

Rare World: Towards Technology for Rare Diseases

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ABSTRACT

Researchers have created innovative technological solutions to support people with common chronic illnesses. In this study, we investigate design opportunities for people with rare diseases who are not well studied or have smaller populations to work with, because although an individual's disease may be rare, the number of people living with a rare disease is substantial. We conducted an interview study with 19 individuals with rare diseases from around the world to understand common problems and experiences that could be supported through design. We found that communicating with friends, family, and providers about her disease were challenges for participants. Additionally, participants thought of their disease as being a large part of who they were. We discuss these findings in the context of prior work on common chronic illnesses, addressing the potential relevance of existing technological interventions for people with rare diseases.

Author Keywords

Rare diseases; chronic disease management; health.

ACM Classification Keywords

H.5.m. Information Interfaces and Presentation (e.g. HCI): Miscellaneous

INTRODUCTION

Common chronic illnesses, such as diabetes (e.g. [20]), kidney disease (e.g. [26]), cancer (e.g. [16]), and asthma (e.g. [36]) impact a large and growing subset of our population. Researchers responded with technologies that address the symptoms, causes, and management of chronic illnesses. These chronic illnesses are well studied in the medical literature and have a constrained set of symptoms and treatments to address with technology. To this end, the technologies are customised to a specific chronic illness. The research community has not found an intervention sweet spot that will work for every demographic and disease. Since there is not a one-size-fits-all solution, rare diseases are left out of the health design space.

While an individual's disease may be rare, the number of people living with a rare disease is substantial. It is estimated that 10% of the world's population has a rare disease [32] (in

comparison, only 3% of the world's population use Twitter¹). If everyone with a rare disease lived in the same country, it would be the world's third most populous nation. Since some rare disease symptoms overlap with common chronic illness symptoms, we set out to investigate the common ground between rare disease populations and common chronic illness populations, and what the scope of technology could be for people with rare disease. We conducted interviews with 19 individuals living with a range of chronic rare diseases recruited from online communities. They lived around the world representing a range of health care systems. Our goal was to examine the experiences of people living with rare diseases and to identify common problems that could be addressed through design. Our main contributions are:

1. A profile of people with rare diseases;
2. A discussion of similarities and differences between rare diseases and more common chronic illnesses; and
3. How technologies could address some of these opportunities for design.

RARE DISEASES

Each country defines rare diseases slightly differently. In the US, rare diseases are those affecting less than 200,000 people (or 0.06% of the population). In most of Europe, a rare disease affects no more than 5 out of every 10,000 people (or 0.05% of the population). World wide, 10% of the population, or about 350 million people, are living with a rare disease. There are around 7,000 different rare diseases. [32] For simplicity, we relied on the NIH's list of rare diseases² to limit our study. Rare diseases can be challenging to diagnose — patients usually receive 2-3 misdiagnoses over five years in the UK and over seven years in the US on average before receiving a correct diagnosis [32]. Diagnosis requires visits to many different primary care physicians and specialists.

RELATED WORK

Patients as whole people & experts.

Researchers encourage us to think about people as people, instead of as patients, emphasising humanness over disease [1, 3, 19, 21]. Further, people can have an expertise that is uniquely different from clinicians' expertise [9], one that is gained through lived experiences. Researchers explore what it means to support the whole person through design [11] by acknowledging that people may prioritize life differently from clinical best practices or be unable to integrate best practices into their lives [1]. People sometimes make complex [19] and flexible [21] negotiations of their actions to accommodate their lifestyle. There is also a tension between

¹Twitter Inc. 2Q 2014 Earnings Report. 2014.

²<http://rarediseases.info.nih.gov/gard/categories>

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CHI 2015, April 18 - 23 2015, Seoul, Republic of Korea
Copyright 2015 ACM 978-1-4503-3145-6/15/04...\$15.00
<http://dx.doi.org/10.1145/2702123.2702494>

empowering people and enabling them to make false inferences or poor decisions, and although intelligent data analysis and careful visualisations [21] can support people, there still exists a need for medical professionals in this process [18].

Social Support & Online Communities

There have been several studies on the use of online communities by people with common chronic illnesses [10–12, 27]. These online health communities tend to be disease specific and are separate from existing social media sites like Facebook or Twitter. People view sites like Facebook as too public and do not use them to share health information. Posts on Facebook or Twitter are often presented to “collapsed contexts” [4] where many environments or social groups overlap in the audience, although Pang et al. [24] found that people are hesitant to even use private messaging features to share health information — either because they were afraid they might be publicly visible or because they did not know how to create private messages. Gibson et al. [7] found that private Facebook groups worked well for support between new mothers as a privacy mechanism within an existing platform.

One exception is [31], where Suh et al. designed a Twitter-based intervention for parents to track their children’s milestones, understanding that busy participants are more active on sites that are already part of their regular routine — although some participants did express concerns about sharing their information publicly. Newman et al. [23] found that conflict existed between the benefits people hoped to gain from online communities and their goal of impression management [14]. For instance, needing emotional support conflicted with a desire to have others view their health status favourably. Text message interventions were explored in the context of chronic illnesses [36] as a middle ground between convenience and privacy. Text messages may give greater control over the spread of their information, while still leveraging a medium that is part of their existing routine.

Patient-Centered Information Management

Living with a chronic condition requires a great deal of information management; people with chronic conditions often keep track of their own medical records, collect their own notes and data, and sometimes collect articles related to their condition. Given the amount of information that is managed by the patient, Pratt et al. [25] call for technologies to manage health information that focuses on the patient at the centre of the information repository. Further, Moen et al. [22] outline strategies used to manage this information in the home.

Typically, research into these patient-centred information repositories has been in the domain of Personal Health Records (PHR’s), although these have been discounted in some cases for being designed from an overly “clinical perspective” [1], failing to account for the “unanchored” [16] (i.e. outside a traditional workspace) and “invisible” [34] work performed by patients during the management of a condition. A patient-centred tool that strays from this traditional clinical perspective is My Journey Compass [13], a tablet with a suite of applications, pdf informational resources, and relevant website links tailored to cancer patients.

METHOD

We conducted interviews with people who were living with a rare disease to understand their experiences and to inform possible design opportunities. This study was approved by the Institutional Review Board at Indiana University.

Recruitment

We recruited through online forums specifically targeting people with rare diseases (RareShare³, RareConnect⁴, the Rare Disease Foundation⁵). Each online forum had sub-communities for different diseases. We recruited from any that had been active within the past three months. We also recruited from Facebook groups targeting specific rare diseases. We randomly chose 30 conditions from the NIH list in different categories to target a range of diseases. 15 conditions had Facebook groups from which we were able to recruit.

Participant Information

Eligibility required a chronic rare disease diagnosis, and membership in an online community for that disease. Over 150 people responded to our recruitment notices and we selected 19 participants who represented a range of conditions. Seven participants were male (37%). Participants ranged in age from 20–66 (avg=45, sd=13). Most (53%) were living in the United States (Table 1). All recruitment notices were posted in English. All interviews were conducted in English, except one, which was conducted with the help of a translator to allow the participant to better express himself. We interviewed participants with 13 different conditions (Table 2). Two participants had more than one condition. We identify these participants using M1 and M2. We identify participants having only one condition with S1–S15.

We respected that, especially in the case of rare diseases, it can take a long time to obtain a diagnosis. We included two participants who were undiagnosed because they were active in online rare disease communities and had reported that their doctors agreed that their condition was rare. We identify these participants using U1 and U2.

Procedure

We conducted semi-structured interviews to understand participants’ perceptions of their own lives. Most participants undertook a one-hour video interview (using Skype, Google Hangout or Facetime). Some were uncomfortable using video

³<http://www.rareshare.org/>

⁴<http://www.rareconnect.org/>

⁵<http://www.rarediseasefoundation.org/>

Continent	Country	Health Care System	N
Asia	Pakistan	Mostly private	1
Australia	New Zealand	Mostly public	1
Europe	England	Mostly public	2
	Ireland	Mostly public	1
	Macedonia	Mostly public	1
	Norway	Mostly public	1
North America	Canada	Mostly public	1
	United States	Mostly private	10
South America	Brazil	Mostly public	1

Table 1. Participant Locations

Disease Name	Description	Categories	N
Chiari Malformation	Dizziness, muscle weakness, numbness, vision problems, headache, balance problems	Brain Diseases	1
Ehlers-Danlos Syndrome (EDS)	Symptoms range from mildly loose joints to life-threatening complications	Connective Tissue Diseases	1
Congenital Anosmia	Lifelong inability to smell	Ear, Nose & Throat Diseases	3
Familial Amyloid Polyneuropathy (FAP)	Loss of sensation in the extremities. The autonomic and central nervous systems may be affected.	Nervous System Diseases	1
Hereditary Angioedema (HAE)	Recurrent episodes of severe swelling (e.g. the limbs, face, intestinal tract, and airway)	Immune System Diseases	1
Hereditary Spastic Paraplegia (HSP)	Increased muscle stiffness and weakness of the legs leading to difficulty walking	Musculoskeletal Diseases	1
Inclusion Body Myositis (IBM)	Progressive muscle inflammation and muscle weakness	Musculoskeletal Diseases	4
Kallmann Syndrome (KS)	Delayed puberty, abnormal development of secondary sex characteristics, and infertility	Reproductive Diseases	2
Morgellons	Abnormal skin sensations, co-existing psychiatric conditions, fibres in affected skin areas	Behavioural and Mental Disorders	1
Multifocal Motor Neuropathy (MMN)	Weakness in hands/lower arms, cramping, involuntary contractions/twitches, wasting affected muscles	Nervous System Diseases	2
Ocular Cicatricial Pemphigoid (OCP)	Chronic cicatrizing conjunctivitis. Can also affect skin/mucous membranes	Skin Diseases, Eye Diseases	1
Systemic Capillary Leak Syndrome (SCLP)	Fluid/proteins leak from capillaries resulting in dangerously low blood pressure	Blood Diseases	1
Wilson Disease	Excessive copper accumulation, leading to kidney, brain, and eye damage	Kidney/Urinary Diseases, Digestive Diseases	1

Table 2. Participant Diseases. (Note that two participants had multiple conditions, and two participants were undiagnosed)

conferencing technology, so we conducted six interviews by phone and two via instant message. We began with an informed consent process and preliminary demographic questions. The main interview was divided into four topics: experience living with the condition (When/how did you first start to notice the condition? How did you arrive at your current diagnosis? What does a good/bad day look like? How does it impact your day to day life?), relationships and support (What medical support do you get for the condition? Are you satisfied with it? How does this influence your relationship with friends or family? Do you receive support from elsewhere?), use of technology and information management (What, if anything, do you bring to medical appointments? What do you do with records or information you receive from your doctor? Do you conduct research about your condition? If so, what sources do you use? Do you find them easy to understand?), and sensemaking (How well do you feel you understand your condition? Has that changed since you were diagnosed? What do you do if you have a question or concern about your condition?). We relied on an interview guide to ensure coverage of all relevant areas, but allowed the participants' responses to guide the flow of the interview. After the interviews concluded, three participants emailed additional thoughts, which were added to their transcripts. Quotes from these additions are identified with asterisks in this paper.

Analysis

In addition to the two instant message transcripts, we transcribed the 17 interview recordings. We analysed these documents using inductive qualitative methods [29]. Three authors separately developed a set of codes based on the inter-

view transcripts and discussed them. Each author iterated on these themes several times until converging on a set of common codes. We finalised the codes, and used them to analyse each transcript. Because of the highly sensitive nature of the interviews, all participants were given the chance to review this paper before publication to ensure they were comfortable with the content. None requested changes to this publication.

FINDINGS

Participants' conditions pervaded many aspects of their lives, and having a rare disease put them in a unique position. S13 describes the experience as *"Rare World. It's very difficult. The receptionist doesn't understand, the doctor doesn't understand. No one's heard of it. It's a really strange spot to be in."* In this section, we discuss challenges participants faced in these different aspects of their lives, and strategies they employed to overcome them.

Role As Friends

Participants varied widely in how much they shared with their friends about their experiences. Several spoke of the challenge of communicating their experiences when their diseases were both invisible and unfamiliar to others. S11 said, *"There's nothing to see. Nobody's ever heard of it. I mean, if I said I had Multiple Sclerosis everybody would know what I was talking about and they would know what to expect . . . It's not obvious that there is anything wrong with me at all. So I think that some people think that maybe I'm putting them on a bit."* U1 told people she had a common condition, because it

was easier for them to understand. *“It’s easiest just to say Lupus than anything else because people can wrap their heads around that.”* Some participants had friends who tried to be supportive, but were not helpful. S13 said, *“A lot of friends would be like, ‘Well just call another doctor’, ‘Just make an appointment with another one’, ‘Well, you just go tell them this.’ ‘You tell them you’ve got to do this’. I’m like, ‘You don’t understand, I can’t find anybody else that will take me.’”*

Some participants found that, throughout the process of diagnosis and treatment, they saw who their true friends were, and that some friends handled the change in friendship better than others. S12 said, *“Some want to be there for you, but they are so scared that they don’t even know how to talk to you. Then you find the friends who . . . come out of the woodwork when you don’t know what to do . . . When shit hit the fan, some of the people who you thought would really be there actually weren’t.”* Several admitted to downplaying their illnesses. U1 explained, *“beyond my true true inner circle of friends . . . I don’t tell people. The most you’ll hear from me is ‘I’ve a little bit of arthritis.’”* S3 downplayed her own symptoms, but turned to her friends for additional support after her son was born with the same condition. *“I never complained about it because it was like a part of me and ‘Okay, that’s who I am.’ [My friends] never realised that it can be life-threatening. When I started my fight for my son, and when I called them [for support] they would say ‘We can’t come because we have to go shopping’ and ‘we can’t come because we have to go here and there.’ And then I said, ‘I’m having this life struggle for my baby. You guys are my friends, I expect you there.’ . . . So they were a little slow in understanding how important this was but it’s because I never made a big fuss about it. It’s who I am.”*

Having a rare disease also impacted the experience of forming new relationships. S3 had previously dated people that were uncomfortable with her condition. *“But you have to tell them at the beginning. You can’t go on lying to them and not telling them. When they fall in love, what are you going to do? Say, ‘Okay, now, I was kidding. I have this rare disease.’”* U1 had similar experiences until she met her husband because her condition did not have a clear diagnosis or treatment.

Role As Family Members

Very few participants had friends that they considered really supportive and helpful; they relied primarily on family members for support, since family members were better able to understand what they were experiencing. Although grateful for this support, participants worried about the impact of their conditions on those around them, especially when participants did not have the disease when they first met their partners. S5 explains, *“I feel sorry for [my wife] because she has to take care of a husband who is in much worse shape than her . . . So now she’s had to cope with a husband who’s lost a lot of his abilities and she’s done very well with it . . . I’m sure she doesn’t like it but what are you going to do? I mean, you know, that’s what marriage is about in a lot of ways.”* S6, on the other hand, felt she needed support from her husband, but he was not accepting enough to be helpful. She asked him to read a book about living with her condition, but he told her he did not want to have to think about it. She said, *“Earlier*

I just had to tell him, ‘You can stay in denial all you want to, but when I say I need you to do something . . . you just have to do it.’ And for a long time he used to say things like . . . ‘Do you really need that cane?’ And I said, ‘Yes I do. Stop asking me questions like that.’”

Beyond problems with romantic partners, a genetically transmitted disease added further complications; participants who passed their conditions on to their children faced unusual challenges. Some made great sacrifices to ensure their children received necessary care — even putting their own lives at risk. S3 explained, *“I buy [medicine] for my son because our medical fund . . . does not cover the medicine. So I actually have to smuggle it from other countries. For my son, one shot is around [€600 or \$800] . . . so I have to have three or four vials of the medicine. And I’m keeping it only for my son because I don’t earn that much money that I can have a shot for me. So it’s on destiny whether I will live another day.”*

Role As Advocates and Patients

Participants spoke frequently of needing to advocate for themselves and for others with their conditions. Particularly in countries with predominantly public health systems, participants worked to improve access to medication. S2 described, *“the associations [for rare diseases] are united and are pressuring the government . . . Health is a duty from the government and a citizen’s right, but there are a lot of things that we can’t reach, like expensive medications, so we need to sue for them, and this is a huge frazzle. So, we are trying to approve some laws in the [government] for this kind of medication to start to be available for the population without needing to go through the justice system to win them.”* S3 was similarly engaged in advocacy and awareness campaigns, explaining, *“We can climb mountains. We can have children. We can live 100 lives. We can live 100 years. All we need is medicine.”*

In addition to advocacy to governments and the general public, participants needed to advocate for themselves when dealing with medical professionals. Many told of times they felt their care was poorly handled by physicians who were ill informed about their conditions. *“I just hope and pray that, if I have an attack, what happens at the hospital is that they listen. We did have an incident where . . . they tried to tell me I had a tummy bug and it wasn’t until I actually passed out lying down that then they thought, ‘Okay, we need to start listening to this person.’”* (S14) Such experiences often lead patients to seek replacement doctors. Not all patients had the option to simply switch doctors if they are unhappy, however, because some doctors were not willing to treat unfamiliar diseases, and health insurance or government regulations further limited options. S13 explained, *“I had to suck it up and go back because no one would take me. You’d call them, you’d tell them what you’ve got and they look it up, they’d say, ‘Oh, no, I’m sorry we don’t treat that.’ I heard that over and over and over again. But I had to suck it up and go back, because no one else would take me.”* S6 described how she tried to set the tone with her doctor right from the beginning. *“I just let him know right away that I was going to ask questions, I was going to read, I would come and talk to him about anything I wanted to try. And I think I kind of let him know that I was*

really going to be active in my care and he could either get used to it or I might move on."

Some patients worked to reduce their reliance on doctors. S3 believed that *"Nobody wants to treat you because they know you will probably be the only patient with that rare disease that they are going to meet. So they're not very interested and motivated in working with you."* After she and her son vacationed in a different country where several doctors refused to treat them, she learned to administer shots herself. *"They were all like, 'We don't know what it is. We don't want to take this risk. So go away. We have other problems, other patients.' So I told them, 'Okay, go on Google and Google it.' 'No, no, go to somebody else's door.' So we lost a lot of hours. My son had this vomiting episode and it was very scary to have him in the car and going around ... to find a doctor that is going to be brave enough to give him those shots. And that's when I said 'Okay, I'm going to learn to give myself the shot.'" S3 did eventually find a doctor who was willing to treat her and her son, and like many other participants, spoke highly of doctors who, although unfamiliar with a rare condition, were willing to take the time to learn. She said, *"She had this purple hair and I thought she was a student because she didn't look very official to me. And she came with a cup of coffee and she sat down and she read the leaflet that is in the medicine ... And she said, 'Okay, this is the first time that I am in this situation, but in the next two hours I'm going to learn everything about it.' ... She believed me. She gave him the shot. She kept us there for two hours ... and in the meanwhile she was on Dr. Google and she Googled it."**

While most participants had to advocate for themselves, when U2's doctors said her symptoms were psychosomatic, she had to enlist her friend's and professor's help before her doctor would take her off a medication, *"finally after six months I got my instructor and a friend to write letters, and then they commented changes they had seen in me and then the doctors told me to stop that medicine right away."* For many participants there was tension between advocating for yourself and becoming your own doctor. S13 stated, *"[The doctors] think you're crazy, like 'Oh, you don't know.' And so I think that's probably a key thing right there is you have to have sense enough to not diagnose yourself. There's a line. But you have to have sense enough to know when to keep pushing. 'cause I know some people are like, 'Oh, well I read it on the Internet', and that's it. I don't ever want to be that person, but I do want to be that person who says, 'Well I read this and I thought about this, and I just wanted to run it past you, wanted to know what you thought.'" It is worth noting that this same participant nevertheless maintained a strong distrust of doctors. *"I was going to these doctors that I didn't trust; they didn't act like they knew what they were doing. And I would give them tests. They didn't know it, but I would ask them things like 'How do you know that this is an allergy?', and they'd say 'Well, we don't have really any way of knowing.' 'Yes you do!'" S2 also expressed an attitude of knowing more than his doctor. *"We call it 'medical ego'. Some doctors think that they are superiors or gods, so, they don't admit that the patient knows more than them."* U2 began actively research-**

ing her symptoms after her doctor said her symptoms were all in her head and she wanted to *"prove him wrong."*

Role As Record Keepers

Because people with rare diseases often see numerous specialists, the amount of medical records can quickly multiply. Some participants kept track of their records for their own use. Due to a series of misdiagnoses and conflicts with doctors, U2 asked for personal copies of her records so she could manage them on her own. She used her lab results to research conditions that could lead to a diagnosis. S11 said she kept a *"vague journal"* of significant occurrences (e.g. bad episodes, cramps) because her symptoms slowly progressed. S13 did not methodologically maintain her records, *"but I try to keep up with like when there is a medication change, because they will always ask you when did you start it, how long were you on it, what was your dosage, and you're like 'I don't know, I can't remember what I did yesterday!'"*

For other participants, their records played a greater role in their interactions with physicians. While S6 kept a spiral notebook for personal use, she also found it helpful when she visited her doctor. *"When I first went to see the neurologist, I had this little book with me and he said 'What is that?' And I said, 'Well, this is my little book where I write down what you tell me to do, or changes that we make.' And he said, 'Oh you won't need that. We keep charts of everything and blah blah blah'. Then he started asking me all these questions, 'When did you do this? When did you do that?' So I just kind of whipped my little book out and I said, 'Okay, let me just look at my little book and I'll tell you.' It was really kind of funny."* U1 described her *"medical résumé,"* a technique she picked up from her parents who were both medical doctors. *"When I was a very young child ... [my mother] would just hand that to the doctor. We would go in to see a specialist or an expert and she would just immediately, first thing, hand the résumé to the doctor and say, 'Read this first before we start. You might as well just read this. I'm a medical doctor, here's her medical summary, it's on two pages. Read this first and then we'll start the appointment.' And I saw the power in that ... As I aged, I started maintaining it myself."* She further explained, *"I call it speaking surgeon-ese. The surgeons speak in very bullet point kind of language, so a patient has to get used to boiling things down to one or two sentences — to speaking in surgeon-ese, bullet-ese."*

Similar to a medical résumé, S8 made *"business cards"*. *"I've had these cards made up through the myositis association. It looks like a little gift card. It's got your name on the front ... It tells you what to do about the disease and it's one of the greatest tools to have because when I go somewhere and if something happens to me, I just give them that card. And they've got everything there that they need."* In addition to using these cards to communicate with physicians, S8 uses them to connect with other patients for support. *"I give them to everybody actually. I left a stack of them in the physical therapy department because they had said I was their first patient with this and then I think they had two more that were diagnosed with it. So I left my card there and I was like, 'You know, if they need to talk or whatever, here's the card and I would be happy to talk to them.'"*

Role As Copers

The interview data reflects a wide range of attitudes and emotions; it was common that participants experienced anger and fear, as well as acceptance and even gratitude. Many discussed the depression they experienced, and some strategies they use to stay positive. Several discussed learning to accept their new circumstances. S5 chose to make the most of his time while he was able, since his physical condition was likely to worsen, *“I do a lot of things now. I play golf. I travel quite a bit with my wife because we know that at some point in time I won’t be able to travel so much. So we kind of compress some of the plans that we might have had into a shorter time span because we don’t have as much time as we’d like.”* S12 also talked about learning to accept his condition, *“I do yoga. I meditate. I try to keep myself busy . . . I have realised that for whatever reason, this is a part of my story. I am walking with this. But I can’t let it overcome me. And it is easy to tell you, but not easy to practice.”*

Balancing a positive attitude with the negative emotions was a constant process for participants. *“I have this little mantra that if I’m in pain, I’m alive. So that’s sort of how I try and twist that off, I suppose. I try to be really positive about myself. It’s hard but I have a physical disability now and it does stop me from doing things.”*(S4) Some participants held a sense of optimism, *“It was a big thing to understand and it sounded scary. And now I’ve realised, in me at least, it’s going to be a lot slower progression than I originally thought and that I can keep it at bay a bit with treatment.”* (S11)

Maintaining this optimism was challenging sometimes. S11 said*, *“There does not seem to be any chance that the outlook will change. No one is interested in rare diseases. They are not economically viable . . . I’m not holding my breath. No one seems to know the mechanism for MMN. So I watch my kids. That is the worst thing.”* Participants were concerned about the progression of the condition and the availability of a cure. Even if no cure is possible, some participants hoped there would be more information available, *“I hope that at least . . . they’ll be able to tell me why. So the biggest thing for me isn’t to be able to smell, I just want to know why.”*(S1)

Participants found comfort in knowing they could have worse diseases, *“I always tell people it’s not great having KS, but there’s people with a lot worse”*(M2). S5 and S12 both had conditions that are commonly misdiagnosed as ALS. S5 initially considered ALS to be one of many possible diagnoses, but it was ruled out fairly quickly. He said, *“At that point I immediately felt better because ALS is going to kill you pretty quickly. So there was a certain you know, ‘Wow, goodie! I’m going to make it here for a while!’”* S12 was misdiagnosed with ALS for almost a full year before arriving at his current diagnosis. *“So when they tell you that you have that disease, they are pretty much saying that you’re getting ready to die because most people don’t make it past two years . . . That was a really bad trip. You have to deal with this huge problem all of the sudden. [When I was diagnosed with MMN instead] I was very happy. They pretty much lifted a curse off me. They told me that there’s a chance you’ll live your whole life.”*

Role As Researchers

Participants took an active role in seeking out information about their diseases. Participants relied not only on their peers’ first-hand experiences, but also turned to scientific literature for answers. Participants found academic papers through searches of Google and Google Scholar. Some participants were successful in learning about their conditions through their research. S3 described how, *“Dr. Google helped a lot because I Googled it a lot. Actually it was Google that helped me finding the medicine in the first place because no one in [my country] have ever took care of us or ever told us that actually there is a cure, because it is very expensive.”*

Most participants, however, found that the information they wanted was not available. S11 explained, *“I’ve looked at a lot of things online...there only seems to be a very limited amount of treatments...So there’s not an awful lot to look up really and no one knows what causes it anyways.”* S14 felt that she understood it as well as she could, given that there was little information about it. *“I don’t know what triggers it and they don’t know what triggers it. And they don’t know what stops it and I don’t know what stops it. So what’s there to know?”* U1 also felt that she understood things as well as she could, given the limited information available. *“I don’t believe anybody, even the experts really have a handle on what my disease is, or what exactly is going wrong in my immune system . . . You’re not going to get direct answers to your questions. . . sure, I wish I could go to some magical leprechaun and say okay, sit down with me, here’s pencil and paper, draw for me exactly what’s wrong with my immune system and how I can fix it. You’re not going to get that. You’re not going to get answers to the questions you really want answered.”*

Many participants described themselves as more research literate than the general population. Twelve participants cited a reason for their knowledge or a connection to someone with a medical background - their pharmacist friend, geneticist father, researcher parents, or past career as a phlebotomist. One participant worked as a nurse for many years, while another had two parents who were both medical doctors. Some participants had less directly applicable medical background, but clung to what science background they did have. *“I present professional development to teachers for elementary science, so I know science.”*(S13)

Role As Peer Supporters

Participants used online communities as a way of sharing research findings and trading resources and support. A few participants also participated in in-person groups, or had encountered other people with their diseases at symposia, conferences, or medical associations. Participants actively sought out others with whom to connect, explaining that they felt helped by *“that instant connection, and knowing you’re in company who have gone through the same thing”*(M2)

Online communities were mainly used to ask questions of people with similar experiences and trade tips. Many felt that their peers were a better source of information than their doctors, who did not know or were not prepared to discuss what they wanted. S6 explained, *“None of the doctors really address . . . what you need to manage your day. So you have to*

rely on somebody else who has walked those steps or similar.” Sometimes participants shared tips in the groups, often in response to someone else’s question. S5 explained, *“I will pipe in and say ‘Well here’s what I ran into and I did this about it.’ . . . I had to have a feeding tube and I couldn’t eat. Well, turns out the feeding tube is a pain in the ass because you have to do something with it when you’re not using it. And women end up tucking it in their bras, but men, you know, I ended up clipping it to my shirt . . . And then I saw issues like that on the Facebook thing and I typed in and said ‘Here is what I did. Here is what I learned. You might want to try this.’”*

Participants who connected with others through online communities were unanimously positive about the benefits this provided. Many spoke of how this helped them feel less alone, *“It is amazing to hear other people with the same thing because when I was diagnosed they told me there was only 60 other cases in the whole world. But of course there’s certainly a lot more than that.”* (S14) S5 agreed, *“You sometimes think you’re alone. If you have a problem and you start talking to people who have the same problem or a similar problem then all of a sudden you feel better about it, because all of the sudden you understand you’re not alone.”* S13 even described how excited she was the first time she met someone with the same condition. *“I’m like a child. I’m like ‘I have a friend!’ and she’s the same way . . . She will email me and say, ‘Have you ever had this? Have you ever had that?’”* Since there are a small number of, if any, people with the same disease locally, many people with rare diseases turn to the global community for connections. The international nature of these online communities was both a benefit and a barrier for participants. For S2, language was a barrier — he found it difficult to communicate with other people. For S8 however, the international community was a benefit, since it gave her access to research from other countries. *“I’m always online looking up different things. I just want to be informed . . . I know that Australia is doing an awful lot of research . . . the United States isn’t really doing the kind of research that they are in Australia.”*

Privacy was a concern to participants as well. S13, for instance, was hesitant to use a Facebook group because, *“there are platforms that you would want to say ‘Hey, I’ve got this and here’s what I deal with.’ and there are others you don’t. So it’s for that reason that I think a lot of people are shy about it, or they’re not technologically savvy, or they don’t want to air their personal business.”* There was concern from participants that their information was being seen by *“snake oil types that prey on desperate people and will give them all this information about how they’re going to cure them and make them better and I hate those kind of people. I just really wish they would go away, but unfortunately they don’t, they just get worse and worse.”* (S10)

Despite these privacy concerns, participants who used Facebook groups had more positive experiences than those who used the rare disease specific online communities. Participants reported that there was little activity on these communities; we observed this throughout our recruitment process too. Most participants from these separate sites had recently

registered and were disappointed by the lack of activity. M1 posted a question and did not receive any responses, *“Nobody said a word. And it isn’t about me; it’s just that no one is saying anything. In many communities there are one or two very active people, but relatively little activity from other members. I think there is one person who answers all of the questions for them, so I decided not to get involved with that.”*

DISCUSSION

Many of the issues uncovered in our study are extreme examples of what has been seen with more common diseases. Participants expressed concerns about communicating their disease to their friends and family, and were worried about care network fatigue. We draw from literature on older adult care networks to provide recommendations on expanding this care network to reduce care network fatigue. Participants also discussed their relationship with their doctor as being particularly challenging; we identify several approaches taken in prior research and discuss how they can be extended to apply to rare disease populations.

We also found that people with rare diseases faced unique challenges that differ from previous studies of chronic illness populations. Where previous studies have encouraged researchers to focus on the whole person outside of just the disease, participants saw themselves as representatives of their disease and considered it to be part of their identity. We also found that unlike prior research, separate online communities were not effective at engaging participants.

Expanding the Care Network

Participants spoke a great deal about the toll their conditions took on relationships. It was difficult for participants to form new relationships, and it was also difficult for existing relationships to adjust to the change in dynamics following a diagnosis. Participants were frustrated by the challenges of trying to communicate what they were going through to those around them, but also worried about the fatigue their experiences were causing their informal caregivers.

This care network fatigue is something that technology can help to address. Consolvo et al. [6] describe care networks comprised of many different people (family, friends, neighbours, etc.) who play diverse roles in the care of older adults. For people with rare diseases, technology is likely to be used not so much to coordinate this care (as in [6]), but rather to communicate about these rare diseases, thereby helping to increase the size of the care network, and to reduce the burden and fatigue placed on each caregiver. **We encourage HCI researchers to design for a slow discovery process through which people with rare diseases can disseminate information about their conditions** so that their friends and family members can slowly transition from a general level of awareness (understanding what the condition is, building empathy) to a specific level of understanding (how to be helpful). This would help to gradually bring more trusted people into the care network and reduce care network fatigue.

Liu et al. [17] found that people with chronic illnesses (HIV, diabetes, and cancer), used video blogs to educate viewers about their conditions and share their own experiences in a

diary-like format. This strategy could also benefit people with rare diseases as a way of helping them communicate their experiences to others. In fact, many of the experiences of people with rare diseases are extremely unique and engaging. This format could also be useful to trade tips and tricks. Participants expressed that some of their friends wanted to be helpful, but did not know how. Technology can facilitate the sharing and navigation of this information, mindful that there are likely to be case-specific limits to the extent of information deemed necessary for adequate helping. Our study draws attention to carefully balance a potential friend's informational needs with the individual's dignity and right to privacy.

A new diagnosis can cause a quick shift in the dynamics of an existing relationship — people are rapidly forced into caregiving roles they had not anticipated. S6's husband reacted to the role change by going into "complete denial," which was frustrating for S6. This story of a primary caregiver in denial, when considered in light of the higher divorce rates [8, 30] for couples where one person has a chronic illness, suggests that **we need to investigate how to help caregivers accept a new diagnosis and work towards providing them with the ability to catch up when they are ready.**

It's Who I Am

One notable difference between people with rare diseases and people with more common chronic illnesses involves their self-perceptions in relation to their conditions. Recent work in the CHI community has encouraged us to think about people as people instead of as patients, emphasising their humanness rather than their disease [1, 11, 19, 21]. To the contrary, however, participants in our study, regularly made statements saying, "*It's who I am.*" (S3) or "*This is part of my story.*" (S12). They spoke of themselves as ambassadors, as if they could not be understood apart from their medical conditions. This diversity of views affords the research community opportunities to use technology in a way that not only encourages people with rare diseases to think of themselves as whole people, distinct from their diseases, but also facilitates the exploration of multiple senses of self [33], particularly when rare disease communities are couched in larger social networks with mixed audiences and collapsed contexts [4, 35]. We can also leverage technology to support these individuals as advocates and help them to achieve the public awareness they are working towards.

Social media is a free mechanism with relatively low overhead and huge potential to enhance awareness of rare diseases. It is worth noting that increased awareness of rare diseases can sometimes invite unwanted comparisons with other, more common diseases. The ALS Ice Bucket Challenge, for example, has come under criticism because, although amyotrophic lateral sclerosis (ALS) is a terrible disease, it is rare and does not impact many people. Some have argued that more attention and money should be given to diseases impacting larger populations [2]. **We must investigate mechanisms people use for advocacy outreach, public response to these initiatives, and ultimately how the initiative improves the initial outreach aims** so that we can assist small groups, such as those with rare diseases, effectively and ethically raise awareness.

Leveraging Existing Platforms

We found that disease-specific types of community that have been the subject of prior work (e.g. [11, 24, 27]) were not actively used by people with rare diseases because it is extremely difficult to achieve the critical mass necessary for these communities to be successful; rare diseases, by definition, involve small numbers of people. **We must design ways to integrate communities into platforms that people are already using to reach people with rare diseases and build that critical mass.** It may be possible to address the privacy concerns expressed by participants in our study, through closed social media groups, although [24] found that private mechanisms within social media sites were not enough. Text message based interventions, as in [36], may assuage these concerns and are worth further exploration. However, it is important to balance this desire for privacy with the desire for awareness about the condition; a possible solution might be a private platform with options to share to more public-facing platforms.

Patient-Provider Relationship

The patient-physician relationship was a major source of complaints, frustrations, and stress for participants. They reported physicians not believing them, not knowing about their diseases, not being willing or able to take time to learn about their diseases, or not being willing to treat someone with a condition with which they were unfamiliar. Especially in emergency situations, these challenges can lead to serious if not lethal health consequences. However, technology can be valuable to enable physicians to (1) more clearly differentiate empowered patients from obsessive hypochondriacs, and (2) quickly learn about unfamiliar conditions and trust the information.

Past studies of chronic illnesses populations have encouraged HCI communities to consider patients as "experts", especially in the day-to-day management [1, 19, 21]. In cases of rare diseases, the line between considering a patient as empowered and an expert and giving too much weight to someone's psychosomatic concerns is a very thin and dangerous one. If doctors are unfamiliar with the presenting symptoms of a condition, and unable to easily diagnose them, they may assume such patients have been heavily influenced by things they have read online. They are challenged to distinguish those with legitimate, but unusual complaints from those whose symptoms are purely psychosomatic, imagined, or fabricated. Technology can help make this distinction. **People could leverage technology to provide their doctors with evidence to support the diagnostic process.** For example, an individual with OCP could use photographs of the eye collected over time to observe the effectiveness of treatment. Kientz et al. [15] adopted similar approaches in their design of technologies to evaluate treatments for children with autism and help their caregivers communicate. Applying Kientz et al.'s approach to rare diseases may hold promise as an area of further study.

There is also a difference between doctors seeing patients as experts and patients seeing themselves as experts. **We can design technology that leverages the patient's perception of herself as intelligent and knowledgeable to encourage**

the adoption of new technologies and compliance with disease management practices. Participants often referred to their own medical or research background or to someone they knew who was medically knowledgeable, which indicated it was important to them to be perceived as intelligent and as experts in their own situations. Past studies of end-stage renal disease patients [26] showed similar results — participants were proud of their ability to use scientific terms, and Siek et al. recommended this eagerness be leveraged to get patients to adopt technologies.

Researchers create technologies to manage health information that focuses on the patient as the information repository [16, 22, 25]. We agree that putting patients at the centre of this information would be appropriate here as well. Participants in our study were, for the most part, highly motivated and engaged in the management of their own health. People with rare diseases, in particular those active in online communities, represent a population that is highly motivated to try new approaches and put forth effort if there are benefits to be gained. This differs from studies of chronic illness patients and data enthusiasts, which have revealed a resistance to self-tracking that requires too much effort from patients (e.g. [18, 28]). Of course, this is not to say that technology should be needlessly complicated, but there are clues that people with rare diseases may be willing to tolerate additional burden for greater returns. Putting patients at the centre of managing their medical information empowers them to feel in control.

We need to take these patient-centred information repositories a step further, enabling patients to convey knowledge about their illnesses to health professionals, and enabling health professionals to quickly learn about new illnesses from authoritative sources. **We recommend a physician-endorsed personal health record that provides a quick overview of the illness, patient care, and sources for more information.** Since the information would come from multiple sources — especially the patients — this would involve connected personal health records where people with rare diseases would use their online networks to find physicians who are trained in their conditions. These personal health records could generate multiple views depending on the need; the system could provide a layman’s overview (like S8’s business cards), a quick medical summary (for hospital emergency visits), a “greatest hits” view of the most pertinent information for new doctors (like U1’s medical résumé), and the complete history (for deeper discovery). Current personal health records allow for navigation in a few simple ways, but allowing for more tailored information displays would be extremely helpful. We recognise that this notion of a personal health record is fraught with political and cultural challenges that need further investigation, however the growing use of tethered Personal Health Record systems might predict this to be a similarly feasible plan in the foreseeable future.

LIMITATIONS

Our decision to recruit participants through online communities gave us access to a wider range of conditions from different types of health care systems around the world. It also allowed us to explore the benefits those communities provide

to people with rare diseases. However, people who are active in online communities tend to be more empowered about their health [5] and so their experiences may differ from less engaged people with rare diseases. Additionally, individuals who did not have access to online communities (either because none exist for their conditions or because they do not have access to technology) may also have a different perspective, since they would need to seek support elsewhere. Because we are interested in sociotechnical interventions, we chose to study individuals who are likely to be early adopters — they are already willing to be helped through technology.

CONCLUSION

We provide the HCI community with a glimpse inside a rare world where people with chronic rare diseases have to take on many roles — from educating to advocating to providing support for people within their immediate and broader community. People with chronic rare diseases are concerned about the toll their disease puts on their caregiver network. They constantly have to educate, advocate, and research their disease to ensure that they can effectively communicate with concerned parties about their illness and treatment. Because these diseases are so rare, there is not always the critical mass of people facing the same battle that is necessary for online health communities, and thus must risk some privacy to connect with people with similar illnesses on popular social media platforms. Some of these findings are similar to what researchers have reported on in common chronic illness populations, however based on the limited knowledge, resources, and support for people with rare diseases, the experiences presented here highlight extreme issues that can be addressed with sociotechnical systems. We expand on the current literature on chronic illness populations by illustrating how people in rare worlds think of themselves and their disease synonymously. We conclude with examples of how we can improve personal health information management for people with chronic illness so that people with rare diseases can efficiently communicate their disease to health professionals — which is imperative during emergency situations. The HCI community can use these results to create the next generation of sociotechnical tools for people with chronic illness to connect, educate, research, and share information with everyone in their care network.

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