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Needs Assessment for a Web-Based Support Resource for Patients with a Pathogenic Variant in *LMNA*

Dylan M. Allen
University of South Florida

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Needs Assessment for a Web-Based Support Resource for Patients with a Pathogenic Variant in

LMNA

by

Dylan M. Allen

A thesis submitted in partial fulfillment
of the requirements for the degree of
Master of Science
with a concentration in Genetic Counseling
Department of Global and Planetary Health
College of Public Health
University of South Florida

Major Professor: Deborah Cragun, Ph.D., M.S.
Kathleen Pope, M.D.
Thomas McDonald, M.D.

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ABSTRACT

Pathogenic variants (PV) in the gene *LMNA* cause autosomal dominant inherited “laminopathies” that can affect multiple different organs, most specifically the heart. Current resources for LMNA patients are sparse and disjointed, leaving a need for a comprehensive resource catering to the wants and needs of the patients. A needs assessment of *LMNA* support resources was completed through reviewing published literature and existing support resources and conducting 11 semi-structured interviews with individuals who have a PV in *LMNA*. The Social Support Theoretical Model and thematic analysis of interview transcripts were used to identify discrepancies between the support that affected individuals *receive* and their *desired* support. Key support elements desired by affected patients were: relevant educational resources on *LMNA* explained in layman’s terms, a list of specialists throughout the US and world who have experience treating patients with PVs in *LMNA*, resources to help aid a patient in managing their own healthcare plan with their provider(s), and others. By identifying gaps between desired and received support from the *LMNA* resource landscape, we were able to identify which aspects are most beneficial and most feasible to provide in a new web-based support resource for individuals with a pathogenic variant in *LMNA*.

CHAPTER 1: INTRODUCTION

The total number of people affected by any rare disease accounts for nearly 6% of the world's population (Crowe et al., 2020). Around 80% of all rare diseases are caused by a known genetic condition, and a lack of understanding of genetic disease etiology in the medical field poses a significant barrier to diagnosis and care (Ferreira, 2019). In many cases, those suffering from a rare disease will spend many years undiagnosed unless they happen to meet with a physician who has specific knowledge of their rare disease (Graper & Schilsky, 2017). In the event that a patient is finally diagnosed, having a rare disease can be extremely isolating and confusing. Support groups represent an integral building block in helping reduce the negative effects of a rare disease both physically and mentally (Kisling & Das, 2021). Support groups not only foster an environment where people can share stories, but also coping strategies, educational resources, and information, all without the presence of time constraints that physicians and healthcare providers are often placed under (Southall et al., 2019).

Something that has been shown to influence an individual's ability to cope with the daily and lifelong adversities of living with a chronic illness is social support (Schwarzer & Buchwald, 2004). The concept of social support was first introduced into the social psychology sphere in the 1970s, and although it has been studied and developed throughout the proceeding decades, there is yet to be a consensus on the exact definition of social support (Mao et al, 2018). A social support system can consist of specific social ties such as friends and family, but it can also refer to an individual having access to free services and education. Individuals with a social support

system generally have access to various types support that can consist of: informational support, emotional support, and material support. Informational support can be described as resources that promote education, provide guidance, or give advice on relevant topics to the individual. Emotional support can be described as any sort of emotional aid as perceived by the individual. Material support can be described as any sort of tangible support such as training or money (Mao et al, 2018). Research has shown overwhelmingly that social support can affect both physical as well as psychological health, by providing the individual with tools to better cope and manage daily and lifelong stressors, whatever they may be (Schwarzer & Buchwald, 2004). Although there has been considerable research conducted showcasing that rare disease support groups are a rich source of emotional and practical informative support for patients across many rare diseases, there is a clear gap in existing knowledge on the specific needs of patients with rare, variably expressive, highly (but incompletely) penetrant genetic conditions. The population affected by a pathogenic variant (PV) in the gene *LMNA* is a perfect example of a group that would benefit from an increase in available support resources.

PVs in the gene *LMNA* cause autosomal dominant inherited “laminopathies” associated with an array of phenotypes that can affect multiple different tissues and organs, most specifically the heart (Crasto et al., 2020). This genetic condition is variably expressive, meaning that different individuals can display a range of different phenotypes as well as having differing severity of these phenotypes across individuals with the same genetic condition. Penetrance refers to what proportion of individuals with a genetic condition (or individuals with a specific PV) will develop symptoms associated with that condition (Coll et al., 2017). There are 15 different syndromes currently described under *LMNA*-related disorders, and they span five different phenotypic groups. The groups consist of: cardiomyopathies, muscular dystrophies,

neuropathies, lipodystrophies, and progeria syndromes (Atalaia et al, 2021). Heart manifestations can include progressive dilated cardiomyopathy (DCM), supraventricular and ventricular arrhythmias, and even sudden cardiac death (Hasselberg et al., 2018). PVs in *LMNA* are currently estimated to make up 4-6% of cases of familial DCM. To meet clinical criteria for a diagnosis of familial DCM, there must either be two family members diagnosed with DCM, or a proband diagnosed with DCM who has at least one relative who has died of sudden cardiac death (Rosenbaum et al., 2020). In addition to DCM being variably expressive, it also shows incomplete penetrance. Penetrance of DCM is age-related, with a majority of individuals presenting with symptoms in the third and fourth decades. Symptoms can present as early as early adulthood, and it is currently estimated that about 5-10% of people with a PV in *LMNA* will never show symptoms (Crasto et al., 2020; Hershberger & Morales, 2016). Due to the variable expressivity and incomplete penetrance of this disease, diagnosis can be difficult or delayed until sudden cardiac death presents within the family. Unfortunately, sudden cardiac death may be the initial presenting symptoms in an individual living with cardiomyopathy (Coll et al., 2017).

Treatment recommendations for individuals with PVs in *LMNA* are still largely limited at this time (Atalaia et al., 2021). Considering laminopathies that affect heart muscle come with risks of arrhythmia and sudden cardiac death, current literature agrees that there is a need for an implantable cardioverter-defibrillator (ICD) for individuals with a PV in *LMNA*. There is currently no specific treatment for the types of muscular dystrophy (MD) associated with PVs in *LMNA* (Autosomal dominant Emery-Dreifuss MD, and Limb-Girdle MD type 1B)(Granger et al., 2011).

Affected individuals may represent an educationally and psychosocially underserved group that could benefit from alternative sources of information and support that they may not

receive through the traditional medical sphere. Moderated support resources for these patients could foster an environment where patients and families can share stories, coping strategies, resources, and information, without the typical time constraints physicians and healthcare providers are often placed under (Southall et al., 2019). Current resources for patients with PVs in *LMNA* are limited to unregulated Facebook groups and a network web-initiative of “The *LMNA* Related Cardiac Disease Information and Network” based in The Netherlands that is focused on research and development. Resources specifically for patients with a PV in *LMNA* are sparse and disjointed, leaving a need for a comprehensive resource providing numerous forms of educational information for patients and providers, as well as psychosocial support catering to the wants and needs of the patients. We conducted a needs assessment by reviewing currently available resources and conducting interviews with affected individuals to further identify their specific educational and support needs. Identifying the gap in available resources, followed by analyzing the benefit and feasibility of desired support elements, will help us to determine what we can provide with our novel web-based support resource.

CHAPTER 2: METHODS

The IRB at the University of South Florida reviewed the proposed methods and agreed that this was an educational needs assessment and did not constitute human subjects research as defined by FDA and DHHS regulations.

Participant Recruitment and Interview Procedures

Interview participants were recruited through one of two different convenience sample populations. The first population consisted of members of the private Facebook group “Laminopathies Support Group (mutations in the LMNA gene)”. After getting permission from the group, the first author used a recruitment post on their Facebook page inviting individuals to provide feedback about existing and desired resources or support needs. The second population sampled was from a clinic of patients seen by a cardiologist who specializes in treating individuals with familial cardiomyopathy. The cardiologist provided information about the needs assessment to individuals who met inclusion criteria within his clinic. Inclusion criteria for all participants were: individuals who report having a PV or a likely pathogenic variant (LPV) in *LMNA*, who are 18 years of age or older, speak English, and live in the U.S. Interviews were conducted and recorded virtually using Microsoft Teams. Prior to recording, participants were informed about the study and agreed to participate and be recorded. Interviews were transcribed using Otter.ai, a speech to text software that utilizes artificial intelligence to transcribe, followed by a final review and minor editing to ensure accuracy done by D.A.

Resource Review

The first author scanned the LMNA support resource landscape using three sources of information. The first source was a patient-focused online search for information and community-centered resources for individuals with PVs in *LMNA* using the term “LMNA support resource” on Google to mimic what patients may do when looking for relevant support resources. The second source was a targeted literature search of peer-reviewed articles assessing current known information on *LMNA* symptoms, treatments, and in one case, psychosocial concerns of individuals with DCM. The third and final source was the collection of semi-structured interview transcripts from participants.

Interview Guide

A semi-structured interview guide was designed using a modified version of the Social Support Theoretical framework to identify gaps in support resources for individuals with a PV in *LMNA*. Questions were split into two main groups. The first group of questions on the guide were focused on the participant’s own experience with available information and support resources they are aware of and have used. The second group of questions was focused on support resource desires, needs, and preferences.

Qualitative Data Analysis

Interview transcripts were analyzed using the qualitative data analysis software “NVIVO”. Interview transcripts were first reviewed to extract lists of current and desired support resources. All identified support resources were placed into one of three categories: informational support, emotional support, or both informational and emotional support. Our modified Social Support model, shown in figure 1, helped organize and identify discrepancies between the support elements that individuals desire and the actual support elements that have

been received by affected individuals. Support elements in each of the three groups were reviewed and aligned in order to identify gaps between desired and received support from the *LMNA* resource landscape. Additionally, in order to determine which resources are most beneficial and most feasible to provide in a new web-based support resource for individuals with a pathogenic variant in *LMNA*, data were further categorized into which gaps and issues could likely be resolved through the creation of a web-based support resource, versus what issues will remain for this population moving forward. Interviews were conducted, transcribed, and coded on a rolling basis to ensure emerging or evolving themes were documented and explored in subsequent interviews. This iterative process helped to clarify any ambiguity and to ensure saturation was reached, meaning that no new resources and ideas were coming up in the final couple of interviews.

After coding content related to resources that participants have accessed and desired, thematic analysis of interview transcripts was completed to better understand common experiences individuals spontaneously shared about living with a PV in *LMNA*.

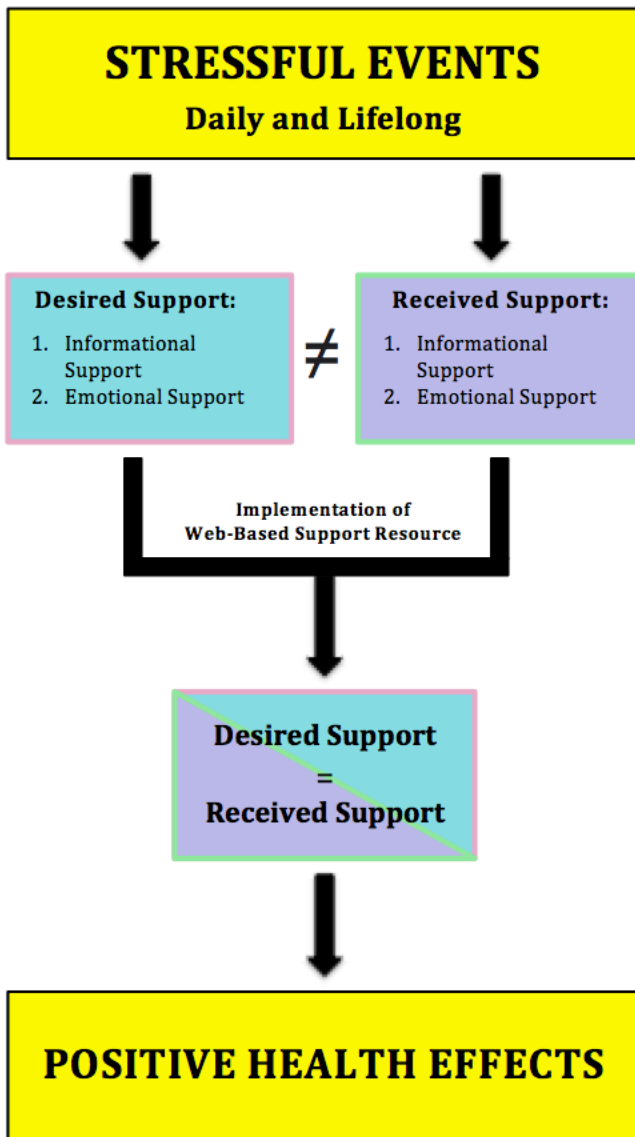


Figure 1. Modified Social Support Model Used for Data Categorization

CHAPTER 3: RESULTS

Demographic Data

Characteristics of interview participants are presented in Table 1. In total we interviewed 11 individuals, with 10 of them being women and only 1 being male. 9 of the interview participants came from the Facebook group, and the other 2 participants came from the clinic. The ages of the individuals ranged from 31 to 67 years. The reported phenotypes from participants included: dilated cardiomyopathy, muscular dystrophy, familial partial lipodystrophy, and asymptomatic.

Table 1. Characteristics of the 11 interview participants

		# Participants (%)
Gender	Male	1 (9.1%)
	Female	10 (90.9%)
Age	30-39	2 (18.2%)
	40-49	5 (45.4%)
	50-59	2 (18.2%)
	60-69	2 (18.2%)
Reported Phenotypes ^a	Dilated Cardiomyopathy	6 (54.5%)
	Muscular Dystrophy	2 (18.2%)
	Familial partial lipodystrophy	1 (9.1%)
	Asymptomatic	3 (27.3%)

^aThis category adds up to more than 11 because one individual experienced two different phenotypes, which is possible due to variable expressivity across the affected population.

Thematic Analysis

Results of our thematic analysis are presented first because these results provide context and rationale for many of the desired support elements identified. Thematic analysis of interview

transcripts identified 8 specific themes representing the lived experience of individuals with PVs in *LMNA*.

Theme 1. Participants were often their own advocate

This theme captures the experience of affected individuals taking on the role of care coordination for themselves, with some describing how they had to convince providers of the legitimacy of their health risks. Individuals described experiences where their health concerns were dismissed by their providers, finding their own specialists proved to be difficult, and the feeling that, without being their own advocate, they would possibly be deceased at this point in time. The difficulty of achieving self-advocacy in this domain is furthered by limitations in available treatment of certain *LMNA*-related disorders, as well as a general lack of knowledge of specific cardiac risks associated with PVs in *LMNA*.

There were multiple participants who expressed worry over how other people affected with this condition, but with less self-efficacy or less ability to advocate for themselves, would manage without necessary support. Having this diagnosis seems to demand the patient to be extremely active in their own healthcare, in many ways to the point of taking on roles outside of what is expected of an average patient. An overwhelming majority of the participants interviewed reported doing a lot of personal research about their disease on their own, often times due to the lack of knowledge or instruction given by their current providers. In some cases, the participants themselves were the first with their diagnosis to present for any of their providers, and these affected individuals often played the role of teacher during a medical appointment rather than patient. “And I remember he [the cardiologist] was like, thank you for teaching me something, which is like, very sweet, but at the same time, like, that's what it is” [P4].

One participant described how a cardiologist they met with to discuss care significantly downplayed their risks regarding their diagnosis. “And his [the cardiologist] answers were like this:

‘Well, sure, I see you have that gene. But you know, that doesn't guarantee everything in your future either. So just because you have that doesn't mean, you know, you're gonna get it’. I 100% do not disagree with that, but there's an extremely *higher* likelihood that I *am* going to get it. I mean, I do keep the window of opportunity of not becoming what my dad's situation is. But still, it was, it was just kind of like pushed to the side” [P5].

This prompted the individual to continue to seek care elsewhere, despite having done extensive research to find their current provider. This individual also has done considerable background education on this condition and had already known the penetrance of this condition approaches 90-95% by the seventh decade, and was able to trust her own knowledge over her provider.

Other less informed individuals who trust their providers to be the expert may receive suboptimal care as a result.

Theme 2. Barriers to appropriate clinical care

This theme encompasses a large variety of participant experiences demonstrating a barrier that leads to a lack of appropriate clinical care and in some cases neglect, harm, or death. Many participants encountered a lack of knowledge of *LMNA* within the medical sphere, including the specialties most necessary for treatment of this condition such as cardiologists and electrophysiologists. “I would say that 99.9% of cardiologists out there have no idea what the *LMNA* gene does, like they just don't know. And it's not to their fault. It's just it's a very highly specialized situation” [P1].

Some individuals even felt that their care was directly affected as a result of their doctors being uninformed. “If he’d [the cardiologist] had called in one of the younger genetic specialists in their cardiology group, [participant’s son] wouldn't have had to have the second stroke [because he would have received his *LMNA* diagnosis sooner]. He’d have never had that prolonged one hour and 17 minute VTAC episode. And we don't know how much that made the game worse than it had to be” [P3]. Some participants shared personal experiences that described feeling abandoned after being given their diagnosis, and difficulty finding providers that provided adequate care. “Yeah, um, my cardiologist just retired, which bugs me out, because I fired four to find him” [P4].

Participants also expressed how it was difficult to find a provider that would meet the demands of a patient with a complex condition like this. “My heart failure doctor that I had just left the hospital that I go to. He was, like, ‘Oh, yeah, you're gonna need a heart transplant someday probably. And I'm going to be there to advocate for you’... and then he left. But I really appreciated that in him. Because I hadn't found that in any of my other cardiologists” [P9].

Multiple participants expressed that, when they do find a doctor with knowledge of *LMNA*, they have run into differences in medical opinions regarding the course of treatment. “You know, my sister got two opinions, when they wanted to put in her pacemaker and ICD. One cardiology group said ‘we don't think you should get a pacemaker and ICD’, with the understanding that she has this *LMNA* gene, but they said, ‘you know what, it's non-conclusive’...these doctors have to get on the same page to have those treatment protocols sort of refined” [P6].

This same participant reported that they had been denied disability benefits for their muscular dystrophy symptoms with the assumption that they were not “sick enough”. “I've recently

applied for short-term disability, mostly for the anxiety piece. And my psychiatrist and I are trying to figure out the pain issue, you know, so being away from work has helped me sort of identify when does the pain start? What am I doing? What can I do to stop it? You know, can I, can I do a meditation practice that helps relieve that? And you know, the unfortunate piece is I have to have the time to do these things, and working full time doesn't allow that, but they've denied my short-term disability. And with, with sort of the saying of, well, 'you're not really sick enough'" [P6].

Another participant explained how, due to lack of proper medical guidance, she was not approved for an ICD until after her sister succumbed to sudden cardiac death. "I wasn't able to get my ICD, even though my sister and brother and my mom had been shocked on their ICDs appropriately. I didn't qualify for my ICD until my oldest sister, who didn't qualify for an ICD, she only had a pacemaker, died of sudden cardiac death. And then they're like, 'Oh, okay, well, you qualify now'" [P10].

Theme 3. Participants have experienced systemic healthcare barriers

This theme reflects numerous systemic barriers to appropriate clinical care that individuals have experienced. This includes experiences of bias such as sexism against women, by way of healthcare providers. Multiple female participants talked about experiences that could be taken as indirect or, in some cases, blatant sexism. "And I think also being a woman in my, at the time, late 30s going on now my early 40s and trying to do really great in my career and being driven that it can be just, 'Hey, she's just a neurotic, white woman that's needs some, you know, anti-anxiety or stress reduction in her life.' And so it's kind of seemed like a mental game like, Oh, it's my fault that I have heart palpitations or anything that I'm feeling. Oh, there must be just

because she's, you know, really driven, and needs to relax or something like that. So those have been my overall experiences.” [P5].

Two participants reported incidences of providers telling them that they need to lose weight as their primary mode of treatment, without really taking into account the their genetic diagnoses and the implications of that. “And after his second angiogram, this [healthcare provider] who had never mentioned really [my son’s] weight, because it's not really a factor, because he doesn't have any of the health conditions that being overweight causes. So he came in, in the recovery area and said, ‘Well, no, that was all normal. So I guess you just need to go home and try and lose some weight, like we talked about’” [P3].

Theme 4. Participants were often information-seeking individuals

This theme was discovered through identifying that a majority of the participants expressed that they want to know as much information as possible regarding this condition and everything it affects. However, a few participants also recognized that this need for information does not reflect every individual with this condition. “Yeah, I think this is the thing that is like, so personal to people with like the struggle of like, is more information helpful or not? And for me, I'm like, I want to know, I want to know, all of the things. But for some people, they don't want to know” [P11].

It was also common for participants to have a previous background in healthcare and/or science, which seemed to contribute to their desire for more information. “I want to know, all the, you know, I want the details because that's my background...but my perspective, I think is somewhat different than just your regular new patient that hasn't that, you know, that doesn't know healthcare” [P3].

Theme 5. LMNA can affect all aspects of life

This theme describes how living with this genetic condition affects not only the person with the PV but also their family and loved ones. It was clear from the first interview onward that this diagnosis is not a diagnosis solely for the individual, but for the support network around them. Symptoms can also impede one's ability to work full time or increase the risks associated with being physically active. "It absolutely affects my wife, our children, everything that we do. It affects my work, it affects... I mean, it's, it's, it's really intense, and it's really stressful. And it's really sad. And we have not figured out how to deal with it well" [P10].

Living with this condition can also affect one's mental health, be it through a fear of sudden cardiac death, or anxiety over the uncertainty of their future health. "It's actually very, been very traumatic for me. And I work with psychiatry to try to not panic that I have this. It's been interruptive in my life only recently, as my mother's condition is failing. My sister's condition is also getting worse. And I certainly worry about myself and what that looks like" [P6].

Theme 6. Living with this condition causes feelings of uncertainty and fear

This theme recounts the common thread of uncertainty over the future as a main driver of anxiety and fear in individuals living with this condition. The uncertainty is the result of two main factors. The first factor is the variable expressivity of the phenotype for this condition, even within families harboring the same PV. "I think the fact that it presents so differently, even within families, is really hard for people to grapple with. Because you just don't know how it's gonna affect your family" [P2]. The second factor is fear due to the current lack of a cure for this genetic condition. Most of the time, this fear actually stemmed from worry due to risks to other loved ones such as children. "You know, I'm too old to die young. And so I don't ever worry

about myself at all. The only thing I worry about is that something happens to me, and I'm not there to help him [participant's son]" [P3].

Theme 7. Participants often have difficulty with scientific jargon

This theme represents the finding that many individuals living with this condition are often presented with (or find) educational resources that are not presented in accessible, plain language. Some of these resources include the test reports that they are given at the time of their diagnosis, webinars presented by *LMNA* researchers, clinical trial information, new scientific literature on *LMNA*-related diseases, and laws that protect individuals from discrimination in regards to genetic test results. "And of course, I do not have a science degree. So language wise, is a challenge sometimes, you know, like, I literally look at it with a dictionary a lot and like, look up words at the same time, so that I can kind of process the information a little bit more" [P1].

Even participants who have a background in science or healthcare expressed difficulty taking in and understanding some informative resources about *LMNA*, usually because they were written or presented for an audience of a higher scientific literacy (or educational background in genetics and/or *LMNA*). "It is pretty heady stuff... And, you know, for me with, after 17 years of this, you pick up a lot of those words. I don't know some of the jargon in the genetics part. But like, it kind of makes sense. But to a newer patient, or somebody who doesn't have a science background, like myself, it would be difficult for them" [P8].

Theme 8. Participants seek out alternative treatment

Individuals with PVs in *LMNA* often rely on trying alternative treatment modalities and therapies to help keep their heart and/or muscles as healthy as possible in the absence of a known standardized course of treatment. This can range from treatments that are expensive, or even

experimental in nature. One participant's experience highlighted the difficulty of managing their muscular dystrophy symptoms. "I spend a lot of time and money on alternative therapies, treatment modalities, to try to keep my muscles as happy as possible, whatever that means. And it takes up an exquisite amount of money" [P6].

Another participant expressed their jealousy over an individual they know who also has a PV in *LMNA*, whose doctor was a research cardiologist and was prescribing new medication that has had promising results. "Their cardiologist also is a research cardiologist. And it seems like they're just doing their own private "let's try this" kind of thing. Because she's like, "oh, yeah, he's got me on this drug now". And I'm like, Oh, so you guys are just having fun. Like, you're just experimenting. But I feel like that should be an option for anyone who wants it, because I'll try it. I'll be like, Yeah, let's go for it. What's it gonna do, make it worse? You know?" [P4].

Received and desired Support elements identified

Transcripts were reviewed to pull out and categorize the support elements that interview participants have had personal experience with and/or have access to (considered support elements *received*), as well as the support elements that they do not have access to currently but *desire*. The identified support elements fell into three groups: informational support elements, emotional support elements, and support elements that provide both informational and emotional support. The received support elements are shown in figure 2, and the desired support elements are shown in figure 3. In some cases, certain participants desired support elements that other participants have previously received, however a majority of desired support elements represented a clear gap in the current resource landscape. Below we highlight some of the most common support elements desired by participants.

Perhaps the most desired element was that of a list of specialists that have knowledge of or experience treating individuals with PVs in *LMNA*. This stems from the nearly universal experience of the participant's having difficulty finding a care team that was informed enough to be able to help, listen, and advocate for them. "It [a list of specialists] would give patients the ability to identify someone, maybe near them where they could go and get specialized care. And then the other thing would be, could be that if someone can't go to one of those places for specialized care, they can point their provider to this website to show, these, here's a list of providers that you can get in contact with to learn more about this" [P10].

A majority of participants also desired educational resources on a plethora of different topics written in plain language for them, such as a list of possible phenotypes associated with the condition and specifically genotype-phenotype correlations, information on insurance and discrimination, and current research and clinical trial information. However, they also desired educational resources specifically designed to be utilized by (or given to) providers who are treating these individuals. "Anything you can do to train or to inform or alert other doctors, nurses, caregivers of this and the dangers of it would be probably life saving in, in the highest level in terms of what your site, or what you guys could do as a resource [P5]."

Participants also expressed the utility in having a frequently asked questions page discussing common questions that diagnosed patients (or at-risk family members) may have regarding this condition, or even having the option to submit questions to be answered by a medical provider. "I think frequently asked questions would be like the first thing that I would go on this website and read. And I like the idea of, like, a way to submit an ask questions" [P11].

Mental health resources were also desired by many participants, in terms of managing uncertainty and fear over their future health. A few participants expressed their desire to find a

therapist as a means of coping, with the caveat that they want to be treated by a therapist who has experience with individuals with chronic illness. “It’s like you’re in a constant trauma mode, is how my therapist has talked about it. Because you never can resolve this trauma, it’s always going to be ongoing. And that’s what’s different kind of, that’s what chronic illness is, essentially... And that’s a little bit of a different treatment approach. Actually, it’s a lot different of a treatment approach” [P5].

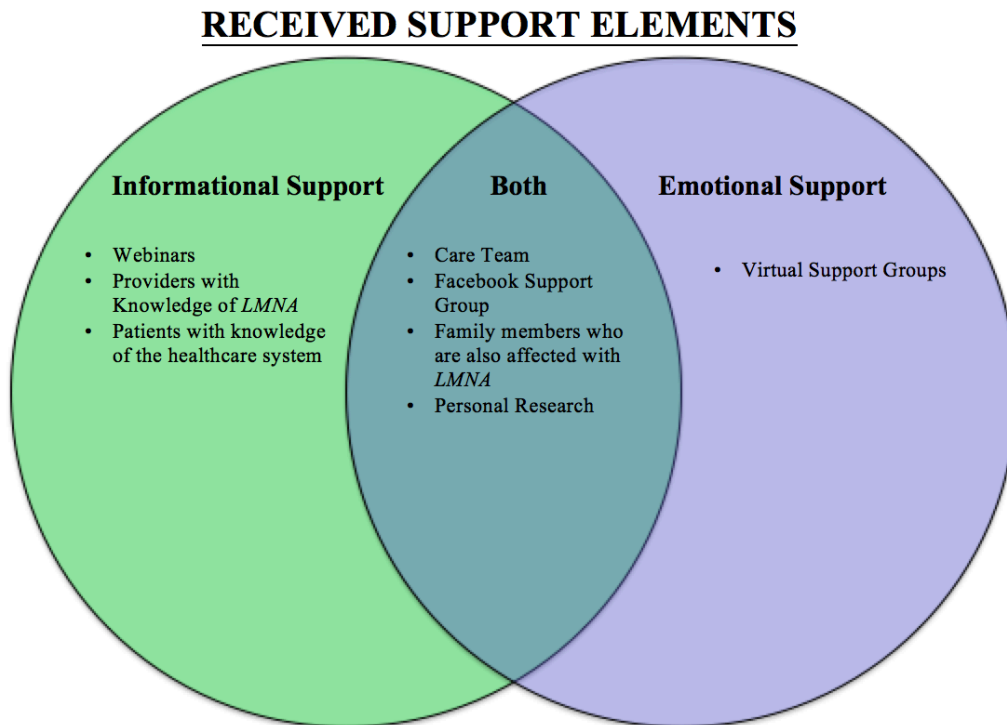


Figure 2. Received support elements identified through analysis of interview transcripts

DESIRED SUPPORT ELEMENTS

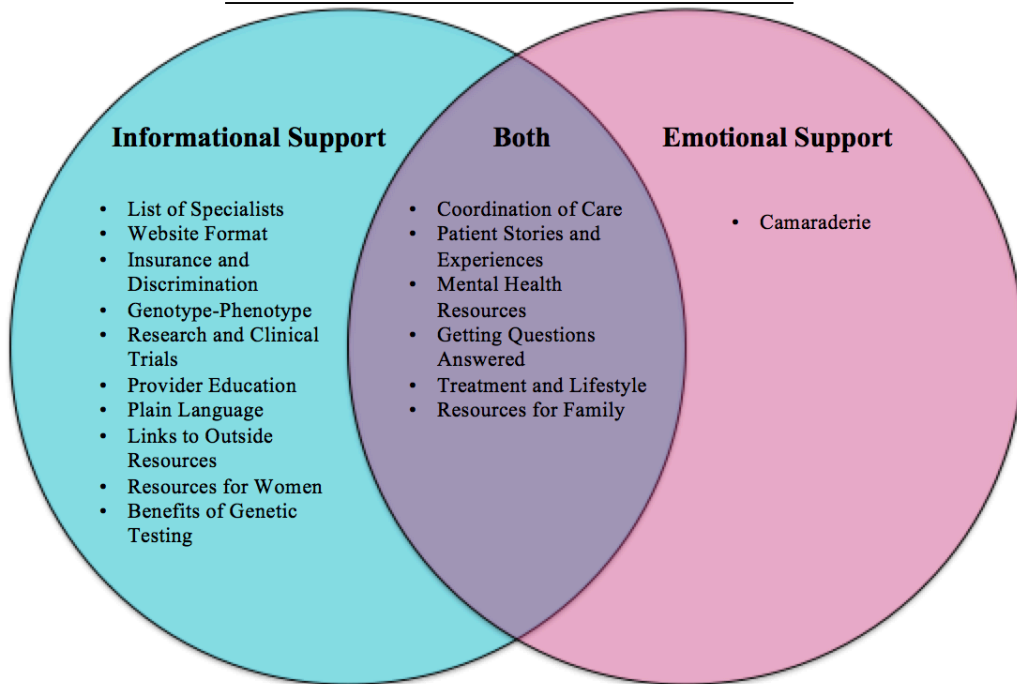


Figure 3. Desired support elements identified through analysis of interview transcripts

CHAPTER 4: DISCUSSION

This needs assessment successfully identified gaps in the resource landscape for individuals with PVs in *LMNA*, and provided context on which desired support elements were of most importance to the individuals interviewed. Taking into account the reported *received* support elements and the *desired* support elements, we have a much clearer understanding of what elements we can provide on our web-based support resource to best fill gaps in the current landscape.

There were a few limitations of this needs assessment that we have considered when looking at our findings. First, our sample size was limited to 11 individuals obtained through convenience sampling. The first author felt confident that saturation was reached with regard to the main purpose of this study because there were no new support resources mentioned in the last couple of interviews that had not already been mentioned. However, the sample of people willing to be interviewed were primarily female and well educated; thus they may not reflect the desired support elements of all individuals with a PV in *LMNA*. Finally, the first author analyzed the interview transcripts on his own without a second, independent coder. However, themes and illustrative quotes were reviewed by another researcher who agreed with the themes and categorization of resources that had been accessed or desired.

We identified many informational and social support needs that our planned website can fill, but there are other gaps in the resource landscape that cannot be filled through the creation of this web-based resource. For example, many individuals desired resources on known genotype-

phenotype correlations, however some identical *LMNA* variants can show varied phenotypic expression, which could be due to epigenetic factors, differences in expression levels, or other factors that current research is not robust enough to account for (Atalaia et al., 2021). Our web-based resource will also provide the most current recommendations for treatment, but the scope of treatment across known phenotypes of this condition is severely limited, and will require continuing research efforts.

Due to the convenience sampling done for this needs assessment, a majority of our participants were naturally advocates for themselves including when managing their own healthcare, and they seem to represent affected individuals who are highly informed and display high self-efficacy. Participants expressed concerns that individuals who are less informed or perhaps less able to make healthcare decisions for themselves, many individuals may end up receiving suboptimal care with this diagnosis, and there were a couple who shared stories of physical harm or psychological distress due to uninformed providers. Our web-based support resource could be extremely beneficial to individuals by providing a list of specialists that have experience treating individuals with *LMNA*, to create less barriers or steps for a patient to receive quality care. This specific support element was unanimously supported and became one of the most desired support elements identified overall. Participants also saw benefit in our web-based support resource containing two separate tiers of information: one tier of information geared towards educating patients with a lower scientific literacy, and a second tier that provides education geared towards providers. Not only would individuals living with this condition benefit from having access to resources that could explain complex yet important topics concerning their diagnosis and care, but also by having the option for their providers to learn using this resource as opposed to learning from the patients themselves. This would take the

burden off the patient, because they could give their providers necessary information to tailor their patient's care as best as possible. Although our resource cannot be expected to educate every single provider that will treat an individual with a PV in *LMNA*, it could be an integral starting point in increasing the awareness of this condition.

Multiple individuals expressed the desire of trying to find alternative medicines or new therapies to aid in the clinical course of their respective symptoms. This brought up the clear issue of a lack of continuity in suggested treatment across medical providers, including providers within the same specialty. Although recommendations for disease symptoms seen in *LMNA* are quite limited, and only a select few are based on genotype, it was clear from this needs assessment that even the few known recommended treatments are often not being followed across providers (Atalaia et al., 2021). The lack of knowledge or clarity with medical management highlights the benefit of providing up-to-date research and literature on current recommended treatments for individuals with PVs in *LMNA* on our web-based support resource, including printable resources that patients can give to new providers to read prior to their appointment. Findings from the patient who expends substantial money on alternative treatments has led us to consider having information about some common alternative therapies so that people can also be informed of their limited evidence and make a more informed decision.

REFERENCES

- Atalaia, A., Ben Yaou, R., Wahbi, K., De Sandre-Giovannoli, A., Vigouroux, C., & Bonne, G. (2021). Laminopathies' Treatments Systematic Review: A Contribution Towards a 'Treatabome.' *Journal of Neuromuscular Diseases*, 8(3), 419–439. <https://doi.org/10.3233/JND-200596>
- Coll, M., Pérez-Serra, A., Mates, J., del Olmo, B., Puigmulé, M., Fernandez-Falgueras, A., Iglesias, A., Picó, F., Lopez, L., Brugada, R., & Campuzano, O. (2017). Incomplete Penetrance and Variable Expressivity: Hallmarks in Channelopathies Associated with Sudden Cardiac Death. *Biology*, 7(1), 3. <https://doi.org/10.3390/biology7010003>
- Crasto, S., My, I., & Di Pasquale, E. (2020). The Broad Spectrum of LMNA Cardiac Diseases: From Molecular Mechanisms to Clinical Phenotype. *Frontiers in Physiology*, 11. <https://www.frontiersin.org/article/10.3389/fphys.2020.00761>
- Crowe, A., McAneney, H., Morrison, P. J., Cupples, M. E., & McKnight, A. J. (2020). A quick reference guide for rare disease: Supporting rare disease management in general practice. *British Journal of General Practice*, 70(694), 260–261. <https://doi.org/10.3399/bjgp20X709853>
- Ferreira, C. R. (2019). The burden of rare diseases. *American Journal of Medical Genetics Part A*, 179(6), 885–892. <https://doi.org/10.1002/ajmg.a.61124>

- Granger, B., Gueneau, L., Drouin-Garraud, V., Pedergrana, V., Gagnon, F., Ben Yaou, R., Tezenas du Montcel, S., & Bonne, G. (2011). Modifier locus of the skeletal muscle involvement in Emery–Dreifuss muscular dystrophy. *Human Genetics*, *129*(2), 149–159. <https://doi.org/10.1007/s00439-010-0909-1>
- Grafer, M. L., & Schilsky, M. L. (2017). Patient support groups in the management of Wilson disease. In *Handbook of Clinical Neurology* (Vol. 142, pp. 231–240). Elsevier. <https://doi.org/10.1016/B978-0-444-63625-6.00020-3>
- Hasselberg, N. E., Haland, T. F., Saberniak, J., Brekke, P. H., Berge, K. E., Leren, T. P., Edvardsen, T., & Haugaa, K. H. (2018). Lamin A/C cardiomyopathy: Young onset, high penetrance, and frequent need for heart transplantation. *European Heart Journal*, *39*(10), 853–860. <https://doi.org/10.1093/eurheartj/ehx596>
- Kisling, L. A., & M Das, J. (2021). Prevention Strategies. In *StatPearls*. StatPearls Publishing. <http://www.ncbi.nlm.nih.gov/books/NBK537222/>
- Mao, Z., Lin, X., Tai, X., & Wang, J. (2018). Breastfeeding Supports for Two-Child Professional Women: A Case Study of Beijing, China. *Asian Women*, *34*(2), 111- 134.
- Rosenbaum, A. N., Agre, K. E., & Pereira, N. L. (2020). Genetics of dilated cardiomyopathy: Practical implications for heart failure management. *Nature Reviews Cardiology*, *17*(5), 286–298. <https://doi.org/10.1038/s41569-019-0284-0>
- Schwarzer, C., & Buchwald, P. (2004). Social Support. *Encyclopedia of Applied Psychology*, *3*, 435-441.
- Southall, K., Jennings, M. B., Gagné, J.-P., & Young, J. (2019). Reported benefits of peer support group involvement by adults with hearing loss. *International Journal of Audiology*, *58*(1), 29–36. <https://doi.org/10.1080/14992027.2018.1519604>

APPENDIX 1:

SEMI-STRUCTURED INTERVIEW GUIDE

Thank you so much for agreeing to meet with me. Before we begin, did you get a chance to read the information sheet I sent you about the interview and my goal to collect information that will help me put together a website with resources?

What questions, if any, do you have for me about this? Are you okay with me recording our interview so that I can go back later and make sure that I didn't miss anything we discussed?

Participant experience with available information and support resources

1. What resources and information about LMNA are you aware of *for patients or their family members*?
 - a. Which of these have you found helpful?
 - b. How helpful did you find them?
 - c. What made them helpful or what could make them more helpful?
 - d. What did you think about the content or the format?
 - i. What information or support content was included?
 - ii. What amount of content is just right, too much or too little?
 - iii. How hard or easy was it to understand?
 - iv. Did you like how it was presented? Why or why not?

NOTE: If unaware of resources, ask:

- e. What types of resources would you find helpful if any?
- f. Have you searched for information and resources? Why or why not?
- g. Where did you search or where would you want to access information and resources?

Support needs and preferences

2. What other types of information or resources would you find helpful?
3. Any of the following of interest?
 - a. Additional medical information (about what topics?)
 - b. Patient stories/experiences
 - c. More ideas for how to deal with uncertainty
 - d. Question and answer forum (just for patients? Or moderated by a genetic counselor and other healthcare providers?)
4. What format do you prefer for these resources? (e.g. downloadable/printable handouts, video, website content, etc.)

5. Now I will go through some other specific topics and resources and I would like to know how interested or important these would be for you as well as any other thoughts on how to improve some of the things I will show you):
 - a. Education on life, disability insurance for at-risk family members
 - b. Protections against genetic discrimination (GINA, FL HB 1189, etc.)
 - c. Profiles showing/describing possible phenotypes associated with *LMNA* (heart rhythm disturbance, muscular dystrophy, progeria, etc.)
 - d. Education of inheritance pattern (AD) of *LMNA*
 - e. Find out where (if possible) research is available
 - f. Educational literature for patients to share with other healthcare providers to ensure proper treatment

6. What other elements **that have not been discussed** yet would you find helpful on a web-based support resource?