recruited through one of the two support groups - FSIG distributed the letter through their list-serve of 991 people, NFDF posted the letter on their official Facebook page, which had 3,054 people following the page at the time of

distribution. Additionally, three participants were recruited directly from the UC Irvine MDC for FD. The flyer was distributed through their list-serve of 32 patients. Of the 37 participants that expressed interest in participating, two were lost to follow-up, two were used as pilot interviews and were not included in the final analysis, and one was excluded due to audio malfunction. A total of 32 participants were included in the final data analysis.

Procedures

Data were collected using semi-structured interviews in order to provide reliable, comparable qualitative data, yet also have the flexibility to explore topics in further detail (Cohen & Crabtree, 2006). The researcher received input on the interview guide from various sources. Bawa's thesis, 'An analysis of current care models and opinions of providers on a multidisciplinary approach for the management of patients with Fabry disease,' provided initial guidance for shaping the interview guide. The article, 'Benefits and Limitations of a Multidisciplinary Approach to Individualized Management of Cornelia de Lange Syndrome and Related Diagnoses' by January et al. (2016) provided the framework for the structure of the interview guide. The survey instrument used in January's research had the same goal of understanding patient opinions about MDCs for the treatment of a complex genetic condition. A committee member, Marta Sabbadini, provided detailed directions in how to adapt the interview guide to best capture the opinions of patients with FD. The interview guide underwent several revisions, received approval from

the research committee, and was piloted on two participants to ensure flow and clarity of language.

The interview guide consisted of 30 closed- and open-ended questions (see Appendix 1 for the complete interview guide). It was split into two main parts, the descriptive quantitative data describing the sample population and the qualitative data derived from open ended questions. The qualitative section is split further into five sections that detail participants personal experience with FD management, current clinical experience, and opinions regarding MDC for the management of FD. The sections are as follows: Personal Experience with FD Management, Non-MDC Participants: Clinical Experience, Non-MDC Participants: Opinions on MDC, MDC Participants: MDC Experience and Opinions, and MDC Participants: Previous Clinical Experience.

In the “Personal Experience with FD management” section of the qualitative data, participants were asked about their personal experience with FD (all participants were asked these same questions). This section inquired into how participants were diagnosed, the level of FD major organ systems involvement, details regarding treatment and about the life impact of FD management. It also included the following statement and question about participation in a multidisciplinary clinic for FD: “In a multidisciplinary clinic (MDC) you have appointments in the same location with many healthcare providers who are specially trained in Fabry diagnoses and management. They work together, across many specialties, to develop a unique and comprehensive plan of care to address all of your healthcare needs. MDC clinics

generally include a geneticist, cardiologist, and nephrologist. Do you currently receive your Fabry care and management at a multidisciplinary clinic?” The language describing an MDC was adapted from the research instrument used by January et al. 2016. From there, the interview guide diverged based on the participant’s response to this same question.

Participants could fit into one of the three categories: *currently seen in an MDC (MDC participants)*, *previously seen in an MDC but not currently (previous MDC participants)*, or *never seen in an MDC (non-MDC participants)*. Each of those diverging sections consisted of two parts, one to understand what their current clinical care looks like and the other to understand their opinions regarding the use of an MDC for FD management. No participants fell into the *previous MDC participants* category.

The following two sections focus on the *Non-MDC participants*. In the “Non-MDC Participants: Clinical Experience” section, Non-MDC participants were asked about their current care: what specialist they are followed by, how often they see each of their FD providers, what they feel regarding the quality of care they are currently receiving for FD management. In the next section, “Non-MDC Participants: Opinions on MDC,” they were asked their opinions regarding the use of an MDC to manage FD: their current knowledge regarding MDCs, possible advantages or challenges they could see to an MDC, if they would find it overwhelming to see multiple providers in one day, if they would be interested in being seen in an MDC, their main reasoning

for why they would or would not be interested, and what they would change to improve their current method of care.

The last two sections focus on the *MDC participants*. In the “MDC Participants: MDC Experience and Opinions” section, MDC participants were asked details regarding the logistics of the MDC they participate in: what specialists they see in and out of the MDC, what a typical day in the MDC looks like, how often they are seen in the MDC, and how many miles or hours they travel to get to the MDC. They were also asked questions evaluating their satisfaction with the MDC care model: if they felt providers were knowledgeable, the advantages and challenges they have experienced associated with an MDC, whether they find it overwhelming to see multiple providers in one day, whether they would recommend this care model to other FD patients. In the final section “MDC Participants: Previous Clinical Experience,” they were asked how they would compare the MDC to their previous FD management, and what extra services they would like to see added to the MDC.

Prompts were used to help participants when they had difficulty coming up with a response to a question. Prompts were most often used when asking about challenges associate with participation in an MDC, particularly for the non-MDC participants. Prompts were based on information gathered from existing literature, personal experience of committee member Marta Sabbadini who has experience working with the FD patient population, and personal experience of the primary investigator.

The PI conducted interviews between October 2018 and February 2019. Prior to beginning each interview, the PI obtained verbal consent from the participant. Interviews averaged 30 minutes in length, were audio-recorded, and transcribed verbatim by REV – a professional transcription service. All data were stored on a laptop with a secure password, and the file containing participant contact information was stored with an additional security measure requiring an additional password to access. All transcripts were de-identified for coding and inclusion in the final research paper.

Data Analysis

Once all the transcripts were collected, the PI and a second coder, Jillian Thrall (JT), used directed content analysis, a qualitative method guided by prior research of theory (Hsieh & Shannon, 2005) to analyze the interview data. This method allowed for the use of existing literature to guide and focus the interview questions. Significant direction in developing the code book was provided by committee member Wendy Packman. Coding was started using pre-determined codes. Two distinct code books were developed, one for participants *currently seen in an MDC* and another for those *never seen in an MDC*. The PI developed the first version of these code books by identifying a set of themes based on the interview guide. A sample of three transcripts were independently analyzed for themes by the two coders. Then, the two coders worked together to establish a consensus. Data that could not be coded were identified, put aside and analyzed later to see if they represented a new category (Hsieh & Shannon, 2005). Coding schemes were refined,

discrepancies were negotiated until both investigators were satisfied with the level of agreement (Campbell, Quincy, Osserman & Pedersen, 2013). The two code books underwent several revisions, the final versions received input from committee members. All interview transcripts were independently coded by DT and JT and then jointly reviewed to finalize one consensus code book for each participant.

CHAPTER III

RESULTS

Part 1. Description of Sample Population

Demographics

Of the 32 participants who were interviewed, 23 were female and nine were male. Ages ranged from 28 to 72, with the average age being 45 years of age. There were no participants in the 18 to 24-year-old age group. Most participants identified as Caucasian, three participants identified as Hispanic, and one participant identified as African American. Participants were representative of 22 states. Geographical distribution was presented according to the National Coordinating Center for the Regional Genetics Network, which identifies seven distinct regions (Table 1).

<i>Participant Demographics</i>			
Characteristics		No. of Participants	%
<u>Gender</u>			
	Male	9	28
	Female	23	72
<u>Age Range</u>			
	18-24	0	0
	25-39	16	50
	40-59	7	22
	60 plus	9	28
<u>Ethnicity</u>			
	Caucasian	28	88
	Hispanic	3	9
	African American	1	3
<u>Geographical Region</u>			
	Western States	6	19
	Mountain States	10	31
	Heartland	2	6

Midwest	4	13
Southeast	3	9
New York Mid-Atlantic	6	19
New England	1	3

Diagnosis

The mean age of diagnosis was 34 years old. Circumstances that lead to their diagnosis are summarized in Table 2. Eighteen participants were diagnosed via cascade testing after a family member's diagnosis. In particular, one of these participants was diagnosed when her son was picked-up via newborn screening. Eight participants were found to have cornea verticillata via an ophthalmology exam. Two participants had cardiac issues that lead the cardiologist to consider FD on the list of differentials. Of the remaining four participants, one was diagnosed after being found to have proteinuria, one was diagnosed after a dermatology exam, one was diagnosed after having a stroke at 25 years of age, even though she had an uncle with a known diagnosis, and one was diagnosed via participation in a research study in which a sample of spinal tap fluid was tested for various conditions.

<i>Source of Participant Diagnosis</i>		
How Diagnosed	No. of Participants	%
Family Diagnosis	18	56
Ophthalmology Exam	8	25
Cardiac Involvement	2	6
Kidney Involvement	1	3
Stroke	1	3
Dermatology Exam	1	3
Research	1	3

Treatment

Treatment status is displayed in Figure 1. All male participants were on ERT, two of which were on a drug that is currently in clinical trials. Four women were not currently on any treatment, the remainder (19/23) were on ERT. Of note, none of the participants were on the newly approved oral CT treatment. Of those currently on ERT, 15 received infusions at a medical institution while 13 reported receiving home infusions. Ordering providers for ERT are listed in Table 3.

When considering response differences among participants not on treatment (4/32) and those on ERT (28/32), participants not on ERT had less overall experience with FD care. One participant was completely asymptomatic, another suffered from acroparesthesia but did not have any organ involvement, and the remaining two participants had been diagnosed in the last year and were still pending their evaluations to determine organ involvement. Overall these participants had less input when considering potential advantages and challenges to being seen in an MDC.

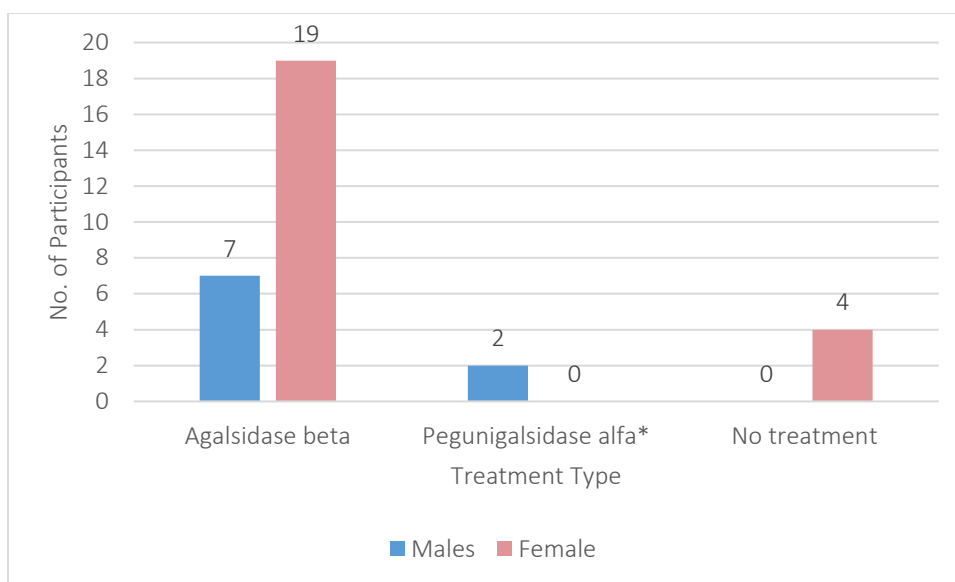


Figure 1. Treatment status of participants. This figure provides a comparison of treatment type between male and female participants. *Pegunigalsidase alfa is ERT currently in clinical trials.

<i>ERT Ordering Provider</i>		
Provider	No. of Participants	%
Geneticist	17	61
Oncologist	3	11
Nephrologist	3	11
PCP	3	11
Neurologist	1	4
Gastroenterologist	1	4

Part 2. Qualitative Data Analysis

First, responses of all participants (N=32) are listed. This is followed by responses of non-MDC participants (N=28) and responses of MDC participants (N=4).

Personal Experience with FD Management

All participants were asked about the impact that FD management has on their life. Four main themes emerged: impact on family, impact on work, managing medical appointments, and toll on emotional or mental health. These themes were not completely discrete, they overlapped and blended within participant responses. Additionally, it was challenging for the coders to separate feelings regarding disease management pertaining to clinical appointments and assessments, from disease management pertaining to the treatment regimen of bi-weekly infusions. Effort was made to focus on the impacts of disease management not pertaining to ERT.

Theme 1: Impact on family (N=5/32, 16%). Five participants, all of them female (5/23, 22%), mentioned the impact of FD management on their family. No males (0/9) mentioned the impact of FD management on family. Female participants noted the challenge of coordinating care for themselves and their children. Additionally, one participant talked about how she delayed getting her care established until she made sure that her children were being cared for first.

“So, you know, you go see a geneticist once a year and then our geneticist, he refers us out to all these other specialties. And some of us have to see this specialty, but not all of us. And then maybe I just have to see this one but not my girls. And especially with having children who are affected too. It does get to be so much to keep track of. And to be honest, since I really started being actively involved with our care as far as Fabry goes, I haven't maintained a full-time job since. And I actually was just awarded my disability claim like in April because I just can't maintain a full-time job and do everything I need to do for myself and my children. You can't always know like if sometimes I might be symptomatic after a treatment, but not always. So sometimes the next day could be really rough or not, or it's rough for my children so they miss school. So then somebody has to be home with them. So it's just really affected definitely my work lifestyle and finances.” (Participant #31, 33Y Female)

“I was busy getting [my children] taken care of and then I thought I needed to be able to take care of them if something happened, so I didn’t start getting treatment until they were moved out on their own.” (Participant #20, 60Y Female)

Theme 2: Impact on work (N=15/32, 47%). About half of the participants mentioned the impact that FD management has on work. They noted that it is difficult to fit in treatments and appointments without taking days off. Often participants did what they could to minimize the impact on work, for example making up hours for missed time, adjusting the number of hours they work, or shifting their schedule to part time or to a flex schedule in order to accommodate time for medical appointments. However, some reported that it was too challenging to keep up with work and not fall behind.

“So I'm a teacher, so I try to schedule appointments when we're out of school so that I don't have to take off work, but when we're in school I do have to miss. Especially when I was having those infusion reactions, I was having to go every other Friday for treatment once they put me back on treatment at [institution], which is two hours from where I live. So it is pretty difficult to try to keep track of everything and different appointments, not just nephrology, but cardiology, and I'm having to go see a neurologist, so yeah. It's difficult.” (Participant #06, 34Y Female)

“Definitely takes up time because like with work I have to take off time, so I have to use my vacation to go to my doctor appointments.” (Participant #07, 28Y Female)

“It is difficult, especially since my work – I've been working and I've kind of jumped a few jobs. So I never really feel like I have the job security, I always feel a little difficult to ask my employers without saying too much information, to ask for days off. So, I have kind of had a hard time with that. What I'm working on now, I have a job that's pretty good, so it's – but it's pretty hard. It's still hard even though I have some kind of job security.” (Participant #01, 39Y Male)

Of participants that were in the 60 and older age group, four participants (4/9, 44%) were retired. These participants all commented that FD management didn't impact their life as much because they had the time to devote to it.

“Because I'm retired ... I would say it impacts me far less than it would if I were working. In fact, I can't imagine how I would have managed it.”
(Participant #24, 72Y Female)

Theme 3: Managing medical appointments (13/32, 41%). Thirteen participants reported that it was challenging to keep track of all their different appointments, difficult to fit them into their busy schedule, and overall time consuming to their day-to-day life.

“I do not miss appointments, I do not miss doses ... My life revolves around making sure I am there for all of these appointments. That is first priority ... Everything revolves around these doctors and what they tell me to do.”
(Participant #33, 46Y Male)

“My life is just pretty much is Fabry's management. Huh. I'm constantly having to go to appointments. It's endless. It's endless and there's just so much that needs to get done. There's so many doctors that I have to see that I haven't seen yet. Like the ear doctor. By the time that I want to get around to seeing them, then something else comes up and then I have to, you know. It sucks. It's an all the time thing.” (Participant #14, 31Y Male)

Others (8/32, 25%) noted that it really wasn't a big impact for them and that they had incorporated the differences into their new normal.

“Since I've been dealing with the symptoms for so many years, I've kind of just gotten used to a lot of doctors appointments because there's so many disciplines that are involved. I'm self-employed and semi-retired, so I can devote the time for the appointments, or for the treatment. So that really hasn't impacted my life, it's just inconvenient to lose a whole day of your life every two weeks. But it's not anything significant by any means.” (Participant #30, 63Y Female)

More male participants (7/9, 78%) reported an impact on either managing medical appointments or impact on work compared to female participants (13/23, 57%).

Theme 4: Toll on emotional or mental health (N=8/32, 25%). Eight participants talked about the psychological impact of dealing with the management of FD. Males (5/9, 56%) were more likely than females (3/23, 13%) to report an emotional or mental health toll from FD management. Two participants (2/8) shared that they felt fortunate to be doing as well as they were.

“When I was diagnosed at the pathetic age of 48, I was pretty well a mess, bad GI problems too, but since I have been on ERT – People are surprised when I tell them – people are surprised that, “Oh, you have a genetic condition? You get infusions every two weeks?” You know what I mean? – So, I’m doing really well – and so I’m blessed.” (Participant #09, 61Y Female)

Others shared more negative experiences associated with disease management, such as experiencing feelings of fear and depression. Some of these feelings were in relation to the difficulty of dealing with the symptoms of the disease, but others were associated with the inability of health care to meet certain needs and expectations of the patients. Some patients expressed that the supportive services were general and did not appropriately account for the specific needs of FD patients.

“I need therapy for sure because I've just been gas lighted about my symptoms for so long. I just have so much on my mind, I've been through a lot. They set me up with this kind of generic therapist. It was just kind of – she would miss a lot of days then the next thing I knew, she was out of town. She left and I didn't even get to say goodbye. It just seemed like such a generic experience. It didn't seem like the fitting that I envisioned what was going to be able to help me.” (Participant #14, 31Y Male)

Participants were then asked about their medical management. The MDC setting was described to each participant, “In a multidisciplinary clinic (MDC) you have appointments in the same location with many healthcare providers who are specially trained in Fabry diagnoses and management. They work together, across many specialties, to develop a unique and comprehensive plan of care to address all of your healthcare needs. MDC clinics generally include a geneticist, cardiologist, and nephrologist.” Following this description they were asked, “Do you currently receive your Fabry care and management at a multidisciplinary clinic?” Twenty-eight participants reported never being seen in an MDC and four participants reported currently being seen in an MDC. This is where the interview guide branched depending on the participants results.

Non-MDC Participants: Clinical Experience

Twenty-eight participants were being cared for in a traditional care model in which they were seeing independent specialty providers for the management of FD involvement. Participants were asked about their current clinic experience. In particular, they were asked about what specialist they are currently followed by for the management of FD. Non-MDC participants reported being followed by several different providers (Figure 2). On average, non-MDC participants reported being followed by four different FD providers, this ranged from the lowest reporting only one provider to at most being followed by nine providers.

Participants were also asked how frequently they were followed by each provider, this varied significantly from one provider to the next for most participants.

Frequency occurred quarterly, bi-annually, annually, every two years, or on an as needed basis. Participants were also asked if their providers were under the same health system, 38% (10/26) reported that they were. Two participants were left out of that calculation because one only has one FD provider and the other is still in the process of establishing her care.

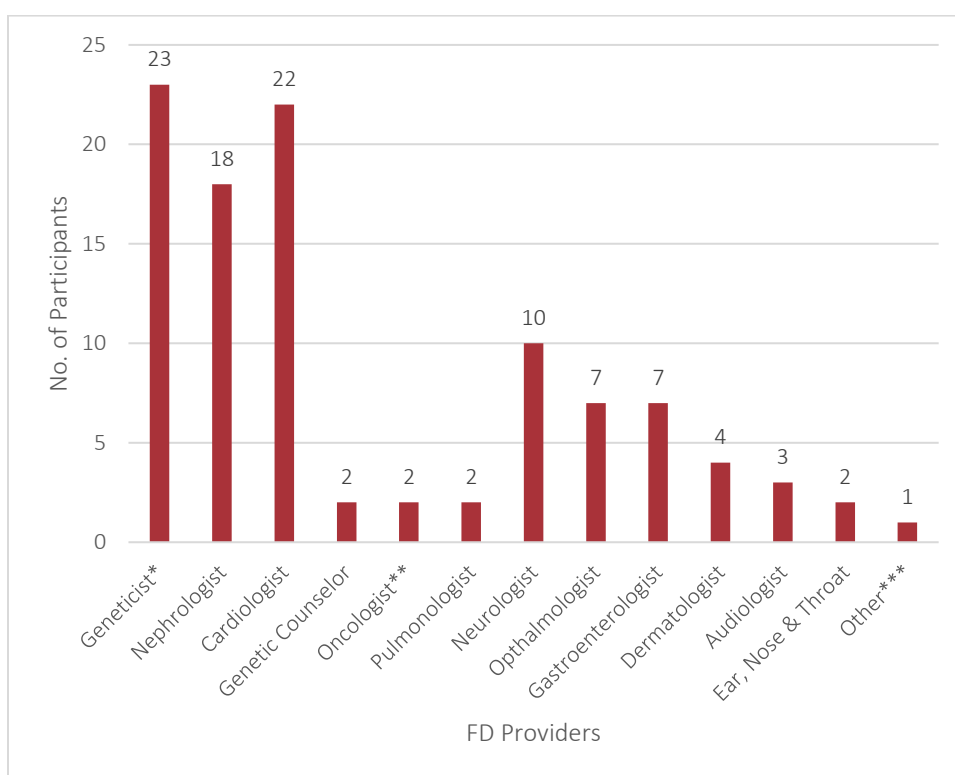


Figure 2. Non-MDC Participants – provider breakdown. This figure provides a summary of the different providers reported by non-MDC participants. *One participant (#08) sees two geneticists but only one is accounted for in the graph. **Two participants reported an oncologist as part of their FD care because this provider is responsible for overseeing their ERT. ***The other category represents additional providers only reported by one individual: a dietician, social worker, endocrinologist, pain specialist, and internist.

Communication. Non-MDC participants were asked about communication between their providers, 77% (20/26) participants reported that communication between their providers was good. Sentiments varied from being very sure of their communication, from not as confident but still feeling that they do communicate.

“They’re really good about communicating ... keeping that circle open.”
(Participant #28, 39Y Female)

“They’re in touch if they need to be.” (Participant #08, 36Y Female)

The remaining 23% (6/26) of non-MDC participants were not as satisfied with the communication among their providers and were either fully aware that it was not occurring or more unclear about whether it was happening or not.

“Absolutely not ... there’s not a lot of contact between them in terms of like, my geneticist is not out there educating them or talking with them, or looking at information saying, ‘oh yes, this is Fabry related,’ there’s none of that going on.” (Participant #29, 69Y Female)

Two of the participants were left out of this analysis because they had a recent diagnosis and did not have care established long enough to consider what communication between their providers might be like.

Of note, four participants talked about a genetic counselor (GC) contributing to their current care. However, when asked what providers they were followed by, only two participants mentioned GCs. The four participants that talked about their experience with a GC had varied experiences, from helping a patient access a drug on clinical trials to referring them to a PCP familiar with FD. One patient referred to the GC as the “quarterback” and another as taking on the responsibility of managing the life impacts of her genetic disease.

Rapport. Participants were not directly asked about their relationship with their providers, but 25% (7/28) volunteered that they had a good relationship with their providers. Some participants expressed a strong bond with their providers, noting how much time and effort it has taken for the providers to become familiar with the patient's disease manifestation, and thus a strong desire not to switch providers.

“The doctors I've had, I've had for several years, and developed a rapport with them. I wouldn't necessarily want to just go to another doctor, but it's taken me several years to get these doctors up to speed on the Fabry anyway now that I have it. So, I wouldn't want to start over with a bunch of new doctors unless I had to.” (Participant #30, 63Y Female)

Provider Knowledge. Non-MDC participants were asked if they felt that their current providers were knowledgeable about FD and its management. Participant responses varied from provider to provider, generally participants would respond that at least one of their providers, more often a specialty provider such as a geneticist, is knowledgeable about the disease and that the others are not as much. Most noted that of their providers, their primary care provider (PCP) was the least knowledgeable about FD. Participants also reported that some of their providers did not know about FD at first, but they researched the condition to become knowledgeable. For 28% (8/28) of participants Genzyme played a significant role in educating their providers about FD and its management. Participants not on ERT (4/32) all felt that providers they had weren't very knowledgeable about FD.

Quality of Care. Management guidelines for FD indicate that individuals should have annual assessments evaluating renal, cardiac, peripheral nervous system,

and cerebrovascular involvement, as well as an assessment of GL3 accumulation.

Participants were asked whether they get annual evaluations. The majority of participants, 96% (25/26) reported that they do get them done. Two participant who are still in the process of establishing care were left out of this calculation.

Participants were also asked if they thought they were getting good care in terms of FD, and thus if they were satisfied with their current FD care. Most participants, 75% (21/28) reported that they felt they were receiving good care.

“Yeah, I think so. I mean my doctor ... he’s always looking out for new and upcoming drugs ... so I think he generally cares about it ... I feel like I’m not really lacking anything that I should be receiving right now.” (Participant #17, 28Y Male)

A more ambiguous answer was obtained in 18% (5/28) of cases. Participants did not state they thought they were getting good or bad care, rather they were more noncommittal in their response.

“I’m receiving as good of care as one can receive when you don’t have very versed [providers], other than the geneticist, about FD.” (Participant #29, 69Y Female)

The remaining participants (2/28) felt that they were not receiving very good care. Specifically, they felt that their providers did not have adequate knowledge about FD and its management.

“No probably not. I mean I see my geneticist very seldom. My nephrologist is the one that’s my main plan of care right now, but as far as FD, she’s not knowledgeable ... so I don’t think that the typical Fabry care is happening right now.” (Participant #26, 31Y Female)

Additionally, some participants talked about other elements that impact their experience with their current FD care. For example, a few participants (3/28) reported

long clinic days or travelling far to access FD care. When discussing coordination of care, some participants (14/28) reported a point provider, or staff on their clinical team, that assisted in coordinating necessary assessments related to FD. For one of the 14 participants it was a Genzyme representative who was instrumental in coordinating her FD care. Other participants (7/28) noted taking on some of the responsibility of triaging information between providers.

Non-MDC Participants: Opinions on MDC

The non-MDC participants were asked to consider advantages and challenges to participating in an MDC for the management of FD. For most participants (22/28, 79%), this was the first time they had heard about an MDC type of care model. Of those that had prior knowledge of the MDC care model (6/28, 21%), one participant (#22) had experience participating in an MDC for breast cancer treatment.

Advantages to MDC. In considering potential advantages, five themes resulted from participants' responses: provider knowledge, provider collaboration, convenience, improved quality of care, and opportunity to meet others.

Theme 1: Provider knowledge (N=16/28, 57%). Sixteen participants noted that provider knowledge about FD would be an advantage of an MDC setting. Participants recognized that being part of an MDC for FD would mean providers would have to be knowledgeable about the disorder, and the experience of seeing multiple patients with FD would eventually contribute to their expertise as well.

“I think it would be wonderful to have somebody who really understands it, who would be able to talk about test results with an understanding of FD A really knowledgeable person who ... can interpret them properly.”
(Participant #29, 69Y Female)

“That would be really amazing to have specialists who knew exactly what they were looking for in respect to FD and it would be really good because you would have [providers] that were obviously up to date on everything that has been discovered recently.” (Participant #32, 56Y Female)

Theme 2: Provider collaboration (N=14/28, 50%). Fourteen participants also noted that communication between providers would be enhanced in the MDC setting. Participants speculated that there would be more coherence among providers regarding management approaches and recommendations.

“I think there probably would be better coordination ... between doctors, if the patient is having some kind of an issue instead of having a lot of back and forth between either different doctors and the patient, it seems like you could probably accomplish more in one day that way.” (Participant #08, 36Y Female)

No clear correlation resulted between these participants and their statement about their satisfaction about their current provider’s communication with other providers. Some noted good communication, others noted that communication could be improved, and other didn’t really have a clear opinion about provider communication.

Theme 3: Convenience (N=26/28, 93%). Almost all participants, recognized the convenience of having their clinic appointments consolidated into one day.

“Being able to see all those [providers] at one time and not having to take off multiple days ... less time off work ... anytime I can get my appointments all together it’s so much better to me.” (Participant #06, 34Y Female)

Theme 4: Improved quality of care (N=4/28, 14%). Four participants mentioned that management in an MDC setting would be more effective. This sentiment takes into consideration several of the other advantages mentioned already.

“You would have a higher standard of care for Fabry if you went to an MDC.”
(Participant #16, 29Y Female recently diagnosed and not on ERT)

Theme 5: Opportunity to meet others (N=2/28, 7%). Two participants talked about having the opportunity to meet others with FD as a potential advantage.

“It’s kind of rare to meet somebody with Fabry’s. The people who have the disease get to meet each other and talk about their experience and maybe kind of share their notes.” (Participant #14, 31Y Male)

Challenges to MDC. In considering potential challenges, four themes resulted from participant responses. Sometimes participants could not immediately think of any challenges, so they were prompted with a list of speculated challenges based on existing literature. Four themes emerged: lack of rapport with providers, long clinic day, location or travel, and loss of personalization.

Theme 1: Lack of rapport with providers (N=5/28, 18%). Five participants noted a disadvantage of potentially not liking one of the specialists that is part of the MDC and not having the option to change that provider.

“I can see possibly not caring for one of the physicians, if that puts you in the position where you’re kind of stuck with that person, yeah, that would not go well.” (Participant #23, 62Y Female)

Other participants (4/28) noted that losing the rapport established with their current set of clinicians and having to start from scratch with new providers if moving to a new care center, would be a significant disadvantage. Two of the participants noted a good relationship with their current providers. One of the two expressed a strong longstanding relationship with her provider in which she has been privy to his

children's life milestones. Some considered it enough of a disadvantage influencing their consideration in potentially participating in an MDC.

“If you already have a set of doctors that have been caring for you and to go to an MDC, that you would have to switch and change a whole new set of doctors.” (Participant #21, 66Y Female)

Theme 2: Long clinic day (12/28, 43%). Twelve participants noted that having multiple appointments in one day would make for a time-consuming event and for a very long day.

“You could possibly be there all day.” (Participant #07, 28Y Female not on ERT)

Particularly, two of those participants felt strongly that seeing the majority of their providers in one day would not be convenient. They were concerned that it would just be too much for one day and were unsatisfied with the intervals of the clinic. One preferred to be seen on an as needed basis and the other disliked not being able to see her provider on time intervals set by her.

“If it's in the effort of saving time, it just doesn't seem like something that I would be attracted to until I saw it ... I can't [see any advantages] because you just get all your appointments over at one time ... I think that's too much in one day.” (Participant #12, 61Y Female)

One participant also noted that if you can't attend that one clinic day then you can't get your FD management.

“[With a] one day kind of deal, what if it doesn't work out for them that particular day?” (Participant #24, 72Y Female)

Theme 3: Location or travel (N=11/28, 39%). Eleven participants noted the challenge of location that an MDC would contribute, and that individuals might have to travel particularly far in order to reach the nearest MDC.

“Location is probably going to be an issue for somebody no matter where it is.” (participant #19, 32Y Female)

Theme 4: Loss of personalization (N=6/28, 21%). Six participants considered that an MDC could result in a loss of personalization in the care.

“It’s kind of like you’re just a number kind of thing, like they’re trying to get you in and out really quick.” (Participant #06, 34Y Female)

MDC setting overwhelming. Participants were asked if they thought it would be overwhelming to see multiple providers in one day (Figure 3). A majority of participants (N=16/28, 57%) did not think that seeing multiple providers in one day would be overwhelming.

“No, I would prefer it. Just because it’s wham, bam, and you’re done for either six months or a year. For me I would really like it, I would probably be tired by the end of the day but I think it’s worth it.” (Participant #03, 44Y Female)

Seven thought that it would be overwhelming (N=7/28, 25%) and five thought that it might be overwhelming to some degree (N=5/28, 18%). Participants noted that it would be emotionally difficult to spend so much time focusing on their disease. Others responded that they could see how it could be overwhelming but considered that they might manage well. Most did not consider it significant enough to completely disregard the idea of participating.

“I think anytime you start to do a lot of testing and things like that, it does slightly get overwhelming because I don't think of myself as a sick person. Well, I do when I go through a lot of testing in a period [of time]. It kind of

brings to mind, oh geez, I've got some stuff going on here. So, in that sense, yes it could be slightly anxiety producing, I suppose. But I think it'd be fabulous to do it in a day ... I think it'd be wonderful to be that efficient.”
(participant #29, 69Y Female)

“Yes, It just would be very overwhelming to me ... just too trying on my body and too emotionally draining.” (Participant #12, 61Y Female)

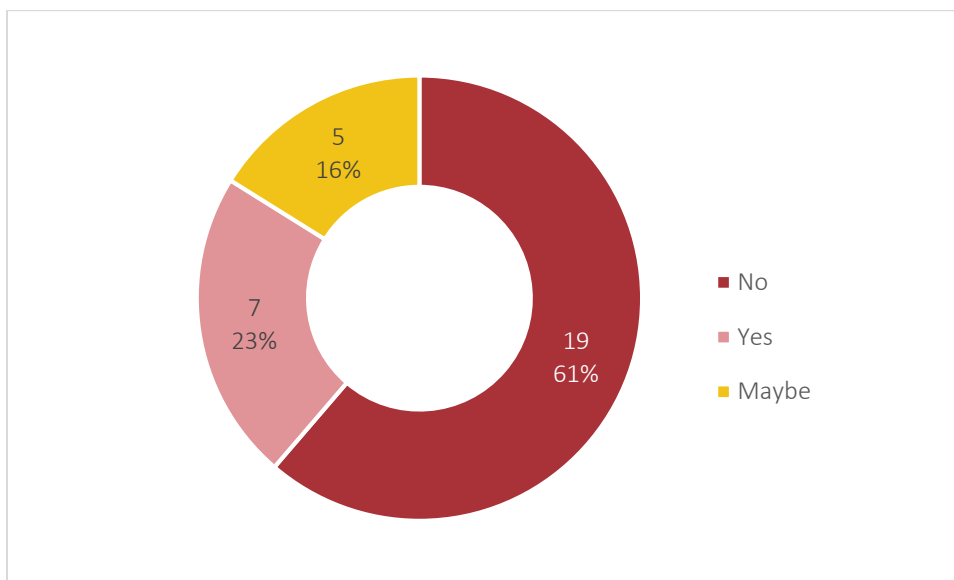


Figure 3. Non-MDC Participants – Overwhelming to see multiple providers in one day? This figure illustrates the responses of non-MDC participants to the question, do you think it would be overwhelming to see multiple providers in one day.

Interest in participating in an MDC. After considering the potential advantages and challenges to participating in an MDC, and after considering if seeing multiple providers might be overwhelming, participants were asked if they would be interested in being seen at an MDC, if that were a possibility (Figure 4). Most participants (N=20/28, 71%) reported that they would be interested. Participants that responded with no (N=3/28, 11%) or maybe (N=5/28, 18%) were primarily not

interested because they were not interested in switching providers. All three of the participants who reported not being interested in being seen in an MDC setting were from the 60 and older age group. Participants in this age group expressed disinterest in having to change providers. They reported that they had taken time to establish good rapport with their providers and that they were happy with the way their FD care was being managed. Five of the eight participants reported satisfaction with their current provider's level of knowledge and communication in their current clinical setting.

“If it meant that it couldn't be my same doctors, that I wouldn't have to start all over again, then no I would not be interested in that.” (Participant #21, 66Y Female)

Of the remaining individuals who said no or maybe (8/28, 29%), two noted that they needed more information in order to decide.

“Well I guess, I don't want to say no, cause when you first hear of something new you don't know enough about it to really know if it's going to be worth your effort.” (Participant #10, 71Y Female)

The other two individuals were steadfast about not being interested because they would find it overwhelming.

“It just would be very overwhelming to me, and I don't think that I – I think that when you're doing the – I don't know how you wanna call it, when you're doing the mass flu shot thing, that your doctors aren't gonna, you're not gonna get specific care.” (Participant #12, 61Y Female)

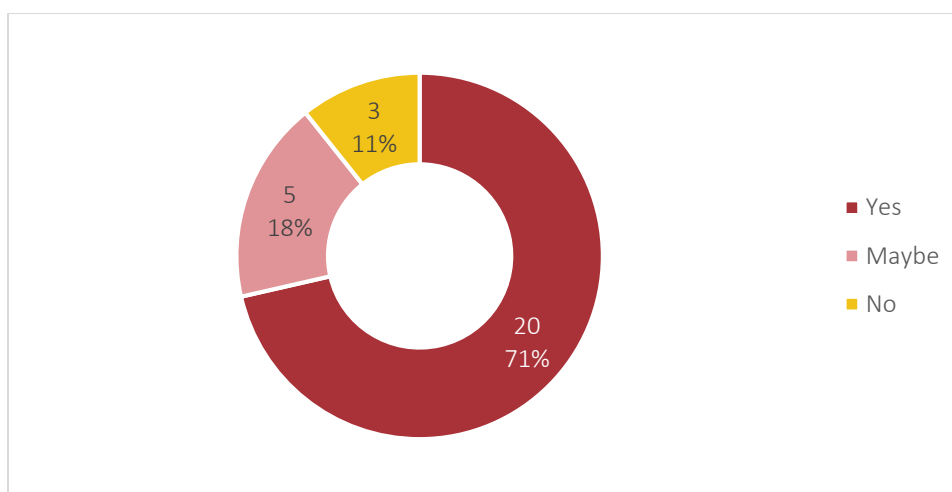


Figure 4. Non-MDC Participants – Interest in being seen at an MDC. This figure illustrates non-MDC participants’ interest in participating in an MDC for FD management if possible.

Those who were interested, or reported maybe being interested in attending, were asked what keeps them from attending an MDC. Most participants (N=22/25, 88%) noted that either this was the first time they were hearing about his care model for FD or that they didn’t know where they were offered. One participant noted that if an MDC was far away, he would not consider it. And the other two participants noted that they did not consider that their disease was severe enough to warrant participation in an MDC.

“I don’t really see multiple doctors right now, I only see the nephrologist ... so I guess really until I need it. Like I think it’s a great idea for people who have to see multiple doctors at one time, I mean that’s excellent.” (Participant #06, 34Y Female)

Additional insights. Non-MDC participants were asked what additional things they would like to see incorporated in their FD care or additional comments about MDCs. Five participants who had children affected by FD would want an MDC

that sees adult and pediatric patients, that way multiple affected individuals in the family can receive their care at once.

“It would be amazing to have the doctors in one place if I have to travel far. And that would be for my whole family, because we’ve thought about doing it as a group type thing.” (Participant #05, 29Y Female, recently diagnosed, not on ERT)

Additionally, another patient mentioned that FD management should include a bigger emphasis on the quality of life issues related to FD.

“Well, what everybody is worried about is the kidney number, you know. Or my heart and stuff like that. What really suffers the most is just my experience, my experience of just being in the world. I feel like I need really special help to kind of like fine tune some kind of a way of dealing with Fabry's.” (Participant #14, 31Y Male)

MDC Participants: MDC Experience and Opinions

MDC Structure. The four MDC participants were asked details about their MDC experience. Participants are not identified in order to preserve anonymity. One participant reported being seen at an MDC for FD at a university medical center in the Southeast region. Three participants attended the same MDC at the University of California, Irvine (UCI). These participants were directly recruited from the MDC in order to provide more participants with this perspective. The following clinic descriptions are based on the information provided by the four MDC participants.

The MDC in the Southeast is a half day if patients are only seeing providers but a full day if assessments such as bloodwork or imaging, are being done as well. The clinic is organized by an oncologist and participating specialty providers include a nephrologist, cardiologist, and pulmonologist. The clinic is working on recruiting a

neurologist to join the MDC. The participant attends clinic every six months, alternating from half day to full day each time.

At UCI, the clinic is split into two half days, with patients either attending the morning or the afternoon appointment sessions. The providers organize an educational luncheon that all patients are encouraged to attend. The clinic is organized by a geneticist and participating specialty providers included a nephrologist, cardiologist, and genetic counselor, although not all patients reported meeting with each of them. Patients attended at different frequencies, one participant reported attending clinic every three months while the other two participants reported attending annually.

Specialty providers reported by all MDC participants are listed in Table 4. On average, MDC participants were followed by five providers. All participants reported seeing additional specialty providers outside of the MDC. One reported seeing a local cardiologist because they wanted to minimize the amount of time spent at the MDC since the commute is long to get there. Another has an additional nephrologist that is working with the patient regarding kidney transplantation.

Table 4							
<i>MDC Participants – Provider breakdown</i>							
Participant ^a	MDC Providers						Outside Providers
	<u>Gen</u>	<u>Nephro</u>	<u>Cardio</u>	<u>GC</u>	<u>Onco</u>	<u>Pulmo</u>	
A	PP	X	X	X	-	-	Neuro & Optho
B	PP	X	X	X	-	-	Naturopath
C	PP	-	-	-	-	-	Cardio
D	-	X	X	-	X	X	Trans Nephro

Note. PP = MDC Primary Physician, Gen = Geneticist, Nephro = Nephrologist, Cardio = Cardiologist, GC = Genetic Counselor, Onco = Oncologist, Pulmo = Pulmonologist, Neuro = Neurologist, Optho = Ophthalmologist.

^aParticipant ID number is not provided in order to maintain anonymity.

Distance traveled to get to the MDC varied significantly for all participants and had different impacts on shaping what care looks like for each. The shortest travel distance reported was 30 minutes. One participant travels one and a half hours to clinic and stays overnight at a hotel to accommodate for the long day. Another one travels three hours to get to clinic and chooses to only see the geneticist at the MDC in order to shorten time in clinic in order to avoid getting home significantly late. The last participant lives in a different state and flies into the city to attend the MDC.

Advantages to MDC. MDC participants were asked about advantages regarding participation in an MDC. In direct response to this question, three participants noted the convenience of seeing most of their providers in one day. One participant commented on the benefits of communication between providers. Collectively the MDC participants spoke about various positive aspects associated with the MDC. Seven themes emerged: provider knowledge, provider collaboration, rapport with provider, convenience, personalization, opportunity for meeting others, and recommending MDC to others.

Theme 1: Provider knowledge (N=4/4, 100%). All participants found their MDC providers to be knowledgeable about FD and considered them to have a high level of expertise in FD management.

“The doctors are more focused on Fabry disease and the different types of things it does to you from the other doctor's perspectives because they're sitting and watching the Fabry clinic ... they can see the new treatments, they

can hear about the patient's questions and stuff. They're more involved it seems like when they're all together.” (Participant #13)

Theme 2: Provider collaboration (N=3/4, 75%). Three participants talked about the advantage of providers being able to communicate with one another regarding approaches to patient care.

“The most important thing actually is that the doctors are communicating so I'm not getting one set of directions from one doctor, and a different set of directions from another doctor that conflict, and everybody knows what's on the playing field.” (Participant #33)

Theme 3: Rapport with provider (N=4/4, 100%). All participants reported having a good relationship with their MDC providers.

“It seems like [my geneticist] cares a lot more than other doctors that I meet ... you need some doctors that have more passion for what they do ... I get to see my doctors who have become like friends almost” (Participant #13)

Theme 4: Convenience (N=4/4, 100%). All participants talked about the convenience of being able to see most of their providers in one day.

“The timing, scheduling. I only have to take one day off. You know, instead of many and juggling different things. It is, it's been a big change, going once compared to going two or three times. And just driving to one place is better than driving four times to the same place or across town, so I do like it. There is a lot of benefits.” (Participant #01)

Theme 5: Personalization (N=2/4, 50%). Two participants felt that the MDC provides an opportunity for individualized care.

“I think the biggest advantage is actually seeing the doctors, I don't know if they've been doing it behind the curtains, that they talk to each other. Me actually seeing that they talk to each other, recommend it, more like they tailor something for me.” (Participant #01)

Theme 6: Opportunity to meet others (N=2/4, 50%). Two participants spoke about benefit of being able to meet and talk to other individuals who also have FD.

“It’s nice to get acquainted with somebody who has the same problems as I do.” (Participant #20)

Theme 7: Recommend MDC to others (N=4/4, 100%). When asked if they would recommend the MDC to other individuals with FD, all MDC participants responded that they would.

Challenges to MDC. MDC participants were asked about challenges regarding participation in an MDC. In response to this question, two themes emerged: travel difficulty and lack of organization.

Theme 1: Travel difficulty (N=2/4, 50%). Two participants noted travel as a difficulty in participating in an MDC. Interestingly the participant who commutes across state lines and takes a flight to attend clinic, did not report travel as a hardship, even when directly asked. On the other hand, another participant noted:

“The only [challenge] that I see is the travel ... It’s a long drive because I live pretty rural.” (Participant #20)

Theme 2: Lack of organization (N=1/4, 25%). One participant found that the organization of the clinic was not consistent and could be improved.

“I think I just mentioned that disorganization. If there would be someone that would help organize because sometimes I'd hear [about] – when they're gonna have them, and they're short notice ... They were a little disorganized, in the past two, three years, I feel like they have been more organized than they were before.” (Participant #01)

One participant did not think there were any challenges to participating in an MDC. Collectively three of the MDC participants, in some way or another, also noted that the long clinic days could be a bit challenging to work with.

“Sometimes I feel like I wait a little too long, but that still over weighs having to go at a lot of different times. So, sometimes the time, the long day can be kind of hard.” (Participant #01)

Quality of MDC care. MDC participants were asked if they thought they are getting better overall care from the MDC, all four said yes.

“Even when my doctor sees an issue, and she consults with the other doctors, I feel like they come up with a better plan. And one example of that would be my high blood pressure, how they are kind of, not attacking it, but they're coming up with a plan. From the nephrologist's point of view, from the cardiologist's point of view, and you know the geneticist I think she just, she's overlooking but I do feel like it's better as a whole. I can see it as a bigger picture, not just this as a single discipline.” (Participant #01)

MDC setting overwhelming. MDC participant were also asked specifically if they found it overwhelming to see multiple providers in one day. Three participants responded that they do not consider it to be overwhelming. One participant who attends the MDC but only sees the geneticist, responded that they would find it overwhelming to see multiple providers in one day, but would still prefer it.

“Not at all because it's all in one and you're done. You don't have to go to four different appointments. The doctors can talk to each other about, say like I have a cardiac arrest or something and the doctor's ... they'll all come in and talk to me at one time and they'll kind of figure out things together. They'll be like, well you need to do this it'll help you, and the other doctor's like, yeah I agree that will help. It's just cool because they're all there and they can coordinate with each other.” (Participant #13)

“It might be a little overwhelming in the moment, yes, but I think, overall, for me it would be a benefit.” (Participant #20)

Adherence. MDC participants were asked if they found that they were adhering better to management recommendations from participation in an MDC. Three participants responded that they were. One attributed it to hearing the same recommendations from different providers, another because providers were explaining things in more detail, and the other because they felt that the providers are more involved and remind her to keep up with assessment. One participant felt that the MDC has not directly influenced his or her adherence, he or she noted always prioritizing managing personal health and reported always doing what the doctors recommend.

“Yeah, they give me way more advice and stuff ... [they really explain why they are making specific recommendations] ... I haven't had any other doctors tell me that kind of stuff.” (Participant #13)

Improvements to MDC. MDC participants were asked what recommendations they would have for the improvement of their MDC setting. Three recommendations emerged from this. A participant was interested in receiving a summary sheet after the visit that includes when labs and or assessments are next due. Another participant was interested in having assessment, such as MRIs, done during the MDC as well. Patients would be interested in seeing incorporated into the MDC a phlebotomist, gastroenterologist and neurologist. Additionally, a patient requested support for psychological services included as part of the MDC.

MDC Participants: Previous Clinical Experience

The four participants who were cared for at an MDC were asked about their previous clinical experience in order to compare to what their experience is currently

at an MDC. Two participants did not have a previous clinical model to compare to as their first experience being managed for FD was at the MDC. Following are more details regarding the care for the two participants with no previous clinical experience. One participant started being managed by the geneticist who eventually transitioned care into an MDC, and the other participant was not being managed for FD until attending the MDC. The latter did note some trouble getting their primary care provider engaged in FD management.

“I didn’t really have any Fabry care [before participating in the MDC], because my primary care was really not too interested in getting things taken care of. I didn’t really have any [FD] care at all. It was all scary at first because I didn’t really know how to really get him involved. I don’t think he even looked it up.” (Participant #20)

The other participants who did have previous clinical experience were both asked to recall what their past experiences were like receiving FD care in a non-MDC setting. One shared that previous providers were not knowledgeable about FD.

“I had some real big problems with doctors not knowing my disease, and knowing what's happening ... The most important thing [in the MDC] was having somebody in the group knowing the disease, knowing what's happening so the doctors aren't doing things they have no idea ... They may know their specialty, but they don't know my disease, and so they're doing things to me.” (Participant #33)

Another shared that they still receive some care from previous FD providers but that the MDC team takes priority because they have more experience caring for multiple individuals with FD.

“I’m see [local] specialists, I’ll see them again to do the testing and stuff but I’ll still have the doctors [at the MDC] that are my true specialists that I work with, they’re just getting the testing done [at the local institution]. So when I go to [my local specialists] the doctors are all nice and they’re doing their jobs

and stuff but it seems more, maybe more personable to me [at the MDC] because they're all there at one time and trying to learn about Fabry disease more than a doctor would if he's sitting by himself maybe just seeing me.” (Participant #13)

Preference. After recalling their previous care model, MDC participants were asked to compare whatever care they had before the MDC to their current care in the MDC and determine which care model they think provides better care and thus which they would prefer. All four participants preferred the MDC. The patients reiterated the advantages they had mentioned before as reasons for preferring the MDC over their previous care model.

“I have a preference for the multidisciplinary clinic again, for the reason that somebody knows the disease and that the doctor communicating.” (Participant #33)

CHAPTER IV

DISCUSSION

Multidisciplinary care takes on various forms and the utility of MDCs for the management of FD is still under evaluation. The current research compliments Bawa's study of provider opinions about existing MDCs for (Bawa, 2018). The current study focuses on the opinions of the patient population regarding MDCs for FD. Most participants in the current study did not receive their FD care at an MDC, consistent with research findings that 88% of FD providers work in a non-MDC setting (Bawa, 2018). Our data show that most individuals with FD are interested in an MDC for the management of FD. Those being cared for at an MDC felt that they were getting better overall FD care from the MDC compared to a non-MDC setting and preferred the MDC to their previous care model. The majority of those who had never been seen in an MDC expressed interest in being seen in an MDC. Many participants saw the MDC as being able to provide convenience, knowledgeable and collaborative providers, and that it could result in overall better quality of care. On the other hand, participants also noted that clinic days would be long and time consuming and that travel to attend the clinic could be a hardship.

Personal Impact of FD Management

Participants expressed that FD management had an impact on their day-to-day life. Male and female participants expressed a different emphasis on areas affected by FD management. Males tended to report it interfered more with work while the

burden on family responsibilities was reported only by females. Studies looking at gender differences are limited to the biological differences and as such this is a topic that has not been explored in the literature. This difference could possibly be attributed to traditional gender roles, but to determine this additional investigation is necessary. Overall FD management seemed to take up a significant amount of time in participants' lives. This was expected given that management of FD requires the involvement of several specialists, requiring patients to coordinate clinical appointments around work and personal schedules (Hughes et al., 2006; Mehta, 2009). It is likely that disease severity impacts the level of management, such that someone with more severe organ involvement sees more providers and has more medical appointments to manage disease progression. It would be beneficial to compare disease severity and disease management in future studies.

Eight participants reported an emotional or mental health toll from FD symptoms and management. These responses were more common among male participants. Participants expressed feelings of fear and depression and felt that healthcare providers had not met their needs. This is consistent with research from Cole et al. (2007) which found that FD can hinder day-to-day functioning and lead to depression which can be higher among males than females. Additional literature describes that the progressive nature of FD can intensify the psychological distress experienced by individuals with FD (Mehta, 2009; Crosbie et al., 2009). A few participants expressed interest in additional psychological services and supportive therapies that more specifically addressed complications related to FD. Although

neither of the two MDC settings described by MDC participants provided healthcare professionals with specialized experience in managing psychological FD symptoms, a multidisciplinary setting would be ideal for providing access to these services.

Most participants were receiving ERT and several noted how challenging and limiting it is to have a lifelong treatment that requires bi-weekly infusions. It was challenging for the coders to distinguish between participant sentiments regarding disease management based on clinical appointments versus treatment regimen. It was important for us to distinguish the two because this research was focusing on the clinical modality of management, not necessarily on the impact of treatment. Researchers recognize that the challenges associated with ERT can serve as additional stressors for individuals with FD (Crosbie et al., 2009). None of the participants were on the newly approved CT which is taken orally. It would be beneficial to talk to individuals who have transitioned from ERT to CT and explore the impact this change has had on their daily life.

A majority of participants (18/32, 56%) were diagnosed after a family member's diagnosis. This is in concordance with the literature which reports that for every patient diagnosed, on average there are five family members who are subsequently diagnosed as a result (Laney et al., 2013). FD can be a family affair and as such some participants expressed interest in MDCs that can accommodate families. Several participants who also have children who are affected by FD noted that it would be very helpful if the MDC was able to see adult and pediatric patients, thus making it possible for families to further consolidate FD care. This would also make

the eventual transition of pediatric care into adult care much more seamless (Grosse et al., 2009). Two providers in Bawa's study noted this is as an advantage of the MDC setting (Bawa, 2018). This may be feasible, given that genetic specialty providers are often housed in pediatric wings.

Opinions Towards Multidisciplinary Clinics for FD

Interest in MDC

The data show that most participants are interested in a multidisciplinary care model for the management of FD. Specifically, 71% reported they were interested in being seen at an MDC. This is slightly less than what was reported in a study surveying parents of children with Cornelia De Lang or related syndromes (January et al., 2006). For example, in their study of 61 participants, they found that 93.4% of participants who had not visited a multidisciplinary center were interested in attending. Of importance, 71% is still an overall strong interest in participation in an MDC for FD. Participants in January's study have a different perspective as they are parents of children attending an MDC and not actually patients attending an MDC themselves. Additionally, all participants in the present study who reported not being interested were 60 years of age or older, which could signify that their disinterest is more due to a discomfort in introducing change to the established care model. Participant age was not collected in January's study and thus cannot be compared to our findings. Furthermore, individuals who expressed interest in being seen at an MDC also had reasons for not wanting to uproot their current clinical model. MDC attendees (N=4) all indicated a preference for their current MDC setting. This is more

than in January's (2016) study in which 90% of MDC attendees indicated a preference for the MDC setting, however our sample size of four is much smaller than their sample size of 50. Although there are only four respondents in the current study, this supports the notion that individuals with FD would be interested in participating in an MDC.

Views on advantages to MDC

Participants in both groups reported similar actual and perceived advantages associate with the multidisciplinary care model. Both groups found the convenience of being able to get a majority of their FD clinic visits consolidated into one day as the most salient advantage. This is concurrent with the advantages reported by providers involved in FD care, noting the convenience for patients of having multiple appointments done in one day in one location (Palak, 2018). These findings support the idea that patients would embrace a “one-stop” approach to care and that patients highly value when specialist have extensive experience in FD (Tyler et al., 2015). Both participant groups also acknowledged that the convenience of the MDC means that clinic days will be long and time consuming, and most were willing to accept that.

Additionally, both groups noted provider expertise and collaboration as other major advantages that could ultimately lead to receiving better care. This was similarly reported by providers involved in FD care. Providers noted that this setting allows for comprehensive care delivery by knowledgeable providers who can communicate extensively with one another (Bawa, 2018). Participants saw the MDC

as an opportunity to benefit from the expertise of multiple providers knowledgeable about FD in one day, as well as minimizing the frequency of clinic visits.

Some participants also noted the opportunity for meeting other individuals with FD during clinic days as an advantage of the MDC setting. FD patients and families can feel isolated and alone (Crosbie et al., 2009). Having the chance to talk to others affected by the same condition can provide a form of community and support. Two providers in Bawa's study also recognized this patient interaction as a benefit of the MDC setting (Bawa, 2018).

Participants were also asked if they thought it would be overwhelming to see multiple providers in one day. Bawa (2018) explored provider opinions regarding MDCs for the management of FD. In her study she found that providers believed that patients would find an MDC overwhelming and thus a disadvantage of this care model. Our data show that 57% (16/28) of the non-MDC participants would not consider that seeing multiple providers in one day would be overwhelming. And even for those that thought it would be overwhelming (7/28) or were more uncertain about finding it overwhelming (5/28), some noted that the convenience would still make it worth it and were interested in participating.

Views on challenges to MDC

Individuals in both participant groups also had similar responses when it came to listing challenges associated with the multidisciplinary care model. Participants found that travel distance and travel expenses could be a major obstruction to attending an MDC. There are existing non-profit assistance models such as Patient

Services Incorporated (PSI) that help individuals with chronic illnesses access medical services in part by providing financial travel assistance (Types of Assistance). The MDC could facilitate identification of such resources, especially for those who might not attend an MDC due to travel barriers.

Additionally, some non-MDC participants thought an MDC might force them to stick with a provider that they might not like and could lead to a loss of individualized care. Studies exploring patient interest in MDCs are limited and thus it is unclear how relevant this issue is, this response can be attributed to the inherent limitation of asking hypothetical questions. In future research, it would be interesting to inquire further about this issue with participants who attend an MDC, to determine how often this comes up and how it is handled. This was not something that could be explored in the current research as none of the four MDC participants expressed this sentiment. Furthermore, the ability for a patient to change providers was not something that providers considered in their assessment of advantages or challenges associated with an MDC (Bawa, 2018).

Additional findings

MDC participants felt confident in the care they were receiving. Patients attributed this confidence to the expertise and collaboration among specialty providers participating in the MDC leading to the delivery of all-around FD care. Interestingly, MDC participants attributed improved adherence as a result of participation in the MDC. Responses seemed to imply that patients felt more involved in their care in response to improved rapport and communication between patient and

providers. This supports the idea that fully informing, educating, and including patients in the decision-making process regarding their medical plan results in increased compliance (Hughes et al., 2006). It was interesting to note that all MDC participants reported seeing additional FD providers outside of the MDC, meaning that existing MDCs are not entirely comprehensive regarding FD management.

Some MDC patients had comments regarding improvements that could be made to an MDC. Participants want additional FD specialists included in the clinic. Participants specifically mentioned a neurologist, gastrointestinal specialist, phlebotomist, and psychological services. Additionally, it seems that patients are still missing supportive services regarding managing quality of life issues. More emphasis should be focused on helping manage these issues in a manner that is specific to FD, or providers should make referrals to specialty providers who might be able to provide further insight (Grosse et al., 2009).

Study Limitations

With only 32 participants, the sample population of this study was small. Incorporating more patients' perspectives would strengthen the themes that emerged during this study. Additionally, the perspective from participants who currently attend an MDC clinic was extremely small at only four participants, of which three were directly recruited from a known MDC center.

Another limitation is that the majority of the individuals who participated in the study were females, with males being just over a quarter of participants. Since FD is an X-linked disorder that tends to impact males more severely than females, it

would have been beneficial to get more male perspectives. Males tend to have a higher disease burden and might have a stronger or different perspective than females.

Finally, the use of hypothetical questions to ascertain opinions about MDCs is also a limitation. Most of the respondents were not familiar with the MDC care model and had never received treatment at an MDC. Participants' understanding of an MDC was limited to the researcher's description; thus, their understanding of an MDC could be incomplete.

The PI and several members of her family have FD. She previously worked directly with the MDC for FD patients at UC Irvine before starting the Master of Science in Genetic Counseling program at California State University, Stanislaus. Additionally, she has accepted a fellowship position at UC Irvine working with the same department which she will start after receiving her degree. She is aware of her bias toward MDCs for the management of FD given her lived experience. A concerted effort was made to avoid introducing bias throughout this project. During interviews she adhered to the interview guide unless asking probing questions and a second coder independently coded the transcripts to identify themes in participant responses.

Further Research

Further investigation into patient opinions needs to be conducted, particularly towards elucidating patient attitudes in participating in an MDC for the management of FD. Perhaps a survey format would be able to reach more individuals throughout

the United States. Additionally, the perspective of more individuals that are already participating in an MDC should be actively pursued. They have valuable insight into the real advantages and challenges of participating in an MDC and their feedback would be critical for determining how best to optimize patient care.

Participants who were cared for at an MDC had varying experiences and the two MDC clinic setting represented by the MDC participants seemed to vary significantly. It would be beneficial for centers who currently have an existing MDC for the management of FD to publish an in-depth analysis of their clinic model. Bawa was able to collect some information regarding the structure of existing MDCs for the management of FD but the findings are limited (Bawa, 2018). Understanding how existing MDC are run and how they were established can help other providers learn from their experiences. Additionally, it might be beneficial to include information about financing since this seems to be a major hurdle in establishing an MDC.

CHAPTER V

CONCLUSION

The analysis presented here is limited but provides preliminary data suggesting that patients see various benefits and are interested in a multidisciplinary approach for managing FD. Most participants felt that providers in an MDC would have a greater level of understanding of FD natural history, management, and treatment options. Participants also felt that communication among providers would result in cohesive directives. Patients considered that in conjunction these factors would translate into improved quality of care. Additionally, all patients noted the convenience of minimizing the overall number of days spent in clinics which would open-up their schedule to other life events. Having a multisystemic disorder involves the coordination of several different medical specialties so being able to consolidate them and create in person collaboration among providers strongly appeals to individuals that are already having to juggle so much.

The findings from our study can be used to help shape the future of long-term FD care. Eliciting patient opinions and perspectives is critical to care optimization. Taking into consideration patient preferences is always an important variable to investigate when determining how best to approach the delivery of medical care that appropriately meets patient needs.

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APPENDICES

APPENDIX A

PARTICIPANT RECRUITMENT LETTER

Hello,

My name is Daisy Tapia, I am a graduate student in the MS Genetic Counseling Program at CUS Stanislaus. I would like to invite you to participate in a research study as part of my master's thesis exploring Fabry disease patients' perspectives regarding multidisciplinary clinics.

All English-speaking individuals 18 years and older with Fabry disease are encouraged to participate.

I will be conducting an over the phone interview that should take approximately 30 minutes to complete. The interview will be audio-taped, and all information will be confidential. You will not be asked to provide any personally identifying information. Participation in this study is entirely voluntary; you may stop at any time.

Participants will receive a \$5 Target gift card as a thank-you for participating.

If you are interested in participating or have any questions, please email me at dstapia@ucdavis.edu.

This study was granted approval through the California Stanislaus University Institutional Review Board.

Thank you for your time and consideration.

Sincerely,
Daisy Tapia
MS, Genetic Counseling Student
California State University Stanislaus
dstapia@ucdavis.edu

APPENDIX B

INTERVIEW GUIDE

Date: _____

Participant ID: _____

INTRO: Hello, thank you for volunteering to participate in Fabry disease research. Before we get started I'd like to go over the verbal informed consent to participate in this research.

VERBAL CONSENT: see verbal informed consent document

DEMOGRAPHICS

Age:	Gender:	Race/ethnicity:	State of Residence:
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PERSONAL EXPERIENCE

1. How and when were you diagnosed with Fabry disease?
2. Tell me about the level of involvement Fabry disease has had on your major organ systems? *Cardiac/Renal/Central nervous system; gastrointestinal issues, pain, strokes, etc.*
3. Are you on treatment?
 Fabrazyme (Genzyme ERT) *Galafold/migalastat (Amicus oral)* *Trial Drug*
 - a. When did you start?
 - b. Who is the ordering physician?
 - c. Where do you get your infusions? *Is that your only option? Can you do home infusions?*
4. When you visit clinic, do you coordinate to be seen on the same day as other family members? *Such as parents or children?*
5. Some people state that Fabry disease management, such as receiving treatment and going to multiple doctor's appointments, impacts their life in

many ways. For example, some people say they end up missing several days at work or school and have trouble keeping track of all their different appointments. How would you say managing Fabry disease impacts your life?

CLINIC EXPERIENCE

In a Multidisciplinary Clinic (MDC) you have appointments in the same location with many healthcare providers who are specially trained in Fabry diagnoses/management. They work together, across many specialties, to develop a unique and comprehensive plan of care to address all of your healthcare needs.

Generally includes: Genetics, Cardiology, Nephrology

Do you currently receive your Fabry care/management at a multidisciplinary clinic?

A. YES, currently being seen at a multidisciplinary clinic

(Keep in mind, does it relate to ERT?)

We will start by going over some basic information:

1. Have you always been seen at an MDC?
2. What specialists do you see regularly during your MDC visits?

Geneticist Cardiologist Nephrologist Ophthalmologist

Neurologist Psychologist Gastroenterologist Pain specialist

Genetic Counselor Social Worker Other

- a. What specialist have you seen in the MDC, but not on a regular basis?
- b. Are there other specialists that you are aware of that are part of the clinic, but that you have no indication to see? *For example, you know there is a pain specialist available at the clinic but you don't experience any Fabry related pain so you have never had a consultation with that provider.*

3. Walk me through a typical day at the MDC.
4. Does your primary care provider correspond with your MDC providers?
 - a. Are they under the same health system?
5. Do you have specialists, such as a cardiologist/nephrologist, that you see outside of MDC that you see on a regular basis?
6. How long are your MDC visits?
Half day Full day More than one day
7. How often are you seen at the MDC?
Once a mo Every 3 mo Every 6mo/twice a yr Once a yr
8. Approximately how many miles is the MDC from your home?

Now let's explore what the quality of your care looks like to you.

9. Do you feel that your providers are knowledgeable about Fabry disease and its management?
10. What are some of the advantages that you've perceived from being seen in an MDC setting? *Do you feel you are getting better overall care? Does it have any psychological benefits for you? Do you feel more vested in taking better care of yourself?*
11. What are some challenges that you've experienced from being seen in an MDC setting?
What is inconvenient about it?
Others have noted these challenges, do any of these resonate with you?

*insurance issues distance to clinic is too far travel expenses
visit are too time consuming visit are not as frequent as you would like
Specialist that they want is not available in the MDC Do not like the
specialist(s) that are part of the MDC do not feel their opinion is taken into
consideration by MDC team*

12. Some providers think that patients would find it too overwhelming to see multiple providers in one day. Do you agree with this?
13. Tell me a little bit about your thoughts regarding the quality of your MDC experience?
14. If an individual with Fabry asked you if you would recommend MDC, what would you say?

If previously managed in non-MDC:

15. When you were previously being seen in a non-MDC setting, what kind of experience was that like?
 - a. By comparing the two models, what has particularly stood out to you about either care model that has had an impact on your preferences?
 - b. Which model do you feel has provided you with better care?
 - c. Do you find that you are adhering better to management recommendations?
16. Are there any extra services you would like to see added to the MDC?

B. NO, not currently being seen at an MDC, but I have in the past

(Keep in mind, Does it relate to ERT?)

We will start by going over some basic information:

1. What specialist are you followed by?
 Geneticist *Cardiologist* *Nephrologist* *Ophthalmologist*
 Neurologist *Gastroenterologist* *Pain specialist* *Psychologist*
 Genetic Counselor *Social Worker*
2. Tell me about how often do you see each of these providers?
3. Are they all under the same health system?
4. Is communication seamless between providers?
5. Do you feel that your current providers are knowledgeable about Fabry disease and its management?
6. Do you get annual comprehensive evaluations?
 - a. Who coordinated that, and who coordinates all your appointments?

Now moving on to your past MDC experience:

7. What specialists did you see during your MDC visits?
 Geneticist *Cardiologist* *Nephrologist* *Ophthalmologist*
 Neurologist *Gastroenterologist* *Pain specialist* *Psychologist*
 Genetic Counselor *Social Worker*
8. Walk me through a typical day at the MDC.
9. How long were your MDC visits?
 Half day *Full day* *More than one day*
10. How often were you seen at the MDC?
 Once a mo *Every 3 mo* *Every 6mo/twice a yr* *Once a yr*
11. How many miles was the MDC from your home?
12. By comparing the two models, MDC vs current method of care, can you tell me about your preferences?
13. What were some of the positive aspects of being seen in an MDC setting?

14. What are some challenges that you experienced from being seen in an MDC setting?

Others have noted these challenges, do any of these resonate with you?

insurance issues distance to clinic was too far travel expenses

visit were too time consuming visit were not as frequent as you would have like

Did not like the specialist(s) that were part of the MDC Specialist that you wanted was not available I did not feel my opinion was taken into consideration by MDC team

15. Did you find that seeing multiple providers in one day was overwhelming?

16. What is the main reason you are no longer being seen at an MDC?

a. If that changed, would you be willing to go back to an MDC setting?

17. Extra services you would have liked to have seen/things that could have been improved in MDC?

18. Do you feel you are getting better care at the non-MDC setting?

19. If an individual with Fabry asked you if you would recommend MDC, what would you say?

C. No, never seen in an MDC

We will start by going over some basic information:

1. What specialist are you followed by?

Geneticist Cardiologist Nephrologist Ophthalmologist

Neurologist Gastroenterologist Pain specialist Psychologist

Genetic Counselor Social Worker

2. Tell me about how often do you see each of these providers?

3. Are they all under the same health system?

4. Is communication seamless between providers?

5. Do you feel that your current providers are knowledgeable about Fabry disease and its management? Do you know if they see other patients with Fabry disease?
6. Do you get annual comprehensive evaluations?
 - a. Who coordinated that, and who coordinates all your appointments?
7. Do you feel like you are getting good care? In terms of Fabry disease?

Now I will ask a little bit about MDC:

8. What is your current knowledge of MDC?

Do you know someone who is being seen in an MDC or have you thought about it before?
9. What are some of the advantages you could see to an MDC?
10. What are some possible down sides/disadvantages you could see to an MDC?

Others have noted these challenges, do any of these resonate with you?

distance to clinic is too far travel expenses visit are too time consuming visit are not as frequent as you would like Do not like the specialist(s) that are part of the MDC Specialist that you are interested in seeing is not part of MDC team
11. Do you think it would be overwhelming to see multiple providers in one day?
12. Are you interested in being seen at an MDC? *If that were a possibility.*

YES

- a. What is the main reason you are interested? *What peaked your interest?*
- b. What stops you from visiting a multidisciplinary clinic?
- c. Is there anything you would change about your current method of care?

That if it was done in a different way would make life easier for you?

No

- d. What is the main reason why you are not interested?

- e. Are you satisfied with your current method of care?
- f. Can you see any reason you would be interested? Is there any aspect of it that catches your attention?
- g. What would you change to improve your current method of care?

CONCLUSION: Thank you for taking the time to talk to me about your experiences and opinions. I myself am a Fabry patient and I am very interested in optimizing Fabry patient care. The results of this study will be made available to FSIG and FDSG. Do you have any questions?

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