RARE WORLD: INVESTIGATING SOCIAL SUPPORT IN RARE DISEASE AND COMMON CHRONIC ILLNESS COMMUNITIES

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For the zebras and spoonies —

thank you for sharing your stories and guiding me through your Rare World.

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RARE WORLD: INVESTIGATING SOCIAL SUPPORT IN RARE DISEASE AND COMMON CHRONIC ILLNESS COMMUNITIES

Rare diseases impact small numbers of people. In the United States, a rare disease is one affecting less than 0.06% of the population. Although any given rare disease is, by definition, rare, the number of people living with a rare disease is substantial. It is estimated that 10% of people world-wide have one of the approximately 7,000 different known rare diseases. If everyone with a rare disease lived in the same country, it would be the world's third largest nation. In this dissertation, I investigate social experiences in common across a range of conditions and the potential of sociotechnical systems.

I began with exploratory interviews to investigate the breadth of these experiences. I distributed a survey to investigate differences between the experiences of rare disease and common chronic illness populations. I conducted a group-based study focused on people with rare diseases' relationships with informal caregivers, investigating sources of social support and unmet support needs.

People with rare diseases face barriers to receiving support and instead turn to online communities for emotional and informational support, but these communities do not sufficiently facilitate tangible support. It is more challenging for people with rare diseases to exchange tangible peer support because they are geographically dispersed and have similar symptomatic limitations. In the final study, I explored one possible approach to addressing this need: the application of social matching to an expanded network of caregivers for people with different rare diseases.

Conducting this research also necessitated the development of new research approaches. I review methods for analyzing skewed or imbalanced data, I explore ways of conducting group-based research remotely with dispersed populations, and I consider privacy challenges of working with a small, sensitive population.

This dissertation addresses a gap in literature on technology for chronic illnesses, focusing on a population rarely studied in sociotechnical fields: people with rare diseases. The primary contribution is in providing researchers with an in-depth understanding of the experiences of people with rare diseases, towards the design of better sociotechnical systems. A secondary contribution is in providing methods and approaches for conducting research with this unique population.

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> *So I call it* ***'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.*
>
> — Anonymous Research Participant (Chapter 3)

INTRODUCTION

Chronic illnesses such as diabetes, arthritis, or cancer impact a large and growing subset of people. Researchers have responded with technologies addressing the symptoms, causes, and management of these diseases. These conditions are well studied in the medical literature and have a well-defined set of symptoms and treatments to address with technology. To this end, technologies are often customized to a specific chronic illness and diseases impacting smaller numbers of people (i.e., rare diseases) are often left out of the health design space.

Each country defines rare diseases slightly differently. In the United States (US), a rare disease is one affecting less than 200,000 people nationwide (or 0.06% of the population) [300]. In Europe, a rare disease affects no more than 1 out of every 2,000 people (or 0.05% of the population) [300]. Although any given rare disease is, by definition, rare, the number of people living with a rare disease is substantial. It is estimated that 10% of people world-wide have one of the approximately 7,000 different known rare diseases [300]. If everyone with a rare disease lived in the same country, it would be the world's third most populous nation. Where existing research aims to address the medical needs of people with rare diseases (e.g., [140]) or understand the psychological impact of rare diseases (e.g., [300]), in this dissertation I consider the social experiences in common across a range of different conditions and the potential of sociotechnical systems to support these experiences.

Because rare diseases are a relatively new area of study in sociotechnical research, I first conducted exploratory interviews with people with rare diseases to investigate the breadth of experiences and the potential scope of technology to support those experiences. From this interview study, I

identified areas where the experiences of people with *rare* diseases differ from existing research about people with more *common* chronic illnesses. I distributed a survey to investigate these differences further. As a result of this exploratory work, I learned that having a rare disease forms a major part of one's identity and strongly impacts relationships with friends and family members. Next, I conducted a group-based study where I focused specifically on those relationships between people with rare diseases and friends/family, investigating sources of different types of social support and any unmet support needs. In the final study, I explored one possible approach to addressing this need: the application of social matching to a helping network for people with different rare diseases.

People with rare diseases are geographically dispersed, may require a range of accessibility accommodations, and face unique privacy challenges. Thus, conducting research with this unique population has necessitated the development of new research methods and guidelines; I discuss these considerations in this dissertation as well. I have considered the potential for identifying people with rare diseases from online behavior and address challenges of using imbalanced data (i.e., data about people with rare diseases and people with common chronic illnesses where there is a heavy skew towards the more common examples). I have explored ways of conducting group-based research remotely with dispersed populations: although researchers have developed best practices for using methods such as interviews over distance [89, 151, 337], little is known about conducting *group-based* research this way. Finally, I consider the privacy and ethical concerns of working with a sensitive population. Although researchers rarely release direct identifiers (e.g., names, contact information) when sharing research results, there are a number of indirect identifiers (e.g., disease name, gender, country) that make it easy to re-identify people with rare diseases from supposedly anonymized data. This re-identification risk introduces additional risks for people with rare diseases, impacting how we report on methods and results of rare disease research.

Research Questions

This dissertation addresses a gap in the current literature on technology for chronic illnesses, focusing on a population that has not yet been studied in sociotechnical fields: people with rare diseases. In this dissertation, I present the following thesis: *People with rare diseases have social experiences in common that are distinctly different from people with common chronic illnesses. I explore the*

[RQ1] What are the experiences of people with rare diseases?

(a) How are these experiences similar to the experiences of people with common chronic illnesses?

(b) How are these experiences different from the experiences of people with common chronic illnesses?

(c) How might we support the unique experiences of people with rare diseases through design?

[RQ2] What are the support needs of people with rare diseases?

(a) To what extent are these support needs being met?

(b) What are the sources of support for people with rare diseases?

[RQ3] How might we facilitate tangible support exchanges among people with rare diseases?

(a) What kind of tasks do people need support with? Which tasks could people best provide support with?

(b) Which kinds of characteristics do we need to know about two people for them to be interested in connecting with each other?

(c) How comfortable are people sharing these characteristics on social platforms? How comfortable are they sharing this information within a private community of people with rare diseases?

[RQ4] How might we conduct research with people with rare diseases?

(a) What can we learn about people from self-reported online behavior?

(b) How might we analyze quantitative data that is heavily skewed towards people with common chronic illnesses?

(c) How might we conduct group-based, human-centered research when populations are dispersed?

(d) What privacy and ethics considerations are necessary in conducting research with this sensitive population?

Figure 1: Research questions.

application of social matching techniques to address unmet tangible support needs as one possible approach. While investigating this thesis, I address the research questions presented in Figure 1.

Research Design & Dissertation Overview

This dissertation consists of three major sections.

Part I details existing research significantly informing this dissertation. In Chapter 1, I provide a definition of rare diseases and describe the historical context of patient activism in rare disease communities. This serves to define the scope and inclusion criteria of this dissertation and motivate the methodological decisions made throughout my work. In Chapter 2, I explore theoretical foundations of social support and applications of these theories to sociotechnical systems, situating my work within the context of prior research in the fields of health informatics and HCI.

Part II details the four main studies that make up this dissertation. The first three studies are needs assessment studies that provide an in depth understanding of this understudied population. In the first study (Chapter 3), I conducted interviews with 19 people with rare diseases to understand the scope of their experiences. I found that having a rare disease strongly impacts how people with rare disease think about themselves, interact with friends and family, and navigate the medical system. In the second study (Chapter 4), I distributed a survey to people with common chronic illnesses and people with rare diseases and used machine learning techniques to understand if and how they are similar or different from people with common chronic illnesses. I found that people with rare diseases are highly engaged online and especially rely on mainstream social media sites to obtain and share health information. In the third study (Chapter 5), I conducted a 22-week group-based study to focus more specifically on support needs and sources of support. In this study, I found that people with rare diseases often have strained relationships with friends and family and rely more heavily on online communities for many different kinds of social support. However, these online communities are not well suited for exchanges of tangible support because of the in-person nature of many tangible support tasks. I propose a system for connecting people with different rare diseases for purposes of tangible support. In the fourth study (Chapter 6), I evaluate this system through an online experiment to to better understand the design considerations and underlying social matching mechanisms of such a system. I find that the demand for and supply of tangible support tasks are often imbalanced, as is the information people are willing to disclose about themselves compared to the information they want to know about other people.

Part III details contributions in the form of new methods developed for conducting research with this unique and understudied population. Here in Chapter 7 I discuss an approach to handling skewed, imbalanced data in the survey study using Yang et al.'s [362] Soft-Margin Functional Gradient Boosting (Soft-FGB) approach. I compare this Soft-FGB approach against standard machine learning techniques and additional methods for handling imbalanced data. In Chapter 8, I describe the ARC method I developed for conducting group-based research asynchronously and remotely with a geographically distributed population. In addition to describing the method and my experience with it in the group-based study described in Chapter 5, I discuss other researchers' application of this method to other populations, and the insights gained from their extensions. Throughout all of this research, it important to consider the privacy implications of conducting

research with this unique population. In Chapter 9, I explore the possibility of anonymizing a quantitative rare disease dataset such that it could be shared with other researchers to facilitate additional research, finding that the usefulness of such a dataset largely depends on the use case, the evaluation metrics, and the method applied. I additionally explore the ways in which qualitative researchers report on participant data in publications and discuss the implications for research with people with rare diseases.

I conclude this dissertation by summarizing the results and implications of this research and discussing the opportunities for extensions of this work into future research. Overall, this research contributes to the design of better sociotechnical support for people with rare diseases by providing:

1. an in-depth profile of the social experiences and support needs of people with rare diseases,
2. a discussion of similarities and differences between rare and common chronic illnesses,
3. an approach to identifying people with rare diseases from self-reported online behavior,
4. a proposal for the design of a helping network to facilitate exchanges of social support,
5. an exploration of the application of social matching techniques to connect people with rare diseases via existing social networks,
6. an investigation of approaches to analyzed imbalanced or skewed data to better understand people with rare diseases within larger data sets,
7. a method for conducting group-based research remotely with geographically dispersed populations, and
8. an evaluation of the potential for releasing privacy-preserving versions of rare disease data to facilitate additional research.

Part I

Background

In this part of the dissertation, I provide an overview of existing work most significantly influencing this dissertation.

In Chapter 1, I provide background information on rare diseases. I describe what it means for a disease to be considered "rare" and outline the inclusion criteria for the rare diseases presented in this dissertation. I additionally provide a brief history of patient activism for rare disease research.

In Chapter 2, I review existing research on social support, beginning with theoretical foundations. I draw from work in social psychology to understand different types of social support and support relationships. I also discuss social support technologies developed in Human Computer Interaction (HCI) and other sociotechnical fields that address known challenges with social support.

This part of the dissertation serves to motivate my research approach throughout the dissertation; I discuss the influence of this history of rare disease patient activism on my methodological choices. It provides a foundation from which to explore the support needs and relationships of people with rare diseases discussed later in this dissertation (in Part II). Further, reviewing existing social support technologies allows us to consider whether some of these solutions (designed for people with common chronic illnesses) might be appropriate for people with rare diseases and to improve on these designs, towards the design innovative sociotechnical systems for people with rare diseases.

> *Rare diseases are rare,*
> *but rare disease patients are numerous.*
>
> — Orphanet[1]

Chapter 1

Rare Diseases

1.1 Defining 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). Although each individual disease is rare, it is estimated that 10% of people world wide have one of the approximately 7,000 different known rare diseases. [300]

Most of these diseases will likely be unfamiliar to many readers. A relatively familiar example of a rare disease is Amyotrophic Lateral Sclerosis (ALS), also known as Lou Gehrig's disease. This condition came into the public attention in recent years as a result of the "ALS Ice Bucket Challenge" where people challenged each other to dump buckets of ice water on themselves to raise awareness and funding for the condition [179, 308, 333, 351].

Throughout this dissertation, I rely on the National Institutes of Health (NIH)'s list of rare diseases[2] to determine inclusion criteria and consider only *adults* (over 18) with *chronic* rare diseases. These conditions have a wide range of symptoms and treatments. Names and descriptions of the conditions discussed in this dissertation are provided in Appendix F. [3]

[1] Orphanet is an online resource of knowledge on rare diseases and orphan drugs sponsored by the European Commission.

[2] The NIH's list of rare diseases can be found at http://rarediseases.info.nih.gov/gard/categories

[3] Throughout this dissertation, I use these boxes to call out important text and findings that significantly influence future sections of the dissertation. Readers may consult the content of these boxes to identify the key takeaways from a chapter and more closely read the surrounding paragraphs for additional detail.

The Global Genes Project conducted a survey with people with rare diseases and relevant stakeholders (caregivers, physicians, payors, policymakers, and researchers) across the US and United Kingdom (UK) [300] that showed a serious lack of resources and information available about rare diseases, with 92% of physicians reporting that it is more difficult to address the needs of a patient with a rare disease than their other patients during an office visit. More than half of patients and caregivers reported needing to advocate and provide their health care professionals with information about their disease. It takes an average of 7.6 years in the US (5.6 years in the UK) and eight physicians to receive a diagnosis [300]. Patients receive approximately 2–3 misdiagnoses before arriving at a conclusive diagnosis [300]. These numerous visits and diagnostic tests means that people with rare diseases have a higher cost of care than people with common chronic illnesses. Many respondents reported incurring medical expenses not covered by insurance or their National Health Service (NHS) (55% of respondents in the US and 18% in the UK) [300]. Many borrowed money from family or friends to cover these costs.

> There is a heavy emotional toll of living with a rare disease. People with rare diseases reported experiencing depression (75% in the US, 69% in the UK), anxiety (86% US, 82% UK), isolation from friends/family (65% US, 57% UK), less interaction with friends/family (70% US, 68% UK) and worry about the future (90% US, 91% UK). [300]

Bogart et al. [26] found that people with rare diseases have lower quality of life than the general population, and, more significantly, than people with common chronic illnesses. The authors additionally found that women, people with multiple rare diseases, people with lower income, and older adults had even worse quality of life. Interestingly, having a formal diagnosis was associated with an improved quality of life.

1.2 Patient Activism for Rare Disease Research

Existing support for rare diseases have largely been the result of patient activism and including patient activist groups in the research process has become increasingly common [227]. Thus, understanding this history of patient activism shapes the way I conduct research with this population. The US

government introduced the Kefaver-Harris Bill in 1962 in response to public pressure over the infamous thalidomide tragedy in which an unapproved morning sickness drug led to birth defects in thousands of children. The bill was intended to protect the public from potentially dangerous pharmaceuticals and described a requirement of adequate, well-controlled studies [141]. A side effect of the bill was that it prohibited testing on groups too limited in size for Randomized Control Trials (RCTs), including rare disease patients [217]. The bill also meant drug development costs were now much higher, and pharmaceutical companies began focusing on treatments for common diseases with large markets and higher profits [102]. It also made it hard for existing medications to be extended to under-served populations [102]. Medications that were economically unviable and impossible to research became known as "orphan drugs." Over the next decade, the gap between the development of common disease drugs and the development of rare disease drugs continued to widen such that there were no or almost no treatment options for certain diseases.

In 1983, the US government passed the Orphan Drug Act (ODA), stating that drugs developed to treat diseases affecting less than 200,000 Americans would be considered as "orphan." This act introduced economic incentives for development of orphan drugs including seven-year market exclusivity, tax credits, drug development grants, and fast-track approval of drugs indicated for rare diseases [139]. Seven-year market exclusivity was particularly appealing to pharmaceutical companies and was successful in encouraging new drug research. Prior to 1982, only 38 orphan drugs had been approved; by 2014, 373 drugs were approved [138]. However, the Act continues to be criticized as a *"policy that serves mainly to promote private industry by allowing companies to charge outrageous prices for products that rare disease sufferers have no option but to purchase"* [52]. When the European Commission passed a similar act in the 1990s, they counteracted some of this monopolization by stipulating that orphan drugs that were considered to be "extraordinarily profitable" would be stripped of their orphan status after five years [183]. The Australian model imitates the fee waivers and priority evaluation of the US model but does not offer market exclusivity, tax credits, or grants [298]

This legislation around orphan drug development was closely tied to patient activism. Patient groups sought legitimization of their conditions through critiques of *"deficiencies of the drug market"* [9, 29] and *"zones of undone science"* [118, 280]. These groups described rareness as a matter of equity and social justice (e.g., [149, 288, 289]).

In the early years of rare disease activism, initiatives were driven by organizations focusing on specific diseases but quickly led to the *"emergence of new identities, collectives and forms of political action"* [262]. Smaller rare disease organizations began to unite in pursuit of common goals.

This meant that the rareness of diseases was subjected to a *"politics of numbers"* [279]. Rabeharisoa at al. [279] explain:

> "By equating rareness with a series of figures (numbers of individual patients, prevalence threshold), the legislations legitimised concerned [patient organizations'] claims for rareness to be considered as an issue of equity and social justice. By counting and gathering patients, rare disease alliances moved towards the mainstreaming of rare conditions, as summed up in their slogan: 'Rare diseases are rare, but rare disease patients are many.'"

This collective mobilization led to the creation of the National Organization of Rare Diseases (NORD) in the US in 1983. NORD arose from a coalition of patient organizations that was instrumental in pushing for the orphan drug legislation; they can largely be credited with getting the ODA approved in 1983. That same legislation established them as an official not-for-profit organization providing support to people with rare diseases. NORD is largely funded through federal grants and public donations. [246]

The European Organization for Rare Diseases (EURORDIS) emerged much later in 1997, primarily out of the French muscular dystrophy association, L'Association Français Contre Les Myopathies (AFM), along with other French patient organizations of the time. AFM's primary goal was to push for legislation similar to that seen in the US. EURORDIS has since grown to represent more than 700 different patient organizations in 63 different countries [105]. They also sponsor Rare Diseases International (RDI), a collective that includes representation not only from NORD but also from Rare Voices Australia (RVA), Indian Organization for Rare Diseases (I-ORD), Ibero-American Rare Disease Alliance (ALIBER), and Canadian Organization for Rare Disorders (CORD), most of which are newer organizations that benefit from the guidance and expertise of a larger collective [163].

In parallel to pressure on governments to better encourage and facilitate research on orphan drugs, patient organizations (organized collectively under umbrella organizations) were frustrated

by the lack of interest by researchers and clinicians into their conditions [160]. They began to collect their own data, often in the form of "patient narratives" [273] and distributed this data to appropriate specialists to catalyze research. AFM was a major driving organization in France behind biomedical research [278] towards ending *"the vicious circle of ignorance and indifference"* [15]. Rabeharisoa at al. [279] describe this organization as having two main accomplishments:

> "(i) it gave shape to a community of researchers and clinicians ready to partake in the war on myopathies along with patients and families, and (ii) it developed organised cooperation between patients and experts in the orientation of research, and in the production and dissemination of knowledge on the diseases."

Patient organizations *"consider the detour through research as a solution to patients' problems, at the condition that the research system be transformed to include patients as active contributors, and to consider their needs and demands"* [279]. Involving patient advocacy groups in the research process has become increasingly common in rare disease research [227] and has seen varying degrees of success. Some have critiqued scientists for pursuing research avenues that are not well aligned with patients' actual problems [280]. The German Retinitis Pigmentosa Society suggested patients move first, before researchers have the chance to develop their own research agenda [338]. Panofsky [262] assessed nine different US-based patient organizations and similarly found that *sociability* (making links with experts) can make a big difference in getting research accomplished.

> Huyard [159] found that the main goal of patient activists was not necessarily a cure for their conditions stating, *"most participants were satisfied with honest and reasonable efforts to gradually improve their situation."* Understanding the everyday lives of rare disease patients is an essential goal.

The work of these patient organizations has extended beyond just research on diagnosis and treatment or their conditions; they have also built initiatives for broader education and empathy (e.g., [247, 248]), increased access to information and research (e.g., [223]), and devices to support disease management in everyday life (e.g., [241]). In fact, 53% of people with rare diseases report having developed some novel solution for managing their condition [255]; many of these people shared these innovations with other patients.

1.3 Summary

Understanding this history of patient activism informs my research in several significant ways. First, knowing that people with rare diseases have a long history of being interested in research and wanting to be involved throughout the research process informs the methodological choices I make in conducting this research. It means people with rare diseases are motivated and excited to contribute to research, so long as I can overcome geographic limitations of working with this group. It also means I have a heightened responsibility to conduct research that is well-aligned with participants' *actual* needs and interests. To do this, I rely on many of the principles from Community-Based Participatory Research (CBPR) [332], including understanding the existing strengths and resources within the community, finding opportunities for mutual learning, and disseminating knowledge to all partners.

Second, knowing that an improved situation is priority (as opposed to focusing entirely on a cure) suggests that there are issues to address that are well-suited to sociotechnical research disciplines. Positive and ecological perspectives from CBPR [332] and attitudes of designing for everyday life [14] would be valuable, and thus is the focus of this work.

Finally, this history of people with *different* rare diseases bonding together in pursuit of collective goals suggests that, from this everyday life perspective, there are common experiences with rare diseases.

> This philosophy of *"rare diseases are rare, but rare patients are many"* is at the crux of my research which aims to address these widespread problems, rather than focusing on specific diseases with well-defined symptoms.

“ *Kindred spirits are not so scarce as I used to think.*
It's splendid to find out there are so many of them in the world. ”

—L.M. Montgomery in *Anne of Green Gables*

Chapter 2

Social Support

In this chapter, I explore theoretical foundations of social support. First, I define different types of social support, particularly in the context of online health communities. Understanding these types of support allows us to examine how the support needs of people with rare diseases are met (or not met). In this dissertation, I am interested in the *sources* of support (i.e., from friends and family, from other people with rare diseases, from online communities, etc.) and the areas in which support may be lacking. Second, I discuss theories of help-seeking and expectations of support in different types of relationships. This helps to understand barriers to seeking and providing support. Understanding how those norms vary by relationship is helpful to better contextualize different sources of support for people with rare diseases. I conclude with a discussion of the differences between voluntary and obligatory support. I draw from work suggesting a shift towards peer support and away from traditional caregiving models which I propose as a valuable lens for thinking about support for people with rare diseases.

2.1 Defining Social Support

Social support is defined as *“verbal and nonverbal communication between recipients and providers that reduces uncertainty about the situation, the self, the other, or the relationships, and functions to enhance a perception of personal control in one's experience”* [4]. Social support can be beneficial when used appropriately, where “appropriate” is determined the nature of the relationship between

the provider and recipient [75, 152], the timing of the support [165], the personality of the recipient [152, 191], the nature of the stress [61, 74], and the spontaneity of the support [75, 84, 113, 190]. When misused, or provided in the wrong context, it can have a negative impact [192].

> Davidowitz and Myrick [78] found that attempts at making supportive comments are actually unhelpful 80% of the time. One explanation for unhelpful support is that victims have more information available to them about the nature, severity, and duration of the stress. Supporters might not have personally experienced the stress so they may have misconceptions about how it affects a person [304].

People who have not experienced the same stress sometimes discourage open discussion of emotions and encourage the person to stay positive [126, 272], even though the belief that it is unhelpful to focus on the negative aspects of one's situation has been repeatedly shown to be a false assumption [147, 172]. Support providers encourage a quick recovery, believing people should recover faster than they actually do [304]. Under uncertainty, people fall back on scripted support, like giving advice or telling the person they know how they feel [358]. Victims often do not share these perspectives on what is helpful. Lehman et al. [192] explain,

> "Indeed, bereaved respondents might be expected to view efforts designed to discourage open discussion and encourage recovery as unhelpful because (a) they isolate the person by curtailing opportunities to share concerns; (b) they seem to dismiss the victim's feelings as insignificant; and (c) they are judgmental, implying that they victim should not feel as bad as he or she does. Similarly, telling a bereaved person 'I know how you feel' or offering advice may be viewed as unhelpful because it seems to dismiss or trivialize his or her problems."

My research in this dissertation has shown that people with rare diseases often receive well-meaning but unhelpful comments from their friends and family members; especially given the rarity of the conditions, it is understandable that it would be a challenging experience to understand without having gone through it oneself. Two strategies that have been shown in existing research to be helpful are allowing the person to talk openly about their feelings and encouraging contact with people who *have* experienced a similar stress [192].

2.2 Computer Mediated Social Support

Online support groups are known to provide many positive benefits to group members and overcome many of the barriers of face-to-face support groups [348]. For people with rare diseases, these groups are especially valuable as they may *"provide the only way to communicate with others who are dealing with similar problems [...] This international scope permits group members to draw from a wide variety of perspectives, experiences, disabilities and points of view, while at the same time promoting a feeling of universality."* [348]. These groups have the potential to connect people who have experienced similar stresses and may be better at providing support [192]. For this dissertation, I rely on Cutrona and Suhr's [76] taxonomy (Table 2.1) to understand different types of social support because of its suitability for analyzing computer mediated communications and its widespread acceptability in HCI.

> Cutrona and Suhr classify types of support into two categories: *action-facilitating support* (informational, tangible) and *nurturant support* (emotional, network, esteem). Action-facilitating is intended to eliminate a problem, whereas nurturant support focuses on comforting or consoling without directly addressing the problem.

Researchers often rely on this taxonomy to examine support behaviors occurring online, reporting on the presence of different types of support in messages exchanged (see Figure 2.1).

Informational support is commonly the subject of messages exchanged in online communities. Braithwaite et al. [28] hypothesize these communities make it easier to offer information because supporters can take time to look up information instead of having to respond immediately. Posting a request for information online also means it will be read by a larger audience than just one person, increasing the likelihood of a useful response.

Emotional support is also common in online support communities. This is true in rare disease communities [5, 10, 69, 91, 136, 270]. Online communities are successful because they represent a place where people can connect with people experiencing similar health challenges who might better understand what they are going through. There may also be an element of anonymity here: people are more comfortable discussing stigmatized topics online where they can keep their emotional challenges separate from their offline identities [7].

Action Facilitating Support	**Informational Support**	*"advice ('I think you should tell your supervisor'); factual input ('If you don't treat the infection quickly, it will get worse'); and feedback on actions ('You shouldn't have told her so bluntly')."*
	Tangible Support	*"offers to provide needed goods (e.g., money, food, books) and services (e.g., baby-sitting, transportation, housework)."*
Nurturant Support	**Emotional Support**	*"expressions of caring ('I love you'), concern ('Are you feeling better?'), empathy ('You must have been really hurt by his coldness'), and sympathy ('I'm so sorry you're upset')."*
	Network Support	*"a sense of belonging among people with similar interests and concerns ('We'd like you to join our support group')."*
	Esteem Support	*"expressions of regard for one's skills, abilities ('I know you'll do a good job') and intrinsic value ('Losing your job doesn't mean you're worthless')."*

Table 2.1: Cutrona and Suhr's [76] definitions of the five categories of social support.

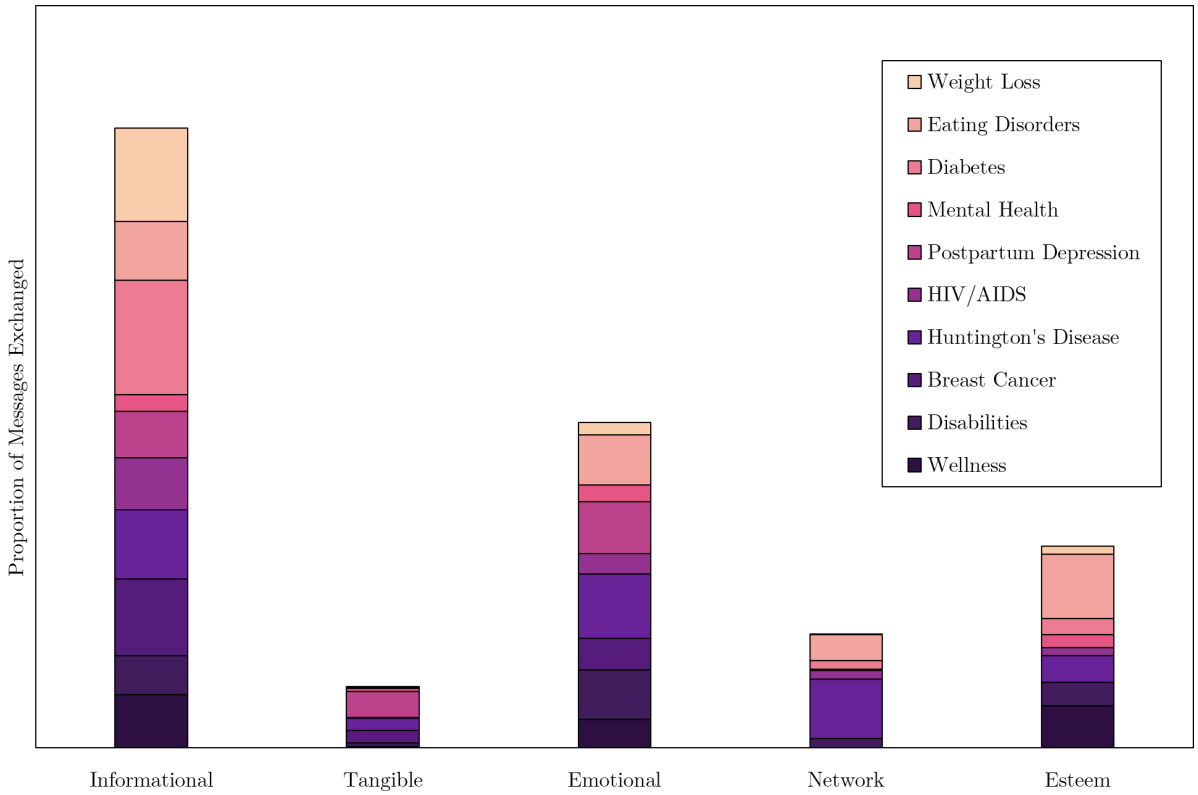


Figure 2.1: Prevalence of different types of social support in computer mediated communications. These ten studies include overall wellness [1], disabilities [28], breast cancer [55], Huntington's disease [69], HIV/AIDS [70], postpartum depression [106], mental health [121], diabetes [198], eating disorders [224], and weight loss [331].

Esteem and network support are less frequently provided via online messages, but research suggests that participating in an online community contributes to satisfying these support needs, even if they are not discussed explicitly [28, 69]. Receiving "Likes" or comments on one's content contributes to esteem support needs and the mere act of being a part of an online community of similar others satisfies one's network support needs [1, 28, 69]. Braithwaite et al. [28] explain that people with disabilities often have constricted social networks and place a high value on network support. For people with rare diseases who have similarly constricted networks, these online communities have much to offer.

Tangible support seems to be much less common online [1, 28, 70, 121, 198, 224, 331]. Although Cutrona and Suhr's original study [76] showed reasonably high levels of tangible support, it is *consistently* the least frequent form of support in digital communities. Most behaviors that are classified as tangible support require collocation between partners (present in Cutrona and Suhr's [76] study but not in online communities).

> There is very little tangible support uncovered in studies of online health communities. In many cases, this tangible support need is attended to by collocated family and friends (as in [305]). Our research in this dissertation suggests this is not the case for people with rare disease.

2.3 Barriers to Help-Seeking

An additional barrier to the exchange of tangible support is that people prefer spontaneously offered support and struggle to ask for help when needed [75,84,113,190]. In *exchange relationships* (between acquaintances or strangers) support is given *"with the expectation of receiving a comparable benefit (or benefits) in return"* [57]. Receiving support from an exchange partner incurs an obligation to provide an equivalent benefit in return. Gouldner calls this norm "reciprocity." [131]. People keep track of how much they owe and are owed [58] and even conduct a brief cost-benefit analysis before choosing to enter into the relationship [101]. In *communal relationships* (between close friends, family members, or romantic partners) support is given without any expectation of repayment, and Gouldner's [131] theory of reciprocity does not apply. The support recipient may still feel motivated

to similarly provide support in the future should the opportunity arise, and the support provider may still *"hope that the recipient will be similarly responsive to his or her needs as they arise"* [57], but there is no expectation that these benefits come with a price tag.

> In exchange relationships, support seekers feel embarrassed when asking for help [84, 190,353]. They underestimate the willingness of others to comply with requests for help [113], feel they are imposing on their partner [113], or worry about incurring a debt they might not be able to repay [57, 75, 84].

Spontaneous support is also preferred in communal relationships: people keep track of their partner's needs, even if they cannot do anything and regardless of reciprocation [58, 59]. Support recipients in communal relationships generally consider discussions about support needs to be a violation of the communal norms [75].

> If a person does not provide support until asked, this causes their partner to doubt the quality of the relationship. If help is provided unsolicited, this reinforces perceptions of the helper as attentive to the needs of their partner and strengthens the intimacy of the relationship. [58, 59, 75]

This suggests that even healthy relationships are filled with challenges. People do not ask for support when they need it but expect people to provide support without being asked.

2.4 Prosocial Behavior vs. Caregiver Burden

Just as support behaviors vary by relationship type, they also vary when voluntary or obligatory.

> Voluntarily engaging in prosocial behaviors (e.g., volunteering) provides positive benefits to the helper [17, 35, 230], including improved mood [54], improved health [355], increased quality of life [296], and reduced mortality [35]. However, when caregiving is an obligation (e.g., caring for an ailing family member), researchers describe practical consequences and emotional burdens [51, 134, 236].

Unfortunately, caregivers deal with these burdens largely in isolation, feeling they cannot vent to someone more vulnerable [51], especially if expressing their frustrations may exacerbate symptoms (as in depression [360]). Caregivers often lose their own social connections through the caring process [360] and can develop depression and anxiety [134]. Sociotechnical researchers have taken several approaches to addressing caregiver burden; these systems often focus on communication, coordination, or providing information [77,195,258,318]. For example, the Estrellita system [195,318] aims to make things easier for parents of high risk infants by facilitating communication between parents and the medical team. Researchers have expressed concerns that these kind of systems may unintentionally increase the burden on caregivers by making it easier to do even more work [51]; caregivers are also concerned about the potential burden of these systems if they have to manage the technology on top of their existing caregiving tasks [161]. Instead, Chen et al. [51] and Tixier et al. [322] both suggest systems for caregivers should include room for personal tasks to bring balance to the caregiver's life.

Chen et al. [51] noted that caregivers often feel under-appreciated for their invisible work. One approach involves expanding what Consolvo et al. [64] call "the care network", arguing care is not handled just by one or two people but by a range of people, including friends, family members, neighbors, professional caregivers, pharmacists, doctors, house cleaners, or many others. Expanding the care network would allow the workload to be distributed across a larger set of people. Hong et al. [154] leverage this idea in their design of SocialMirror, a device for young adults with autism that allows them to seek advice from a trusted network. To decrease the burden on primary caregivers, as well as to avoid overreliance by the young adult on any one adult, the mirror works with a trusted network of friends, family, and professionals. Caregivers can interact with each other and see each other's posts, which helps to overcome the challenges identified by Consolvo et al. [64], namely uneven distribution of responsibility, miscommunication, distrust, and unmet care needs.

Unlike research focused on supporting existing caregivers or distributing the workload across many caregivers, Arreola et al. [8] make a compelling argument for *peer* support. The authors' older adult participants relied more heavily on their peer group for support and hesitated to ask their adult children for help. Arreola et al. [8] argued for a shift from the one-caregiver-to-one-older-adult model to a peer support model in which older adults help care for each other:

> "By replacing the individual caregiver with a peer group, each older adult has more eyes watching for them in case there is a problem. There is also less social stigma associated with asking about the day-to-day business of peers. This allows for less friction in terms of communication and may enhance activity within a community."

Stroomer et al. [313] similarly found this shift towards peer support to be a successful strategy. They explain, *"people may be willing to provide support within their community, proving the barriers are lowered in asking for support. The field trial demonstrated that a social network service could be used to lower barriers towards asking for help."*

> Shifting towards peer support rather than relying as heavily on informal caregivers has the potential to reduce barriers to help-seeking, improve the quality of support provided, and activate secondary benefits of engaging in prosocial behavior.

2.5 Summary

Social support, although potentially beneficial, is not without complexities. In the case of chronic illnesses, people are sometimes forced into caregiving roles they had not anticipated. They are rarely sufficiently prepared to be effective in these roles. Research on supporting caregivers or expanding the care network provides thoughtful solutions in cases in which there is at least oneoverburdened caregiver(s). Online communities are also common sources of support, especially for informational and emotional types of support.

Online communities have the potential to connect people with rare diseases to people they would not otherwise be able to meet, however these communities may not be sufficient if they cannot provide tangible support requiring collocation. In this research, I investigate the support needs and the sources of social support for people with rare diseases.

Part II

Social Support in Rare Disease & Common Chronic Illness Communities

In this part of the dissertation, I report on four studies that constitute the core contribution of this dissertation and answer RQ1–RQ3 (provided again for reference in Figure 2.2.)

[RQ1] What are the experiences of people with rare diseases?

(a) How are these experiences similar to the experiences of people with common chronic illnesses?

(b) How are these experiences different from the experiences of people with common chronic illnesses?

(c) How might we support the unique experiences of people with rare diseases through design?

[RQ2] What are the support needs of people with rare diseases?

(a) To what extent are these support needs being met?

(b) What are the sources of support for people with rare diseases?

[RQ3] How might we facilitate tangible support exchanges among people with rare diseases?

(a) What kind of tasks do people need support with? Which tasks could people best provide support with?

(b) Which kinds of characteristics do we need to know about two people for them to be interested in connecting with each other?

(c) How comfortable are people sharing these characteristics on social platforms? How comfortable are they sharing this information within a private community of people with rare diseases?

[RQ4] How might we conduct research with people with rare diseases?

(a) What can we learn about people from self-reported online behavior?

(b) How might we analyze quantitative data that is heavily skewed towards people with common chronic illnesses?

(c) How might we conduct group-based, human-centered research when populations are dispersed?

(d) What privacy and ethics considerations are necessary in conducting research with this sensitive population?

Figure 2.2: Research questions.

In Chapter 3, I describe an exploratory interview study ($n = 19$) with people with rare diseases. I found that having a rare disease impacts many different roles and relationships in one's life and learned about experiences in common among people with rare diseases, even when symptoms vary.

In Chapter 4, I investigate differences between people with rare diseases and people with common chronic illnesses using a survey informed by the results described in Chapter 3. I distributed this survey to people with common chronic illnesses as well as people with rare diseases ($n = 328$) to better understand the differences between these two groups and to identify the rare examples. I learned that people with rare diseases are active users of mainstream social media platforms for engaging with their health.

In Chapter 5, I explore sources of support for people with rare diseases. I used the Asynchronous Remote Communities (ARC) method where people with rare diseases engaged in discussions with each other and completed a series of activities throughout the 22 weeks of the study. I outline the differences in people's perspectives on the relationships they have with people within Rare World (i.e., people who also have a rare disease) and outside of Rare World (i.e., healthy people or people with common chronic illnesses), identifying support needs that are unmet. I propose an approach to addressing these unmet needs, shifting away from a caregiving model towards a model of networked peer support, framing the system as a "helping network" to overcome barriers to help seeking.

In Chapter 6, I explore the use of social matching techniques to connect people with rare diseases to people with *different* rare diseases to exchange tangible support. I discuss the potential benefits of leveraging an existing platform for such a community, including a specific Facebook feature called "Discover People" which visibly surfaces "Things in Common" between people. I consider the privacy implications of disclosing the information necessary to facilitate this matching to a range of different potential audiences.

This part of the dissertation provides an in-depth profile of the social experiences of people with rare diseases, moving from a broad perpective on the range of experiences in Chapter 3 to an exploration of one possible mechanism for helping people with rare diseases to help each other in Chapter 6. I compare these experiences with those those of people with common chronic illnesses to identify areas where existing solutions might be adapted, areas where new solutions are necessary, and opportunities for thes two populations to overlap.

Limitations

Participants were recruited from social media, through support groups or paid advertisements. Social media as a recruitment source often leads to an over-representation of women [347], and this was true in the research described in this dissertation as well. The decision to recruit participants online provided access to a wider range of conditions from different types of health care systems around the world. It also allowed for exploration of the benefits these communities provide to people with rare diseases. However, people who are active in online communities tend to be more empowered about their health [32] and 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 the relevant technology) may also have a different perspective, since they would need to seek support elsewhere. Because this dissertation focuses on sociotechnical interventions, I chose to study individuals who are likely to be early adopters; they are already willing to be helped through technology. Future work might build on this foundation by considering less digitally connected participants.

I define "rare disease" using the NIH's database because of its reliability and comprehensiveness. A limitation of this approach is that the database focuses primarily on diseases that are rare in the US; these diseases may not be considered rare in other countries and there are additional diseases that would not be considered rare in the US. An ideal approach would take into account whether or not the disease was rare in the country where the participant lived, but rare disease databases do not exist in many of these countries. Given that people from all around the world actively contribute to online communities for many of the diseases from the NIH list, I allow for participation from international participants.

Throughout this research, participants told us the name of their condition and we never asked for proof of a diagnosis. We know from previous work [300] that people with rare diseases are misdiagnosed two or three times on average before receiving a "final" diagnosis. It is possible many participants will be un-diagnosed and re-diagnosed several times in the future; they may not actually have the condition they have been told they do.

There are even questions in academic literature about whether some of the diseases described by participants are real or not. As an example, Morgellons, a diagnosis self-reported by a participant

from Chapter 3, has been the subject of significant disagreement in the medical community over whether this is a tickborne infection [229] (including symptoms such as fatigue, skin lesions, diffuse musculoskeletal pain, cognitive dysfunction, and emotional lability [294]) or some form of delusional parasitosis or monosymptomatic hypochondriacal psychosis. It has been described as everything from a "medical mystery" [294] to an "Internet meme" [199] to a "folie a deux" (a collective delusion) [242]. However, participants in this research describe similar experiences, regardless of my confidence in the legitimacy or finality of their diagnosis. Participants with (relatively) less rare and (relatively) better understood conditions describe being doubted by family, friends, and the medical community just the same as participants with diseases that are more heavily contested in the literature. They described long periods of seeking a diagnosis, actively advocating for themselves to medical professionals, and seeing multiple specialists. There is no good way to accurately screen for hypochondria without also excluding the many people world wide who *do* have a rare disease. Further, I would argue that while a misdiagnosis or made up diagnosis might mean that the proposed physiological mechanisms are invalid, it does not mean their lived experiences are not valid and it is these lived experiences that are the focus of this work. Thus, in this dissertation, I air on the side of believing participants.

Finally, in this dissertation I have included participants with a wide range of conditions and a wide range of physical symptoms. I have deliberately recruited diversely, but have not relied on any measure of quality of life or disease severity to categorize one condition as being "worse" than another or one participant as having a harder time than another. Rather, I respect that participants experiences with their own condition are individual and influenced by their own environment and past experiences.

> *Personally, I always compare myself to Galileo because everyone knows he's fantastic, but he has no spoons*[1] *at all because he's dead. So technically I'm better than Galileo because all I've done is take a shower and already I've accomplished more than him today. [...] But I'm not gloating because Galileo can't control his current spoon supply any more than I can, and if Galileo couldn't figure out how to keep his dwindling spoon supply I think it's pretty unfair of me to judge myself for mine.*
>
> — Jenny Lawson in *Furiously Happy*

Chapter 3

Experiences of People with Rare Diseases

Researchers in sociotechnical fields have sought to understand the experiences of people with a range of common chronic illnesses, including cancer (e.g., [164, 336]), dementia (e.g., [186, 240]), mental health (e.g., [254, 360]), diabetes (e.g., [215, 325]), chronic pain (e.g., [107]), multiple sclerosis (e.g., [11]), tramatic brain injuries (e.g., [109]) and many others. With the exception of a few specific conditions (e.g., Kane et al.'s [171] work on ALS or Rodger et al.'s [285] work on rare muscular dystrophies), rare diseases are largely new to sociotechnical research communities. Thus, I begin here with a qualitative investigation into the needs and experiences of people with rare diseases. I conducted interviews with people with a range of rare diseases from different countries to better understand those experiences. In this chapter, I describe the method and results of this exploratory interview study with people with rare diseases. I additionally discuss some initial observations about the differences between people with rare diseases and people with common chronic illnesses, including their relationship with their conditions and their preference for online health communities to exist within broader social networks. This study was conducted collaboratively with Kim Oakes, Danika Geisler, Kay Connelly, and Katie Siek; the use of "we" in this chapter refers to those researchers. This chapter includes material from our CHI 2015 paper [208].

[1] The Spoon Theory is an essay by Christine Miserandino [232] in which she uses a pile of spoons to explain her invisible illness to her friend. Each spoon represents a certain amount of energy. Each activity throughout the day requires a spoon and a spoon can only by replaced as the person recharges. Once the spoons are gone, the person has to rest until their spoons are replenished. In invisible illness communities "spoon theory" has become *"an Internet shorthand for the experience of living with a chronic illness, having to think constantly about the number of 'spoons' as units of energy that are used up by daily activities"* [128].

3.1 Method

We conducted interviews with adults who were living with a rare disease to understand their experiences and to inform possible design opportunities. This study was approved by Indiana University (IU)'s Institutional Review Board (IRB) (Appendix A - #1405068949R001).

3.1.1 Recruitment

We recruited through online forums for people with rare diseases (RareShare[2], RareConnect[3], the Rare Disease Foundation[4]). Each online forum had subcommunities for different diseases. We posted to any subcommunity that had been active within the past three months, identifying ourselves as researchers and sharing our study information including contact information. Participants could contact us by email to express interest in participating or to request more information.

We also recruited from Facebook groups targeting specific rare diseases. We chose 30 conditions from the NIH list of rare diseases that impact a range of systems in the body to target a range of diseases. We searched for Facebook groups using the names of these conditions (some conditions did not have any Facebook groups, others had multiple groups). We reviewed the group descriptions and any guidelines or policies outlined by group administrators. In cases where the guidelines were unclear, we contacted group moderators to seek permission to recruit. We found 15 Facebook groups from which we were permitted to recruit. Similar to our approach with the rare disease forums, we posted to these groups identifying ourselves as researchers and sharing our study information.

3.1.2 Participants

We recruited participants who were over 18 years old, had a chronic rare disease diagnosis, and were members of an online community for their disease. No financial incentive was provided for participation. Over 150 people responded to our recruitment notices; we interviewed a subset of these people, aiming to target a range of condition and length of time living with the condition. We reached theoretical saturation after interviewing 19 participants. We identify participants in this chapter using P3.1–P3.19.

[2]http://www.rareshare.org/
[3]http://www.rareconnect.org/
[4]http://www.rarediseasefoundation.org/

Health System	Country	N
Private	Pakistan	1
	United States	10
Public	Brazil	1
	Canada	1
	England	2
	Ireland	1
	Macedonia	1
	New Zealand	1
	Norway	1

Table 3.1: Participants by country and health-care system.

Disease Name	N
Chiari Malformations	1
Ehlers-Danlos Syndromes	1
Congenital Anosmia	3
Familial Transthyretin Amyloidosis	1
Hereditary Angioedema	1
Hereditary Spastic Paraplegia	1
Inclusion Body Myositis	4
Kallmann Syndrome	2
Morgellons	1
Multifocal Motor Neuropathy	2
Ocular Cicatricial Pemphagoid	1
Systemic Capillary Leak Syndrome	1
Wilson Disease	1

Table 3.2: Participant Conditions. Two participants had multiple conditions and two participants were undiagnosed.

Seven participants were male (37%) and the remaining 12 were female. Participants ranged in age from 20–66 ($\bar{x} = 45$, $s = 13$). A little over half of the participants (53%) were living in the US in a predominantly private health care system. One participant lived in Pakistan, which also has a predominantly private health system. The remaining eight participants lived in countries with predominantly public health care systems (Table 3.1). All recruitment notices were posted in English. All interviews were conducted in English, except one which was conducted in Portuguese with the help of a translator.

We interviewed participants with 13 different conditions (see Table 3.2 for a list, full descriptions are provided in Appendix F). Two participants had multiple conditions. We identify these participants with the superscript $^{\mathrm{m}}$. We also respected that, especially in the case of rare diseases, it can take a long time to obtain a diagnosis [300]. 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 likely rare. We identify these undiagnosed participants using the superscript $^{\mathrm{u}}$. All other participants reported having a single rare disease.

3.1.3 Procedure

We conducted semi-structured interviews to understand participants' experiences with their conditions. Most ($n = 11$) participants undertook a one-hour video interview (using Skype, Google

Hangout, or FaceTime). Some were uncomfortable using video conferencing technology, thus we conducted six interviews by phone and two via instant message. We recognize the limitations of conducting interviews over instant message and adopt strategies from [89, 337] to maximize the effectiveness of this approach, including asking participants to elaborate on their thought process when they type for several minutes and only share a short response, summarizing our understanding of their response before asking follow up questions, and adapting the length of our pre-written questions for an online format.

We began with an informed consent process and preliminary demographic questions. The main interview was divided into four topics:

1. **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?

2. **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?

3. **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?

4. **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 a thematic interview guide (Appendix B) 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 us with some additional thoughts, which were added to their transcripts.

3.1.4 Analysis

In addition to the two instant message transcripts, we transcribed and analyzed the 17 interview recordings using inductive qualitative methods [311]. Three authors separately developed codes

based on the transcripts and discussed them. Each author iterated on these themes several times until converging on a set of common codes. We finalized the codes, and used them to analyze each transcript. Because of the sensitive nature of the interviews, we conducted a member check [193]. Participants were given the chance to review our CHI 2015 paper [208] before publication to ensure they were comfortable with the content and that we had accurately represented their views. No participants requested changes.

3.2 Findings

Participants' conditions pervaded many aspects of their lives, and having a rare disease put them in a unique position. P3.11 described the experience:

> "So I call it '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, I discuss challenges participants faced in these different aspects of their lives, and strategies they employed to overcome them.

3.2.1 Role As Friends

Participants varied widely in how much they shared with their friends about their experiences. Several participants spoke of the challenge of communicating their experiences when their diseases were unfamiliar to others. This was particularly true in cases where the conditions were externally invisible; participants described how people generally associated disability or disease as being something visually perceptible. For example, P3.9 explained:

> "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'd know what to expect. They might know how worrying that would be. Whereas with this, it's very much me just saying, 'This isn't good.' [...] 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 it on a bit."

P3.13[u] told people she had a common condition, because it was easier for them to understand. She explained, *"It's easiest just to say Lupus than anything else because people can wrap their heads around that."* Several other participants admitted to downplaying or simplifying their illnesses in

conversations with other people. P3.13[u] 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.' "* P3.4 downplayed her own symptoms, but turned to her friends for additional support after her son was born with the same condition. She explains:

> "I never complained about it because it was like a part of me and 'Okay, that's who I am.' [My friends] never realized 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."

Participants had a variety of experiences in the volume and quality of support they received from their friend and sometimes this depended on how much they disclosed about their condition.

P3.5 found that, as long as he did not spend too much time *"whining about it"* and kept a positive attitude, people were *"extremely supportive and respectful and helpful."* P3.12 explained it was hard for her friends to understand because they see her as *"relatively well and positive."* She explained:

> "[P]eople don't really understand because they've seen me well and that's all they see really: that I'm well and I can do stuff [. . .] It is hard in that I know inside me the difficulties that I have because I have a physical disability [. . .] I've had to ask people to help me to get up and they sort of look at you as much and say, 'Why?' Cause I look alright..."

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. P3.10 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. I've really found out who those people are in my life. [. . .] When shit hit the fan, some of the people who you thought would really be there actually weren't."

3.2.2 Role in Relationships

Many participants leaned more heavily on their family members for support, because they were more aware and understanding of what was going on, having been exposed to it more throughout the diagnosis and management process. P3.11 explained that while her friends had never heard of the disease and did not understand her experience, her husband *"has been with me day in and day out, and so he really understands what I'm saying."*

> 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.

P3.10 explained his concerns about the impact of his disease on his wife. He said, *"we had to go through all of it [. . .] I know it's been hard on her. But there is a level of powerlessness that you feel. Like what do you do? You want to make it easier on them, but there are some things you can't do. It's frustrating."* P3.5 felt similarly guilty about the toll the condition took on his spouse. He said:

> "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."

> Other participants described needing support and having partners that had not yet accepted the new circumstances enough to be helpful.

P3.6 asked her husband 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.'"

Having a rare disease also complicated the experience of forming new (romantic) relationships. P3.4 had previously dated people that were uncomfortable with her condition. She explained:

> "[S]ome of my partners decided they couldn't handle [my disease] and it was better that they didn't continue the relationship. 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.'"

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. P3.4 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 [...] You take your life on one side and the life of your child on one side [...] We also take the medicine with an expired shelf life [...] The medicine with the good shelf life, we give to the children and if it's expired, some of the grownups take the medicine."

Relationships become more complicated when a person has a rare disease because dynamics, expectations, and resources change.

3.2.3 Role As Advocates

Participants spoke frequently of needing to advocate for themselves and for others with their conditions.

> Many participants described "broader awareness" as a goal. They were very actively engaged in political advocacy as well as educating their networks and the general public about their conditions.

Particularly in countries with predominantly public health systems, participants worked to improve access to medication. P3.3 described:

> "The associations [for rare diseases] are united and are pressuring the government to approve a series of new laws [...] By constitution, 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."

P3.4 was similarly engaged in advocacy and awareness campaigns, focusing more on awareness in the general public and funding for research. She explained:

> "Together with all the rare diseases we run with different t-shirts and the motto was 'We run for a better life.' So it was covered in the media so that we raise awareness of rare diseases [...] We had a concert to raise awareness [...] Last year we climbed [a mountain] together with the rare disease organizations in order to convince the government that, 'We can climb mountains. We can have children. We can live 100 lives. We can live 100 years. All we need is medicine.'"

3.2.4 Role As Patients

In addition to advocacy targeting governments and the general public, participants needed to advocate for themselves when dealing with medical professionals. They generally believed that doctors were unfamiliar with their conditions and unwilling to learn. P3.4 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."* Many recounted times they felt their care was poorly handled by physicians who were poorly informed about their conditions. P3.12 said:

> "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 didn't listen and 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.'"

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. P3.11 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."

For P3.3, this decision to abandon certain doctors similarly depended on how important the doctor was and how difficult they would be to replace. He explained:

> "If it's a doctor who happens to be new, I don't care and just change doctors. But if it's a doctor that I really need and don't have a choice, I need to try to talk to the doctor [...] The patient-doctor relationship is complex. It's not simple. And some doctors are resistant to learn from patients. But we try the best that we can."

P3.6 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. After P3.4 and her son vacationed in a different country where several doctors refused to treat them, she learned to administer shots herself. She described:

> "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.'"

P3.4 did eventually find a doctor who was willing to treat her and her son, and like many other participants, spoke highly of doctors who were willing to take the time to learn about unfamiliar conditions. She described a doctor who:

> "...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, P3.14[u] additionally had to enlist the help of friends and professors to convince a doctor her side effects were not just psychosomatic. She explained, *"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. P3.11 stated:

> "[The doctors] think you're crazy, like 'Oh, you don't know.' [...] 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 (P3.11) nevertheless maintained a strong distrust of doctors. She recounted:

> "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!'"

P3.3 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."* P3.14^{u} began actively researching her symptoms after her doctor said her symptoms were all in her head and she wanted to *"prove him wrong."*

3.2.5 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. Because of a series of misdiagnoses and conflicts with doctors, P3.14^{u} asked for personal copies of her records so she could manage them on her own. She used her lab results to research different conditions that could lead to a diagnosis. P3.9 said she kept a *"vague journal"* of significant occurrences (e.g., bad episodes, cramps) because her symptoms slowly progressed. P3.11 did not methodologically maintain her records, but tried 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 P3.6 kept a spiral notebook for personal use, she also found it helpful when she visited her doctor. She described:

> "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."

P3.13[u] described her *"medical résumé,"* a technique she picked up from her parents who were both medical doctors. She explained:

> "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, P3.7 made *"business cards"*. She explains:

> "I've had these cards made up through the [rare disease] 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, P3.7 uses them to connect with other patients for support. She explained:

> "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.'"

People with rare diseases document privately to keep track of personal medical experiences as well as publicly to communicate with the medical community and connect with other people with rare diseases.

3.2.6 Role As Copers

Participants in this study reflected 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. P3.5 chose to make the most of his time while he was able, because his physical condition was likely to worsen. He stated:

> "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."

P3.10 also talked about learning to accept his condition. He explained:

> "I do yoga. I meditate. I try to keep myself busy. I try to stay in a perpetual forward motion. [. . .] You could sit there and worry about the disease and while you sit there, your whole life is passing by. [. . .] I have realized 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. P3.6 described:

> " [. . .] 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. "

Some participants held a sense of optimism, such as P3.9 who explained, *"It was a big thing to understand and it sounded scary. And now I've realized, 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."* Maintaining this optimism was challenging sometimes. P3.9 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."* Participants were concerned about the progression of their condition and the availability of a cure. Even when a cure was unlikely, some participants hoped there would be more information available, explaining *"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."*

Some participants took the approach of reminding themselves it could always be worse. P3.2^{m} said, *"I always tell people it's not great having [my condition], but there's people with a lot worse."* P3.5 and P3.10 both had conditions that are commonly misdiagnosed as ALS. P3.5 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. [...] 'Wow, goodie! I'm going to make it here for a while!'"* P3.10 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 another condition 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."

Participants tried a range of strategies to stay positive throughout changing circumstances.

3.2.7 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. They found academic papers through searches of Google and especially Google Scholar. Some participants were successful in learning about their conditions through their research. P3.4 described how,

> "Dr. Google helped 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."

Most participants, however, found that the information they wanted was not available. P3.9 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."* P3.10 agreed, stating:

> "After your diagnosis, the first thing you are going to do is Google it, and read everything you can about what you have. What you find is very limited because there is no research being done. There is literally no research being done. Since there are so little people who have it, researchers have bigger fish to fry, like diabetes."

P3.12 felt that she understood her condition, *"about as much as I can because it is a mysterious thing."* She explained, *"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?"* P3.13^{u} also felt that she understood things as could be expected. She explained:

> "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 [...] 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."

Most participants described themselves as more research literate than the general population, citing some specific reason for their knowledge (e.g., an advanced degree in computer science or past career in drawing blood for medical tests) or a connection to someone with a medical background (e.g., a pharmacist friend, geneticist father, or researcher mother). 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 (e.g., P3.11 explaining, *"I present professional development to teachers for elementary science, so I know science."*) Participants described strategies they used to read research articles. P3.13^{u} explained, *"Reading research papers should not be scary to people. You can find a research paper, print it out, slowly read it line by line, bring it to your doctor, ask questions. People can do that. That's the great thing about the Internet."*

3.2.8 Role As Peer Supporters

Participants heavily relied on online communities as a way of sharing research findings and trading resources and support; this was unsurprising, given our decision to recruit via online communities. A few participants also participated in in-person groups or had encountered other people with their diseases at symposia, conferences, or medical association meetings. Participants actively sought out others with whom to connect and, as P3.2^{m} explained, they felt helped by *"that instant connection and knowing you're in company who have gone through the same thing."*

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. P3.6 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. P3.5 explained:

> "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 piped 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, including P3.12 who said, *"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."* P3.5 agreed, stating, *"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."* P3.11 even described how excited she was the first time she met someone with the same condition. She said, *"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?'"*

Because 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 P3.3, language was a barrier; he found it difficult to communicate with other people. For P3.7 however, the international community was a benefit, since it gave her access to research from other countries. She explained, *"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."*

Self-presentation was also a concern for participants; they were mindful that certain platforms were more appropriate than others for discussing health circumstances.

P3.11, for instance, was concerned about who would be able to see her posts on public social platforms and was hesitant to use a Facebook group. She explained:

> "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."* P3.8 explained that they *"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."*

In general, participants expressed that the benefits of these online communities outweighed the privacy risks. In some cases, participants even shared their actual medical information with each other via these communities. P3.10 described a disease-specific online forum where people post their blood test results and compared their own results to those of other patients to see similarities and differences.

Participants who used Facebook groups described more positive experiences than those who used the rare disease specific online communities. Participants in our study reported that there was little activity on the rare disease specific communities; we observed this throughout our recruitment process as well. Most participants from these separate sites had recently registered and were disappointed by the lack of activity. P3.1^{m} posted a question and did not receive any responses. She said:

> "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."

Even within communities that were active overall, participants wanted to connect with people who were like them beyond just having the same disease. P3.11 grumbled that, *"A lot of them are 75 to 80 years old, which is another problem because most of the people who have what I have are old - older than me."*

3.3 Discussion

Many of the issues uncovered here are extreme examples of what has been seen with more common diseases. We also found that people with rare diseases faced unique challenges that differ from previous studies of chronic illness populations.

3.3.1 It's Who I Am

A notable difference between people with rare diseases and people with common chronic illnesses involves their self-perceptions in relation to their conditions. Recent work in HCI encourages considering about *people* rather than patients, emphasizing humanness rather than disease [14, 158, 211, 214]. Ballegaard et al. [14] explain:

> "The healthcare professionals and caregivers somehow presume that the main goal for the elderly is – almost at all cost – to avoid diseases and health related problems. While this approach has clear benefits [...] [t]he clinical approach should be supplemented with an approach that gives voice to the interests of the citizen. For most people health and disease are just some aspects of life. The people in our case studies explain that their everyday life as a whole is what matters: e.g. spending time with their family and friends, having an interesting job or exciting hobbies. Health and healthcare technology are just small pieces that they try to fit into the larger puzzle of the everyday routines."

To the contrary, participants in our study regularly made statements saying, *"It's who I am"* (P3.4) or *"This is part of my story"* (P3.10).

> Participants identified strongly with their conditions, speaking of themselves as ambassadors, as if they could not be understood apart from their medical conditions.

There are several implications here. Most simply, there may be opportunities to 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. However, 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 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 [22]. There is value in investigating the 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 people with rare diseases, to effectively and ethically raise awareness.

There is a secondary implication here: identifying with a rare disease to the detriment of other facets of one's identity can cause stress on a relationship [117]. Researchers have suggested there are benefits to taking on multiple roles or statuses [302] and people with rare diseases may be missing out on these benefits if their rare disease has become a "master status" (one that *"tends to overpower, in most crucial situations, any other characteristics which might run counter to it"* [157]) or even more specifically a master "sick role" [267, 354] that it interferes with relationships with others. Friedson [117] explains:

> "A chronically ill or permanently impaired person who 'expects too much' or 'makes too many demands' is likely to be rejected by others [...] legitimacy [of the condition] is conditional on limiting demands for privileges to what others consider appropriate."

These different identities and senses of self [330] are further complicated when rare disease online communities are couched within larger social networks with mixed audiences and collapsed contexts [27, 346].

3.3.2 Leveraging Existing Platforms

Unlike in prior research with people with common chronic illnesses, we found that separate online communities were ineffective at engaging participants.

> The disease-specific communities that have the been the subject of prior work were not actively used by people with rare diseases because they lack the critical mass necessary to be successful. Instead, communities within Facebook allowed participants to find people with similar experiences.

People with rare diseases are very active users of technology and existing social platforms [326]. We should consider integrating rare disease communities into platforms people are already using. It may be possible to address the privacy concerns expressed by participants in our study, through

closed social media groups. It is also 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.

3.4 Summary

This chapter provides researchers with a glimpse inside "Rare World" where people with chronic rare diseases take on many roles, from educating to advocating to providing support for people within their immediate and broader community. Participants were concerned about the toll their disease takes on their caregiver network. They constantly have to educate, advocate, and research their disease to ensure that they can effectively communicate 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 people accept some privacy loss to connect with people with similar illnesses on popular social media platforms. The experiences presented here highlight challenges and opportunities that could be addressed with sociotechnical systems.

“ *If you hear hoofbeats around here,*
you can usually bet they're made by horses, not zebras.
Fortunately, when somebody gets sick,
the trouble is usually something common,
not one of those exotic diseases they dramatize in medical schools. ”

— Dr. Hilton Read, quoted in the Reader's Digest, May 1964

Chapter 4

Comparing Rare Diseases & Common Chronic Illnesses

Themes presented in Chapter 3 are reminiscent of existing research on common chronic illnesses, such as the reliance on family members in navigating a new diagnosis [64, 155, 175, 231] or challenges in interacting with clinicians [281]. Other themes, such as identifying strongly with a condition or working to raise awareness, seem more unique (or at least more noticeable) in rare disease populations. People with common chronic illnesses do not seem to face the same battles in seeking a diagnosis, finding information, and managing changing relationship dynamics. We set out to systematically investigate differences between people with rare diseases and people with common chronic illnesses using a survey informed by the results described in Chapter 3 that we distributed to both populations.

Much of what we know about health is the result of rigorous medical research. Recent research however has seen value in using social media data to identify large scale public health patterns such as influenza spread [173, 291, 303], detecting depression [81, 329], or Ebola outbreaks [297]. Additional research has been able to identify drug-drug interactions or adverse drug events from Twitter [142, 293] or Instagram [68]. These studies all rely primarily on people's posts about clinical information (i.e. drug names, disease/condition names, symptoms). In this chapter, we begin to explore the potential of *behavioral* data in identifying and understanding people with rare diseases. That is, rather than looking for social media posts about a disease's symptoms or treatments, we consider how people use the Internet and social media to access health information and support.

Some researchers have taken a similar behavioral approach to identifying people with a specific health condition (e.g., research by Saeb et al. [292] found that GPS and usage data from cellphones were strongly related to the severity of depressive symptoms). These types of approaches allow us to learn a great deal about conditions that are otherwise under-reported (e.g., because of stigma). Identifying people with rare diseases presents an additional challenge because we are not targeting one single rare disease but instead are targeting a large class of diseases united by rarity but having wildly different symptoms. This approach allows us to identify people with diseases about which very little clinical information is known and ultimately provide them with social or informational support, without needing to know specifically what disease they have.

In this chapter, we discuss the design and distribution of our survey, exploring the differences in the responses of people with rare diseases compared to people with common chronic illnesses. This provides further insights on the nature of the differences between the experiences of people with rare diseases and their common chronic illness counterparts. We apply advanced machine learning techniques to effectively predict the occurrence of rare diseases. This confirms many initial insights from Chapter 3 about the differences between the two categories of diseases and lays a foundation on which we could begin to identify people with rare diseases from their online behavior and better direct them to information or social support. Overall, we learn that people with rare diseases are highly engaged online when it comes to health issues. Their use of online communities for support is different enough from people with common chronic illnesses to merit further investigation. This study was conducted collaboratively with Shuo Yang, Kim Oakes, Kay Connelly and Sriraam Natarajan, and this chapter includes material from our CHASE 2016 paper [209].

4.1 Method

We distributed a survey to understand differences between the behavior of people with rare diseases and common chronic illnesses (IRB#1410636078A001– see Appendix A).

4.1.1 Survey Design & Distribution

We selected questions based on findings from Chapter 3, as well as existing literature on common chronic illnesses. These questions were adapted from a similar Pew Internet & American Life Project

survey [116] but tailored the questions to our specific research questions, removing questions that seemed irrelevant and modifying questions to better incorporate prior knowledge. The result was a survey containing 35 questions covering four themes: demographics, technology use, information seeking, and perception of health care professionals. A copy of the survey is included in Appendix C.

We tested the survey internally to identify problems and estimate completion time. We distributed the survey to 26 Facebook groups for people with chronic illnesses as well as to Reddit, targeting eight subreddits of specific chronic illnesses. We did not share the recruitment notice outside of these chronic illness groups because we did not want to create additional privacy risks by encouraging people to indicate to a social network that they had a chronic disease if they had not already done so (by their participation in a disease specific group).

4.1.2 Survey Responses

We received 341 responses and omitted 13 incomplete responses (leaving 328 complete responses). Disease names were entered as text by survey respondents (in many cases respondents had multiple co-morbidities and listed these in the text box). We discretized disease names as one or more rare diseases (30.34%), or one or more common chronic illnesses (60.66%). For the six respondents having at least one common chronic illness *and* at least one rare disease, we created duplicate entries, labelling one as common and one as rare. We did not have enough of these examples to treat them as a third category, but believed they may have characteristics of either group.

Respondents ranged in age from 18 to 71 ($\bar{x} = 31.71, s = 12.71$). We discretized these into bins. The number of years the respondent had been experiencing symptoms and the number of years since the respondent had been diagnosed were similarly discretized into bins. The data set consisted of responses from 39.00% male respondents and 59.53% female respondents (1.46% of respondents did not identify with either gender). We received responses from 22 different countries, primarily English speaking countries like the US (58.46%), Canada (12.46%), UK (10.39%), and Australia (8.01%); our survey was only distributed in English. Other countries included Czech Republic, Denmark, France, Germany, Hungary, Israel, Latvia, Malaysia, Mexico, Netherlands, New Zealand, Norway, Poland, Portugal, Romania, Russia, Sri Lanka, and Sweden. Many of the questions in the survey asked the respondents to "check all that apply." Each of these checkboxes was treated as a binary feature (checked/unchecked). This provided us with a total of 70 features.

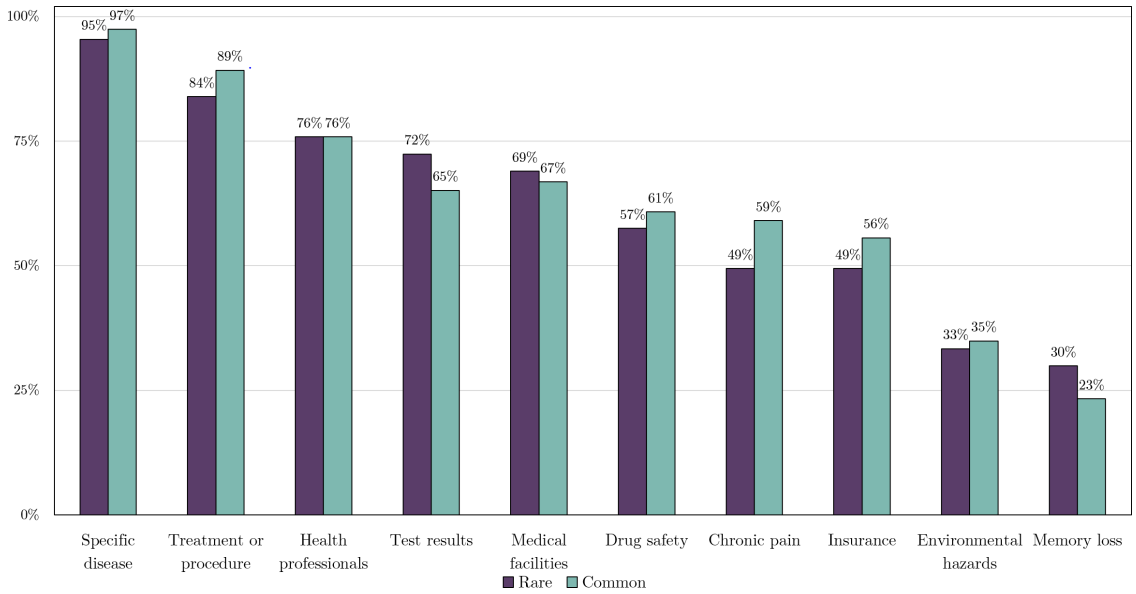


Figure 4.1: Responses to *"Have you ever looked online for information about any of the following?"* by rare disease and common chronic illness respondents.

4.2 Results

In this section, we explore the differences in survey responses from people with rare diseases and those from people with common chronic illnesses. We investigate how they seek information online, use digital technologies to support their health, and use social networks for health purposes. We conclude by applying advanced machine learning techniques to identifying people with rare diseases from their self-reported behavioral data.

4.2.1 Information Seeking

To understand the information people with rare diseases and people with common chronic illnesses search for online, we conducted chi-square tests for each information type. Here, our independent variable was *disease type* (rare/common). Our dependent variable was whether or not the respondent had ever looked online for information about the topic (yes/no). We found no significant differences in the types of information the two populations were searching for online. The vast majority of respondents (see Figure 4.1) reported using the Internet to look up *"information about a specific disease or medical problem"* (95.70% of people with rare diseases and 97.48% of people with common chronic illnesses) and to look up *"information about a certain medical treatment or procedure"*

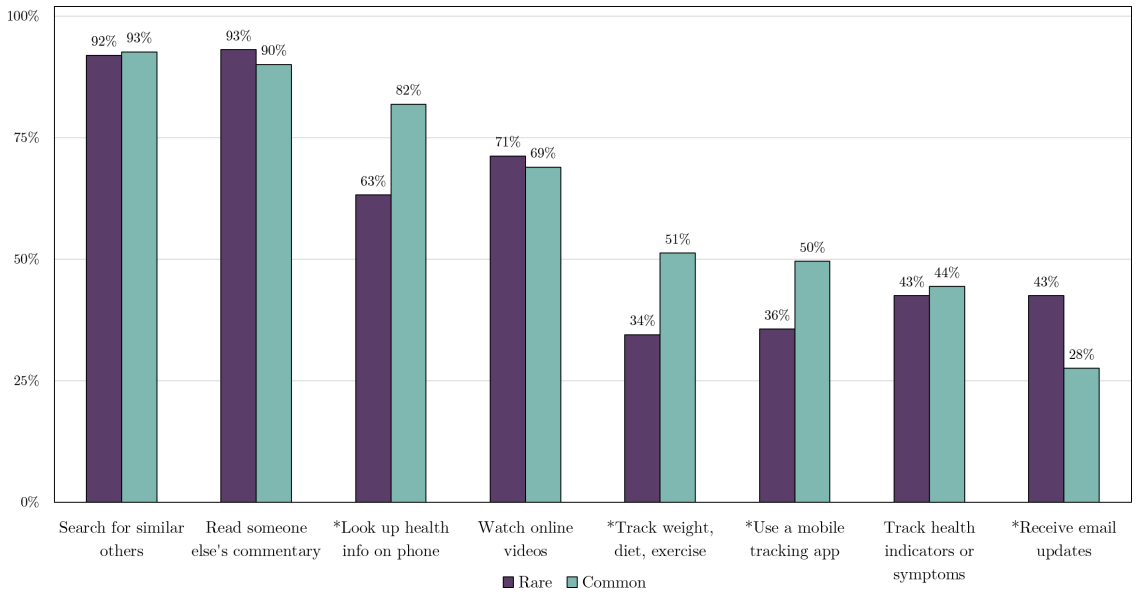


Figure 4.2: Popularity of different online behaviors by rare disease and common chronic illness respondents. Asterisk indicates significant differences.

(83.87% rare diseases, 89.08% common chronic illnesses). Using the Internet to look up information about *"environmental health hazards"* (34.41% rare diseases, 35.29% common chronic illnesses) and *"memory loss"* (30.11% rare diseases, 23.53% common chronic illnesses) was less common.

4.2.2 Online Behaviors

We similarly conducted chi-square tests to understand differences in online behaviors between the two groups. We found few differences when considering self-reported online behaviors (see Figure 4.2). People with rare diseases and people with common chronic illnesses were both likely to have used the internet to *"read someone else's commentary or experience about health or medical issues on an online group, website, or blog"* (92.47% rare diseases, 89.92% common chronic illnesses) and have *"gone online to find others who might have health concerns similar to yours"* (91.40% rare diseases, 92.44% common chronic illnesses).

There were three areas where people with common chronic illnesses were significantly more active than people with rare diseases. People with common chronic illnesses reported using a mobile app to track or manage their health more more often than people with rare diseases ($p < 0.05$, $\phi = 0.12$) and they used their phones to look up health information more often ($p < 0.01$, $\phi = 0.19$). They also tracked their weight, diet, or exercise routine online more often ($p < 0.01$, $\phi = 0.14$). The

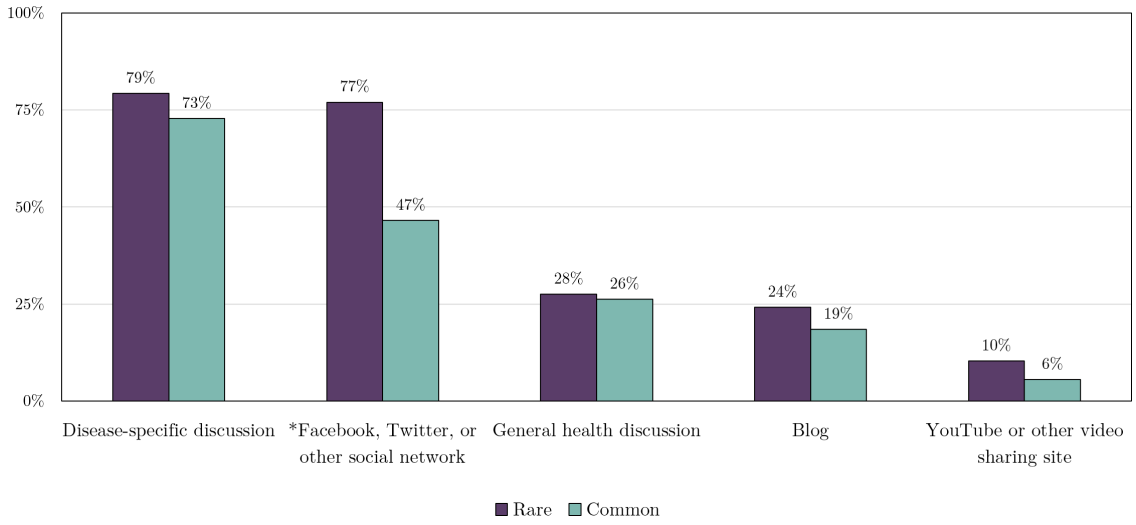


Figure 4.3: Responses to *"Have you posted comments, questions, or information about your health or medical issues on any of the following?"* Asterisk indicates significant differences.

only area where people with rare diseases were noticeably more active was in signing up to receive email updates or alerts about health or medical issues ($p < 0.01$, $\phi = 0.15$).

4.2.3 Social Media Use

Where we saw the biggest differences between rare disease and common chronic illness populations was in their preferred social media sites (Figure 4.3) and use of these sites (Figure 4.4).

> People with rare diseases were much more likely to have posted about their health on a social media sites like Facebook or Twitter ($p < 0.01$, $\phi = 0.27$). People with rare diseases also described using these mainstream social networks much more actively for a variety of tasks than people with common chronic illnesses.

Rare disease respondents were more likely to use online social networks to get health information ($p < 0.01$, $\phi = 0.19$), to start or join a health group ($p < 0.01$, $\phi = 0.28$), to follow other people's health updates ($p < 0.01$, $\phi = 0.21$), and to memorialize someone ($p < 0.01$, $\phi = 0.19$).

4.2.4 Identifying People with Rare Diseases

In this section, we are interested to learn whether advanced machine learning techniques, when combined effectively with data about the experiences of people with different health conditions

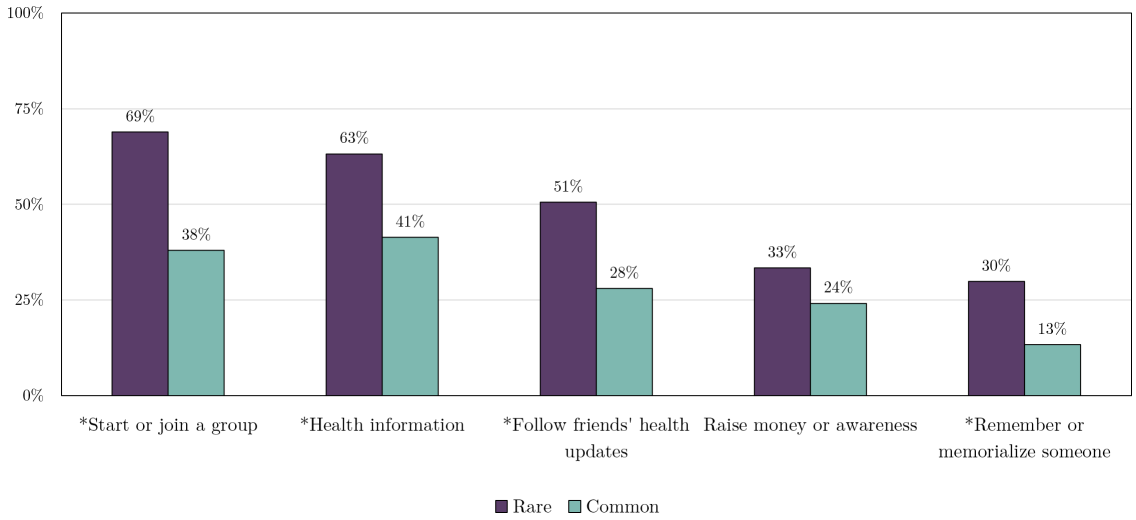


Figure 4.4: Responses to *"Have you ever used a social networking site to do any of the following?"* Asterisk indicates significant differences.

(informed by our domain expertise acquired in Chapter 3 and collected from our survey described here), can effectively predict the occurrence of rare diseases.

The challenge here is that, given the rarity of the diseases, our survey responses are skewed towards people with common chronic illnesses and we have a small number of rare disease examples (i.e., we have more responses from people with common illnesses than we do from people with rare diseases). This problem is called class imbalance in machine learning [49]. If we consider people with a common chronic illness as one class and people with a rare disease as another, there is a huge skew towards people with common diseases. Simply adopting standard machine learning techniques (e.g., decision trees, logistic regression, naïve bayes) can achieve high accuracy by predicting all the examples as the majority class (common chronic illnesses). Thus, to conduct our analysis, we rely on an advanced machine learning classifier (Soft-FGB) that can handle this small, imbalanced data as part of the classification process [362]. Additional details on Soft-FGB and analyzing imbalanced data can be found in Chapter 7.

The nature of class imbalance problems is that the cost associated with misclassifying the rare class (in our case, rare diseases) is higher than misclassifying the common class (common chronic illnesses). The cost matrix does not contain uniform cost values but rather has a higher cost for the instances that are rare diseases but classified as common chronic illnesses than for those that are common chronic illness but are classified as rare. If we assume the class of rare disease as the

Evaluation Metric	Soft-FGB Results
True Positive Rate	0.972
False Negative Rate*	0.028
F_3	0.825
F_5	0.909

Table 4.1: Results of applying Soft-FGB to survey data. Asterisk indicates metric should be minimized. All other metrics should be maximized.

positive class, then we prefer recall (how many relevant items are selected) rather than precision (how many selected items are relevant). Thus, we focus primarily on evaluation metrics that assign higher weights to higher recall regions. Specifically, we consider the true positive and false negative rates, as well as two additional metrics: F_3, and F_5, where this F-measure is:

$$F_\beta = (1 + \beta^2)\frac{Precision \cdot Recall}{(\beta^2 \cdot Precision) + Recall}$$

where β is the importance given to recall over precision (i.e. a higher β indicates more emphasis on recall, a smaller β indicates more emphasis on precision). We use F_3 and F_5 to increase the importance of recall over precision.

We use Yang et al.'s [362] Soft-FGB method to account for the sample size and the skew in the data. Additionally, we use 10-fold cross validation to assess the generalizablility of our model. That is, we partitioned our data into 10 subsets and conducted the analysis 10 times. At each iteration, we treated one partition as the test set and nine partitions as the training set. We achieved a true positive rate of 0.972, and a false negative rate of 0.028. Our F_3 and F_5 scores for this analysis are 0.825 and 0.909 respectively. Results are provided in Table 4.1.

4.3 Discussion

Our work in Chapter 3 indicated that people with rare diseases take on a very active role in managing and seeking information about their health. Given the rarity of the conditions, people with rare diseases do not often have in-person access to others with the same conditions, so they turn to online communities to connect with others. We suggested that disease-specific online communities were ineffective because of a sparsity of participants, and that mainstream social media sites were more

popular and effective. Our analysis in this chapter supports those earlier intuitions; people with rare diseases describe actively using mainstream social media sites, such as Facebook or Twitter, to engage in health activities (more than separate disease-specific health forums). They rely on these social networks to get health information, find groups, raise awareness, etc. People with rare diseases are known to contribute to these online communities by posting their own data/test results and talking extensively about their experiences [120, 274, 352]. People with common chronic illnesses do not have the same need to engage in this online support and information seeking behaviour, because there is more information available through medical professionals in the first place and more support available locally. It seems reasonable that many people with common chronic illnesses would be less likely to have joined a health group on a mainstream social network, less likely to have needed to raise money or awareness for their condition, less likely to have memorialized or remembered someone suffering from a health condition.

There are a few areas where people with common chronic illnesses *do* use technology or the Internet to manage their health; people with common chronic illnesses more actively use their phones and smartphone apps to track or look up health information. We suspect this is because many of the mobile health applications available commercially are designed to target the symptoms, causes, and management of specific chronic illnesses. These conditions are well studied in the medical literature and have a specific and known set of symptoms and treatments, so technologies are customized to specific illnesses. The equivalent applications for rare diseases simply do not exist.

4.4 Summary

The results of this survey study confirms many of our insights about the differences between people with rare diseases and people with common chronic illnesses (informed by the interview study presented in Chapter 3). People with rare diseases and people with common chronic illnesses have different experiences; these experiences are so different that we can use machine learning to effectively pick out people with rare diseases from a mixed data set. If we can effectively identify people with rare diseases, we can begin to provide them with better access to social support. In extensions of this study, it would be interesting to aim to identify people with rare diseases among healthy populations. Our current study focused on the distinction between common chronic illness

and rare disease populations, but it may be interesting to identify those groups from broader social media data. We can imagine that even people who do not have a chronic illness diagnosis may still engage in health information seeking online, if they are interested in general wellness for themselves, or are seeking information for a close friend or family member.

An additional limitation of our approach is that it excluded anyone undiagnosed; everyone who completed the survey knew the name of the condition(s) they had. Future extensions of this work could also considered people who may have a rare disease, but are not yet formally diagnosed. These are people who are likely to be active information seekers online but do not typically have a disease name to work with; including this population in future iterations of this research would allow us to build models that can identify them and help guide them towards additional support. Including people who are undiagnosed may also lead to insights on the differences between people who are pre-/post-diagnosis, thus better understanding the impact of a diagnosis on one's life.

Our survey asked respondents to reflect on their online behaviour and sources of support which may be different than actual behavioral data that could be gathered in real-time. Future work could triangulate this self-reported data against *actual* behavioural data (possibly collected from social media or browser histories as in previous work [68, 81, 263, 292, 328]. Training a similar model on actual behavior would bring us closer to being able to identify people with rare diseases on social media sites and guide them towards appropriate support communities. However, there is an ethical consideration here as well: what are the consequences of diagnosing someone with a condition for which there is very little information and in many cases no treatment or cure? Better understanding the differences between people pre- and post-diagnosis may help navigate this dilemma.

The results of this study also suggest that people with rare diseases are highly engaged online and many of them make use of existing social networks for health purposes. They rely heavily on online sources to search for similar others, read about other patients' experiences, participate in support groups, and follow other people's health updates. In Chapter 5 we seek to understand the *types* of support addressed (or not) by these online support sources. We examine the relationships people with rare diseases have with other people with rare diseases (i.e., those also inside Rare World) and with their healthy friends and family members (i.e., those outside of Rare World).

“ *That's me. And that's three people. And I'm going to help them.*
But it has to be something really big,
something they can't do by themselves. So I'd do it for them.
Then they'd do it for three other people. That's nine.
And nine do three more... ”

— Trevor McKinney in *Pay it Forward*

Chapter 5

Relationships Inside & Outside of Rare World

In Chapter 3, I described the experiences of people with rare diseases, including the impact of a rare disease diagnosis on one's relationship and perspective on the world. In Chapter 4, I explored differences between the experiences of people with rare diseases and people with common chronic illnesses, finding that people with rare diseases rely heavily on online sources for support. In this chapter, I use a group-based approach to triangulate findings from earlier studies and to explore sources of support and support relationships in more depth. I outline the differences between relationships participants have with people within Rare World (i.e., people who also have a rare disease) and outside of Rare World (i.e., healthy people or people with common chronic illnesses). I identify support needs that are unmet and begin to consider possible new sources for this support. This study was conducted collaboratively with Grace Bastin, Leslie S. Liu, Katie Siek, and Kay Connelly. This chapter includes material from our CHI 2017 paper [203].

5.1 Method

For this study, we developed the Asynchronous Remote Communities (ARC) method. ARC is a group-based, asynchronous method that can be conducted remotely with dispersed populations (described in greater detail in Chapter 8.) We provided participants with ten activities (briefly summarized in Table 5.1) via a private Facebook group for the study over the course of 22 weeks. This study was IRB approved (Appendix A - #1412065553R001).

Activity	Description	Shared	Dur.
Diary	Participants tracked interactions they had with other people about their disease. They documented who they talked to, how they communicated, what they discussed, and how they felt about it.	Directly	24 hrs
Circles	Participants used paper clips and other small household objects to illustrate comfort sharing information with different people by drawing circles with themselves at the center and placing people at different distances from the center.	Directly	Once
Questions	Participants made a list of questions they wished their friends/family would ask them about their disease.	Directly	Once
Problems	Participants ranked a list of problems (provided by the researchers) by the relevance to them personally.	Directly	Once
Photo Elicitation	Participants took photos representing two main themes from the problems activity and commented on each others' photos.	Anon.	3 days
Solutions	Building from the problems activity, participants discussed strategies or solutions they had used to address these problems.	Directly	Once
Mad Lib	Participants created a fill-in-the-blank paragraph, detailing the more humorous aspects of conversations they have surrounding their conditions.	Directly	Once
Movie Script	Participants wrote a script for a movie about their life, taking place between them and a friend or family member.	Directly	Once
Rant Line	We set up a "Rant Line", a Google Voice number that participants could call, text, or send photos to anytime they needed or wanted to rant about something.	No	3 days
Personas	We created and posted two personas and asked participants to critique them and discuss how representative they were of their own lives.	Directly	Once
Survey	Participants completed a debrief survey about the study.[1]	Directly	Once

Table 5.1: ARC study activities. **Shared:** Responses were shared *directly* to the group, collected by researchers and *anonymized* before being shared, or were *not* shared with the group at all. **Duration:** Period of time over which the activity occurred.

[1] This survey was intended to evaluate the ARC method, but is included in in this chapter because in reflecting on their experiences in the study, participants also reflected on their interactions with one another.

In addition to these activities, participants independently started new conversations among themselves and these posts were included in our analysis.

5.1.1 Participants

We recruited adults with a rare disease from Facebook support groups because Facebook is actively used by people with rare diseases as a way of connecting with each other (see Chapter 3) and has been shown to be an effective mechanism for recruiting people with rare diseases for research [79, 295, 326, 340]). We did not recruit from other sources because we did not want to create additional privacy risks by encouraging people to share information about their condition on Facebook who were not already doing so. Where possible, we sought permission from group moderators prior to posting our recruitment notice.

There were 13 participants in our study group, although two participants did not engage in activities or discussions with others. Participants had one of five different rare diseases (Table 5.2) and ranged in age from 32–68 ($\bar{x} = 48.9$, $s = 12.8$). Eleven participants identified as female and two as male. All participants were from the US, except one from Australia. Two participants were employed full time, while the rest were either unemployed, receiving disability payments, or retired at the time of the study. They were each offered a $50 honorarium for their participation in the study, regardless of their level of activity.

Disease Name	**N**
Osteonecrosis	3
Ehlers-Danlos Syndromes	4
Hereditary Angioedema	2
Inclusion Body Myositis	1
Kallmann Syndrome	3

Table 5.2: Participant Diseases

5.1.2 Analysis

We iteratively coded all data from the study group for a range of themes, focusing on participants' relationships with friends and family members and their relationships with other people with rare diseases. Three researchers participated in coding this process, ultimately converging on an inter-rater reliability (Cohen's Kappa) score of 0.72. A summary of our codes is provided in Table 5.3. In this chapter, we do not discuss codes with low prevalence (i.e., where they were only discussed by a small number of participants) or where there was a heavy skew towards a single participant (i.e., where a topic was discussed extensively by one participant but very rarely by

Code Name	# of Participants	Total Comments
Outlook & Self-Image		
Identity - Acceptance	7	16
Identity - Positive	9	20
Identity - Negative	7	17
Mourning One's Old Life	4	11
Adjusting Expectations*	4	6
Expectations to Feel a Certain Way*	4	6
Jealousy*	3	4
Relationships Outside Rare World		
Absence of Relationship	6	23
Emotional Distance from Others	9	34
Belief/Validation	8	43
Listening	9	25
Passive Disinterest	8	28
Active Denial	6	22
Unhelpful Comments	9	32
Practical Support	7	28
Recognizing Limitations	8	16
Guilt/Burden*	4	4
Wanting an Apology*	2	2
Relationships Within Rare World		
Support from Other Groups	7	15
Comments about Other Participants	5	8

Table 5.3: Codes used in analysis. Number of participants counts participants who made at least one comment classified as that code. Total comments refers to all comments classified as that code. Codes with asterisks are not discussed because of low prevalence.

others). As in Chapter 3, we conducted a member check [193], giving all participants a chance to review our CHI 2017 paper [203] before publication to ensure they were comfortable with the content. No participants requested changes. Many quotes in this chapter (taken from participants' written submissions) contain typing mistakes or spelling errors. We present them as written by participants to ensure the narrative voice of the participant is not interrupted. In a few instances, it was necessary to edit quotes for anonymity or clarity; these edits are indicated with [ ].

5.2 Findings

Chapter 3 broadly describes the experiences of people with rare diseases and a phenomenon called "Rare World." People face additional challenges as a result of a rare disease diagnosis, which influences their perspective on the world around them. We found this concept pervasive in the study described in this chapter as well. Here, we more formally define "Rare World" to mean that people with rare diseases compare themselves to the "normal" people in their lives (i.e., healthy people or people with common chronic illnesses), leaving them with a feeling of isolation.

We begin with a brief description of the outlook and self-image of participants from our study (bolstering findings from Chapter 3). We then extend this work to focus specifically on people with rare diseases' perspectives on their relationships with people outside of "Rare World." Finally, we discuss the relationships people build and the support they receive from within Rare World (from others with a rare disease).

5.2.1 Outlook & Self-Image

Having a rare disease was a significant part of a participant's identity. P5.10 described telling her son she was *"sharing about a main feature in our lives and something we were proud of."* This echoes findings from Chapter 3 that people with rare diseases identify strongly with their conditions.

> Given the work that goes into living with a rare disease (navigating the medical system, staying up to date with research, raising awareness, advocacy initiatives, etc.) as well as the severity and/or persistence of some of these conditions, it is understandable that it becomes an easy thing on which to fixate.

Participants agreed with this characterization and wished it was not the case. P5.9 explained:

> "I tried not talking about my disease at all or how I feel or what's happening to me medically, because when I respond to their questions of 'how are you doing?' with honesty, their next comment is always that I am dwelling on my illness and I need more activities in my life. To which I always agree. And feel discounted every time it happens."

Even within this strong sense of identifying with the condition, we observed variance in the *valence* of participants' outlooks on their conditions. Although previous research has found that people with chronic illnesses have different needs and outlooks based on their illness trajectory [104], we saw that participants in our study did not fall into a particular mindset based on the progression of their disease (i.e., someone who had been diagnosed with a disease for years did not necessarily feel more accepting of it than someone who was newly diagnosed). They even shifted between mindsets over the course of the 22 week study. Of the 11 participants who were active throughout the entire study, we observed a range of outlooks in all them (see codes for positive, negative, and accepting identities in Table 5.3).

Some participants managed to reach stablility, maintaining an attitude of doing the best that they could and acknowledging their limitations without being defined by them. When faced with insulting or unhelpful comments, accepting participants described laughing it off and making a joke of it. They were not necessarily "proud" of their condition, but had learned to deal with it and accept it. For example, P5.8 joked, *"I don't mind feeling abnormal! I felt normal once...it was the strangest 20 minutes of my life!!!"* Other participants demonstrate a very positive mindset, not only accepting that they were dealing with a new normal, but also wanting to *go beyond accepting* by helping others who were struggling, raising awareness or advocating for the condition, conducting research about the condition, and engaging in other proactive and positive behaviours.

Finally, we saw that some participants saw their disease as a factor that isolated them from the rest of their family and friends. These participants saw the disease as extremely inhibiting. As P5.6 put it, the disease *"pretty much destroyed my whole world."* Even for activities that asked participants to take a more lighthearted approach (such as the mad-lib activity), some participants struggled to look on the bright side. P5.9 expressed, *"I couldn't in anyway make this funny. There is nothing about my disease that is humorous, nothing about my life with it that is humorous. I'm sorry."* These participants seemed to adjust poorly to the limitations their disease now placed on their daily activities, as they were sometimes jealous of those who were healthy and often mourned the loss of their old life. P5.2 explained:

> "I used to have all the nice crap. I owned a condo full of antiques, I drove a brand new BMW, I had amazing jobs, a million friends, NOW, 3 years later, I had to declare bankruptcy, my parents are kicking me out of their basement..."

Even further, participants in these negative mindsets had a hard time relating to people with more positive attitudes; they even felt resentful at time. P5.9 explained:

> "The attitude of 'I have the disease but the disease doesn't have me' [...] when I'm really, really sick and battered by my disease I am resentful of people who can say that. I desperately want a more positive outlook on life, but when you're constantly in the hospital, with incredible pain and misery, I just can't relate."

5.2.2 Relationships Outside of Rare World

Some participants had family members that were supportive, such as P5.10's husband who was a *"dedicated carer the past 11 years and rarely complained."* This was especially in cases of hereditary conditions where a participant's parent also had the condition, as in P5.7's case where her friends and family were supportive because *"they kind of understand that Dad passed this to me :("*

> For the most part, participants described wanting a range of types of support from those friend/family relationships and did not feel that need was being met. In many cases, relationships had broken down over time and many participants attributed this to the rare disease experience.

They made comments like, *"I've been abandoned by most of my friends. Nobody comes visit, nobody calls me. It gets very lonely and very depressing"* (P5.12) or *"Being sick all the time has ruined family relationships, friendships, and put a severe strain on my marriage"* (P5.8). Sometimes this isolation was something participants did to themselves. P5.11 explained, *"I have gone through times where I have insulated and isolated myself because I have felt so different from others."* This isolation and loneliness was evident through participants' stories and discussion, but is also visually discernible through their responses to the circles exercise; an exemplar from P5.9 is provided in Figure 5.1 along with the participant's description of her submission.

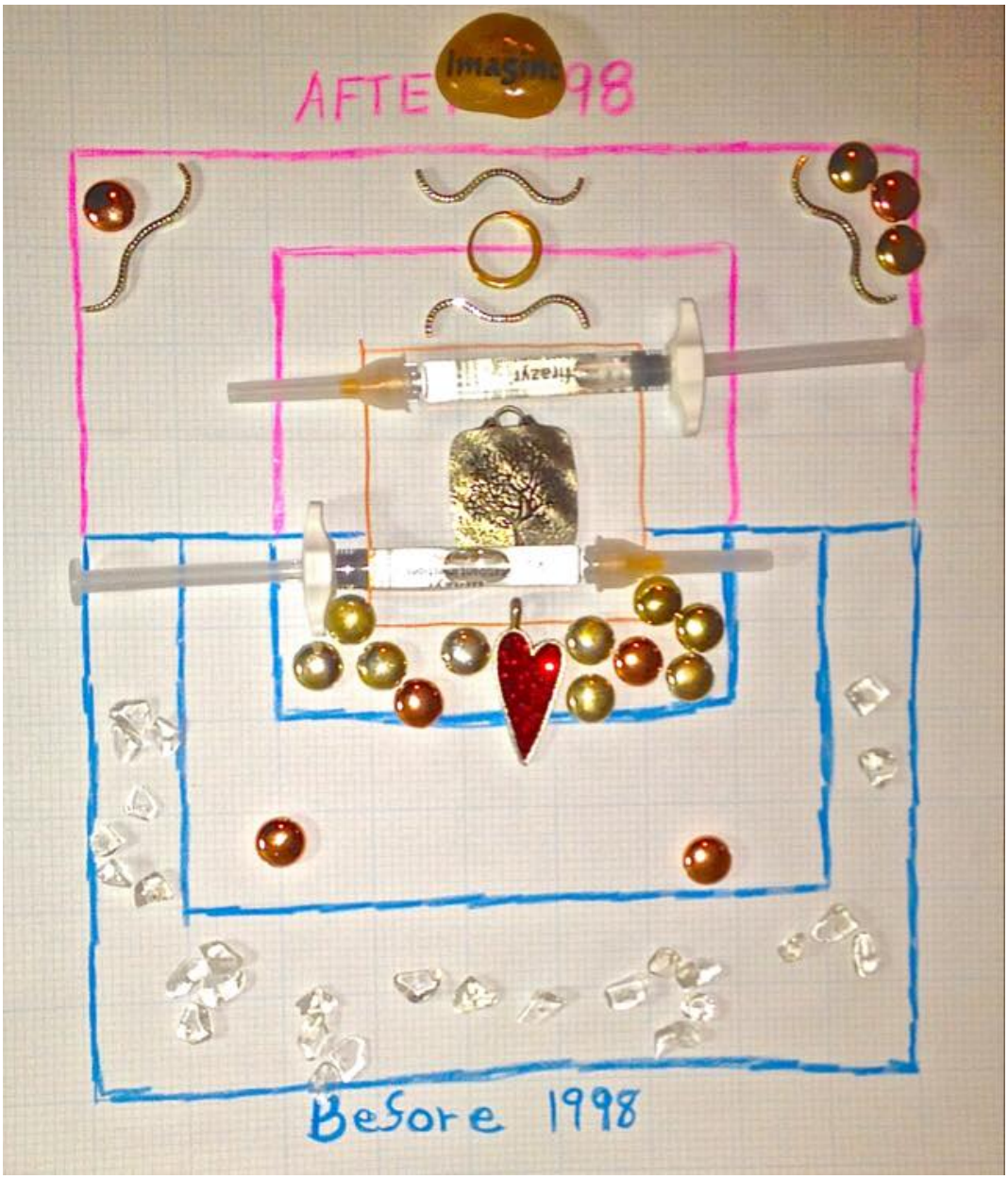


Figure 5.1: P5.9's Response to the circles exercise, showing closeness of different relationships.

"Circles just don't seem to fit for my life, it just feels that squares, and everything that means, are more appropriate. The overlap in the middle is a medallion with a tree in pewter. That represents me. The upper part in rose represents my life now, and the blue represents the past before my disease got worse in frequency of attacks. The injection vials are the medication I have to take at least once a day sometimes three times a day. To keep me out of hospital. They block off not only my past but my future. The heart in red represents my marriage, which is in the past as my husband and I have been separated for almost 6 years largely because of this illness. The ring that is barricaded in silver wavy lines above the pewter medallion is my wedding ring that I don't wear now and may never wear again. The silver and gold stones in the blue area near the heart represent my two children, they are 43 and 45 years old now, and my two grandsons aged 19 and nearly 16. Plus the seven cats and dogs that were so dear to us when over the past 31 years. The two bronze stones in the blue area below all of that represent my sister and my cousin with whom I used to be close. At the very bottom the clear stones represent all of the friends and work associates that I really enjoyed being able to see nearly every day. At the top left in pink that is also barricaded behind silver is a stone representing my son as we are estranged and have been for many months. The three stones on the right that are barricaded by silver represent my daughter and my two grandsons. We have been estranged since the end of October last year."

Nurturant Support

Participants discussed a lack of emotional, esteem, and network support. P5.9 expressed:

> "What I need is the emotional support. When the disease is really bad and I am terribly debilitated, hardly able to walk and in great pain, I just need someone to give me a hug and tell me they love me and that it will get better. I haven't had that in my life for the last 10 years, and even less so in the last six months."

Participants regularly felt like people just simply did not care about them or were disinterested and disengaged. P5.10 lamented:

> "How about someone in my family actually asking and remembering the name of my disease? And how about just one of them wants to know what it is, how it affects me, what it's trajectory is, what is likely to happen to me, and is it hereditary? None of those questions have ever been asked of me the past 11 years."

Often, this desire for nurturant support was related to legitimation of their experiences. P5.5 explained, *"I feel like a cure is something that is a priority but just knowledge to people about the diseases would be just as satisfying for me. Having people know what it is and taking it seriously."*

> Participants suspected that others did not believe they were sick, believed they were sick but not with a rare disease, or thought they were exaggerating their symptoms.

P5.2 expressed that his *"mom is in denial and tells me I am not disabled."* Participants regularly commented that doctors, friends, and family members alike all accused them of being hypochondriacs (*"I get treated as if I'm a know it all and a hypochondriac sometimes too"* (P5.5)). Additionally, they heard accusations of making up the disease to get attention (*"If I had a nickel for every time that I over heard people saying that I was just seeking attention (that one really hurts >()"* (P5.3)). Some participants expressed that their family members felt that because the disease was rare, the participant probably did not have it. P5.9 listed the kinds of things she heard from her family members:

> "'It's a very rare disease so you can not have it.' [...] 'I don't think that's a real condition.' [...] 'Well, I think you need to see a psychiatrist. Because I think you're just faking this 'rare' disease.' [...] 'You are costing the medical system millions of dollars a year that could go for treatment for real diseases! Why don't you go to another doctor and find out what's really wrong with you?'"

P5.8 agreed, responding, *"I feel your pain on that one! I've been told that by too many drs, nurses, and family members....its just stupid. Rare means not common, not impossible!"* Participants sought to combat this disinterest or lack of understanding with education; they were very active in seeking out and distributing information about their condition to their friends and family members. P5.8 participant described her actions:

> "I have TRIED to send email links about [disease name], etc., I have TRIED to send letters from DOCTORS stating that [disease name] is HEREDITARY and the risks and how they should be tested, I HAVE TRIED to personally educate, I have TRIED to take people to dinners and Dr appointments, NONE OF IT HAS HELPED. They simply do NOT UNDERSTAND and think that at sometime I should just 'get better' and feel good enough to be 'normal' again."

Some participants distributed detailed articles, like P5.9 who sent *"medical journal articles to my relatives to read that talk about the disease in some detail."* Others looked for materials that were easier to understand, like P5.2 who looks for *"easy info for my family to read."* The amount of detail in the article did not appear to make a difference and participants had a number of different theories on why this was. P5.2 expressed, *"Not sure if they don't want to face it, or if they are scared, which leads to them not wanting to believe me."* P5.9 explained that her husband did believe she was ill, but that he still did not want to deal with it because *"it's pretty awful to watch someone go through these attacks every week."* P5.10 agreed, *"'Normal' people don't really want to hear about sad things and from people who have a health problem because it makes them feel 'down' or guilty that they are well."* P5.3 similarly theorized that her family members found it hard to face these health concerns, hypothesizing that illness is too threatening for many people. She explained:

> "Talking about it is something we have to do all the time, but people who have relatively normal health histories, don't want to be reminded of their own potential mortality and so they don't talk about it and we end up feeling MORE isolated. I wish my family would ask me WHAT it is, really is, that had me in the hospital for the 5th time this year, I wish they'd realize that I wasn't ill so much to get attention (because this is NOT the kind of attention I'd want)."

Participants often attributed a lack of support to a lack of understanding, feeling that people genuinely *wanted* to be helpful but sometimes made comments that may have been intended as caring but came across as minimizing.

They described how family and friends would make suggestions like, *"Let go and let God."* (P5.12) or *"try eating healthier, acupuncture etc"* (P5.5). Participants found these comments to be minimizing, and P5.5 lamented, *"'Friends' and I air quote because they are proving themselves to be less and less, downsize it and say things like they never heard of it and it can't be thaatttt badd. Omg if I hear that one more time!"* Comparisons to other diseases like heart disease (e.g., *"Well at least you don't have something serious like heart disease."* (P5.7)) or cancer (e.g., *"You ought to be grateful you don't have a real disease like cancer"* (P5.9)) seemed especially common. Participants expressed frustration with these comparisons, even going so far as wishing they *did* have cancer, making comments like, *"actually I think I would have a better chance of survival with some forms of cancer"* (P5.9) or *"If I shaved my head I would bet they wouldn't think twice about asking me"* (P5.12). P5.3 explained:

> "It's minimizing Those kind of comments, especially the first, 'at least you do not have something serious, like cancer.' [Cancer], not minimizing it at all, is not the only financially, emotionally, physically, mentally devastating disease. But it is the only [one that] many have heard of, It has a face."

Tangible Support

Participants also wished friends and family members would be more understanding of their physical limitations (e.g., trouble gripping objects or moving around) or other sorts of tangible support needs (e.g., needing a place to live or financial support).

When we asked participants what kinds of questions they wished their family and friends would ask, many responses described practical, instrumental support.

Examples of these tangible support requests included:

- *"Is there anything you really want to eat, drink, read, etc."* (P5.2)
- *"What kind of routine household chores do you need help with?"* (P5.3)

- *"Does this effect you on a daily/hourly basis? Are there any projects you could use help on? Could I pick anything up for you at the store?"* (P5.8)
- *"What kind of limitations does XXX cause you? What can I do to help you with those limitations?"* (P5.13)

This practical support was needed but not generally present and was not independent from the nurtant support challenges they faced. P5.8 explained, *"I would take some more help with tasks, which if they BELIEVED me, they would know I need."* Participants provided anecdotes like their parents kicking them out of the house or a relative refusing to come inside when picking up his daughter from the participant's house. For P5.10, the main barrier to this support was that *"those who care live on the other side of the country or the other side of the city. The family members just around the corner don't give a damn."* P5.9 overcame some of these obstacles, *"because I have a paid caregiver who comes to my house twice a week to do laundry, housekeeping, and preparing food, I don't really need family members to help with those tasks. I have all of my groceries shipped to my home from soap.com or Amazon.com, and my meat, poultry, fish come frozen from Omaha steaks.com."*

This was not the case for most participants however, largely because of the financial burden. Instead, they had to adjust their expectations based on what they could and could not get done in a given day. P5.8 said, *"I prioritize what I MUST get done today over what I'd like to get done, because I might not have the energy for both....or sometimes either."* Definitions of "busy" or "getting a lot done" had to change. P5.2 considered a "busy day" to be one in which he *"was able to shower and go to the grocery store with my mom."*

Informational Support

Participants did not discuss informational support as being something they expected or wanted from friends and family members. This type of support is something they generally expected more of from other sources (medical professionals, "Dr. Google", or other patients). Instead, they often worked to *provide* information to their friends and family members as part of their efforts to obtain better nurturant support.

5.2.3 Relationships Within Rare World

Support groups for people with rare diseases successfully empower people with rare diseases to connect with each other [5, 79, 270, 295, 326, 340]. In cases of other kinds of chronic conditions, research has shown that supporting each other may alleviate some of the need for outside help and even act as a form of self-therapy [196].

Relationships Among People with the Same Rare Disease

We observed that if there is support needed that is not currently being provided by friends and family members, people with rare diseases turn to each other for support. Participants reported that this was the case for the Facebook groups they were a part of. As P5.10 explained, *"I've been able to count on one hand those who've been there for me. In the global groups I belong to the support has been huge. I really value that."* P5.9 similarly expressed, *"And without loved ones, the only place we have to turn it is to the Internet and our disease specific group pages. That's the only place I get my support."*

Most participants were either retired, unemployed, or receiving disability payments. Some even considered managing their disease as being their job. P5.7 explained, *"I am on disability so I spend my time learning and keeping up on research for my disease so I can keep my doc informed. Sad isn't it? lol."* They discussed how they contributed to their other groups and communities by sharing research they found. P5.7 explained that she shared research with the Facebook support group for her condition, stating, *"quite alot of us do as our docs aren't enformed enough and someone has to be! lol, new things are coming out yearly."* This practice of finding and sharing research was common in quite a few different groups; some participants were even responsible for having started these groups. P5.9 had started *"a handful of Facebook groups and pages"* to share and collect research articles. She explained:

> "sometimes it's easier to access them when stored that way, instead of on my computer [...] And I want people who are interested in knowing more about the research to be able to access the current literature if they want to. [...] I subscribe to Google scholar for research on [disease name] as soon as it's published. I don't always understand the biochem information, but I share the research paper with Facebook [disease] group and there is usually someone who can put the information into understandable English for me LOL."

She notes that she is not the only one in her groups that shares articles like this; several other people with her same condition also *"keep up on the research and post articles as well."* These groups were also useful in trading tips or experiences. P5.9 uses her groups to access:

> "information you can't yet from your doctor, because he doesn't really deal with day today [disease experience] So he doesn't know. And I get decades of experience from folks who have been diagnosed much longer than I have. We have a handful of drugs that are ultraexpensive that treat our condition but of course there is no cure. And we share about which medications seem to be working better for more people, or ones that seem to be causing problems in the ones who have tried it."

Overall, these disease-specific support groups on Facebook were highly valued, such as for P5.11 who commented, *"I wish I had had the internet when I was 17."*

> People described these groups as providing not only better quality support (*"the online support groups are where I find the best support"* (P5.7)) but also more reliable support (*"And when someone is in and emotional/medical crisis, they are there for us. Even while we are in the ER, a hospital bed, or in ICU. My family wasn't there."* (P5.9)).

However, some participants noted that despite the supportiveness and effectiveness of these groups, they still longed for more supportive in-person relationships (whether that be with their friends/family or with other people with rare diseases). P5.8 explained:

> "But I also really miss being able to get out, even though I can't talk like this to my family. I would love to talk to ACTUAL people face-to-face that understand this condition, or at least chronic illness, but considering i have NEVER been able to travel much or get out easily, and am now homebound, that may be difficult. So, i get a little frustrated."

Relationships Among People with *Different* Rare Diseases

For this study, we created a group for participants with *different* rare diseases to come together. We found that they were very interested in learning from one another and were curious about each others conditions. Although there was a certain amount of clustering by disease, participants expressed empathy towards each other and were appreciative of the support they received. Despite

the difference in symptoms, the shared experience of living in "Rare World" provided much common ground. One participant[2] said, *"I am glad that I got to be a part of this experience. I learned a lot about other people and their conditions, and that we are ALL basically fighting the same battles..."* They started to identify things they had in common with each other, and even discussed these commonalities within the group. P5.12 observed:

> "I've noticed a commonality between the span of rare diseases.
>
> 1. The cost is astronomical.
> 2. The lack of understanding from the medical community.
> 3. The lack of compassion from society (not everything should be compared to cancer, we each have our own rare disease).
> 4. Lack of support from families.
> 5. Social isolation.
>
> Anyone can add more but this is my observation from the last few months."

Even within our study group, we observed participants engaging in discussions with each other outside of the formal study activities. Often, these discussions involved a participant describing some experience or challenge they were going through while other participants offered empathy or emotional support. Participants regularly posted updates on their life, such as recent or upcoming medical visits. Sometimes these were topics not directly related to their medical condition, but around notable things going on they wanted to share. They regularly exchanged resources or information with one another. One participant shared a link to their blog and other participants were curious about how to start one, so they began exchanging instructions. Similarly, a participant experiencing financial stress received suggestions about applying for disability insurance and where to find an appropriate lawyer. An international participant asked about health insurance while travelling in the US.

[Participants expressed an appreciation for the support they received from each other, even though their diseases were quite different.]

They shared personal stories with each other, offered support during rough times, and celebrated each others' successes. As an example, P5.3 shared with the group:

[2] Quote was provided anonymously as part of the debrief survey.

> "PUT ON YOUR PARTY HATS AND GET OUT THE NOISEMAKERS, MY HUSBAND GOT A JOB!! Let me tell you, after years of disappointments and tears and fears, learning how to feel again is different. I'm numb [...] The thing is, that most of you didn't know that you supported us; BUT YOU DID! Thank you all."

Participants even expressed that that this diversity of diseases gave them a different perspective on their own experience, such as P5.10 who expressed, *"I came to the conclusion from being in this group that most of you are far worse off than people with [my condition]."*

5.3 Discussion

The results of the studies described thus far in this dissertation indicated that rare diseases impact support relationships in many ways. First, participants described finding it hard to form new relationships, facing challenges in disclosing their conditions and communicating expectations. Second, participants' existing relationships struggled to adjust to changes in the dynamics of the relationships following a diagnosis – friends and family members reacted in many different ways to this change in circumstances. Third, participants expressed that the social support they received from outside of Rare World was limited in both quality and quantity. They received better and more frequent support from communities of similar others, although this support was still limited to that which could be delivered online.

5.3.1 Overcoming Barriers to Social Support

Existing literature suggests people are reluctant to ask for help when they need it [57, 75, 84, 113, 190, 353]. In this dissertation, we learned that participants often felt burdensome or guilty about the toll their condition was taking on those around them; this might suggest they also face barriers to reaching out explicitly for help. We also learned that people with rare diseases identify very strongly with their conditions, and existing research suggests that people who identify strongly with a single part of who they are often see negative impacts on their other relationships [117]. We heard stories of primary caregivers actively being in denial about changes in their relationship following a diagnosis as well as family and friends not believing participants. Some family members tried to be helpful but did not know how and often missed the mark, echoing research from psychology that people often have misconceptions about the best ways of being supportive [78]. No matter what the

cause, it is clear that there is a need for social support here that is not being met presently.

One possible solution might be to address care network fatigue by expanding the care network to reduce the burden on any given caregiver by distributing the workload across a range of helpers [154]. The challenge here is that even when people outside of Rare World *do* want to be helpful, they do not always understand *how* to be helpful; their attempts at being helpful sometimes have a negative impact (e.g., P5.7's example of *"Well at least you don't have something serious like heart disease."* or P5.9's example of *"You ought to be grateful you don't have a real disease like cancer"*). This may be something that could be improved through awareness and education, such as in Liu et al.'s [196] study of video blogging to educate viewers about common chronic illnesses; this strategy could also help people with rare diseases communicate their experiences to others, especially given their interest in engaging with social technologies to share health information (see Chapter 4). This would help to not only gradually bring more trusted people into the care network and reduce care network fatigue, but also help them be better helpers.

Technology can facilitate the sharing and navigation of this information, but we should be mindful that too much information can be counterproductive; family members of participants in this research were often rapidly forced into caregiving roles they had not anticipated and this new diagnosis can cause a quick shift in the dynamics of an existing relationship. P3.6 told us her husband reacted to the role change by going into "complete denial." P5.2 similarly expressed frustration about his mother's denial of the severity of his disability. These stories of a primary caregiver in denial, when considered in light of the higher divorce rates for couples where one person has a chronic illness [125, 312], 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. We could consider designing for a slow discovery process (similar to Raban et al.'s [277] concept of "Progressive Identity Disclosure") 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 research draws attention to carefully balancing a potential friend's informational needs with their readiness to accept that information (and, of course, against the person with a rare disease's right to dignity and privacy).

5.3.2 A Helping Network to Facilitate Peer Support in Rare World

There are any number of different approaches to potentially address some of these challenges, including expanding the care network to reduce the burden on any single caregiver, raising awareness to reduce stigma and misunderstanding around rare diseases, or connecting people with rare diseases to relevant existing local resources for managing health-related disabilities (e.g., nonprofit groups, local faith communities, or other formal social services). In this dissertation, I explore one of these possibilities: facilitating support exchanges among people with different rare diseases.

Researchers who work with older adults [8, 313] have suggested that peer support allows people to tap into much richer, more diverse ecosystems for support and reduce the burden on a traditional single informal caregiver (see Chapter 2).

> Adopting a similar mindset of empowering people with rare diseases to support *each other* may reduce some of the burden and reliance on friends and family members who, despite the best of intentions, may not always provide adequate support. Helping each other may not only address gaps in the support needs of people with rare diseases, but could also serve as a useful distraction from the challenges of living with a rare disease, or self-reported (by P5.9) "dwelling" on one's illness.

Being empathetic and helping someone else with a rare disease would allow people to continue to live in Rare World without as much dwelling or negativity. As was the case in our study, this might also provide some perspective on what others may be experiencing; individuals derive a number of mental and physical health benefits from helping others [168, 230, 256]. Peer support may be a means of empowerment, allowing people with rare diseases to receive much needed support in a way that actually reduces their current isolation by connecting them with people with different diseases and broadening their perspective.

The findings of this study suggested that *tangible* support is particularly lacking for people with rare diseases. Participants reported receiving emotional and informational support from members of their support communities. Tangible support, however, often requires collocation (e.g., helping out around the house). The low prevalence of specific rare diseases makes in-person tangible support

currently challenging to coordinate (i.e., if there is no one with the same condition in an individual's geographic area). Additionally, some of the tasks participants described needing help with are things that it would be challenging for someone with the same condition to provide; if a person requires help with groceries because their condition makes lifting the grocery bags challenging, it is likely that someone with the same disease might have similar support needs, rather than being able to help out.

> We may be able to overcome these barriers (geographic dispersal and symptom-based limitations) by facilitating connections between people with *different* rare diseases.

Despite differences in diseases and symptoms, this dissertation has shown that there are many *experiences* in common between people with different rare diseases. I hypothesize that these common social experiences of "rarity" may serve as common ground, while coordinating support among *different* rare diseases may help overcome the symptomatic and geographic limitations of trying to coordinate tangible support between people with the same condition. There may also be opportunities to connect with people with common chronic illnesses who may have sufficient common ground while also benefiting from and contributing to an exchange of support; we investigate this possibility further in Chapter 6.

Although it might be possible to facilitate one-on-one exchanges of tangible support between people with rare diseases, I suggest this would be more effectively achieved using a timebanking metaphor [63, 143, 184, 299]. Bellotti et al. [21] define a timebank as:

> "[an] organization in which members earn 'time dollars' for providing any kind of service (from apple picking to zoology lessons) to others. Time dollars are essentially 'minted' when a service recipient is debited by the same amount of time dollars that the provider is credited with."

A person with a rare disease would gain time dollars for help they provide to others that they could then redeem for help with their own needs. Research on motivations for participating in timebanks reinforce theories introduced in Chapter 2; people are keen to contribute altruistically and enjoy helping others, but they often struggle to ask for help. While timebanks might minimize some of the stigma of asking for help, it is still not a perfect solution. Bellotti et al. [21] describe

how the banking system *"can be seen as failing if people feel uncomfortable about asking for help, as if they are still obligated somehow, in spite of 'paying' with their hours."* One participant in Bellotti et al.'s [21] study explained that,

> "Offers drive timebank usage. Offers are the driver, not request [. . .] We have a lot of barriers against requesting. It's really easy to scan through a set of offers and go, 'I could use that.' The emotional demand is created when you see the offer, not when you see the request."

One way of overcoming the problem with people not asking for help might be to more explicitly frame and design a system around offers, rather than requests. Bellotti et al. [21] propose reconceptualizing timebanks as "Helping Networks", describing:

> "a social network in which you do not simply connect to people, but offer to do something for them or let them do something for you. You acquire contacts through service exchanges with different people and the more you do this, the bigger your network becomes."

[A "Helping Network" designed around offering help may help reduce the barriers to seeking help.]

Example Scenarios

Here we provide three example scenarios of how we might facilitate this type of cross-disease support. We distinguish between abilities, skills, and expertise as follows: *abilities* are tied to physiological capabilities or symptomatic limitations (e.g., being able to see, hear, walk, lift something); *skills* are acquired abilities, learned through experience or through study (e.g., cooking, programming, knowing a different language); *expertise* is acquired through formal education, certification, or regulated professional experience (e.g., practicing law, real estate, skilled trade). In all scenarios, we will refer to the '"support provider" and the "support recipient" for clarity, but we note that people with rare diseases would shift between these two roles at various times, earning and spending time dollars as they go. It is also possible a "support provider" in a given scenario could be a friend or family member, in a case where there are friends or family members that want to be supportive and helpful, especially if these tasks are scaffolded to help people learn to be good helpers. This would

also be useful in a case where the friend or family member does not live near the person with the rare disease, but still wants to be helpful. The support provider could bank time for their remote friend or family member while helping someone locally.

Scenario 1: Coordinating Support by Abilities Several participants had conditions with severe physical symptoms. For example, the primary symptom of Inclusion Body Myositis is progressive muscle weakening. For other conditions, the physical limitations might not be persistent, but rather temporary debilitating flareups, such as in the swelling episodes associated with Hereditary Angioedema. One task participants specifically mentioned as an example was carrying groceries. Although there are an increasing number of grocery delivery services available, these do not take care of the tasks of gripping the bags, carrying the heavy items into the house, and lifting them into the fridge or cupboards. We imagine that if a support provider in the neighbourhood was going grocery shopping, they could take the support recipient with them, helping them bring the groceries inside at the end of the trip. Alternatively, if the support recipient is not up for the trip to the store, they could give the support provider a shopping list to be filled independently.

Scenario 2: Coordinating Support by Skills One conversation that took place in our study was around how to start a blog. This was something that one participant mentioned doing as a way of sharing their experiences and educating the public about their condition. Other participants in the study found this interesting and considered starting their own, but had questions about how to do so. We imagine that a support provider who is comfortable with technology, familiar with blogging platforms, or maybe skilled with web programming could help a support recipient who is less skilled in this way to put together and/or maintain their own blog. Notably, this scenario relies on skills that might be picked up formally (e.g., through some computing education program), but could also be self-taught (e.g., having taught oneself to put together a personal blog). This example also does not require geographic proximity. To facilitate this kind of support, it would be useful to know details about the skill set of the support provider.

Scenario 3: Coordinating Support by Expertise Another conversation that took place in our study group was about the disability payment claim process. One participant was experiencing financial hardship, and another participant described their experience through the process and

provided advice on how to find a lawyer to help. We propose extending this informational support scenario even further to the tangible next step: What if the support provider *was* a disability lawyer? While it may initially seem like an infrequent combination to find a disability lawyer who also has a rare disease, research suggests that people often choose a career path to which they have some personal connection, especially when it comes to health topics [31, 72]. As a second example, it is possible that someone with a strong background in certain sciences or medicine could help interpret the medical jargon often used in research papers that are of interest to a support recipient.

5.4 Summary

In this study, we saw that people with rare diseases often do not feel they are getting the support they need from their friends and family members. There may be opportunities for designers and HCI researchers to support the relationships between people with rare diseases and their friends and family, but further study of these challenges from the perspective of these friends and family members would be necessary to properly understand the barriers. Instead, in this dissertation I suggest there are opportunities for people with rare diseases to support each other, instead of relying as much on people outside of Rare World. While existing rare disease support groups excel at providing informational, emotional, network, and esteem support, there are barriers to facilitating tangible support within these communities. I propose an approach to overcoming barriers using social matching to facilitate support among people with *different* rare diseases. In the next chapter, I investigate the specific mechanisms for matching people to each other for the purposes of support.

> *Byrdes of on kynde and color flok and flye allwayes together.*
>
> —William Turner in *The Rescuing of Romish Fox*

Chapter 6

Facilitating Peer Support Among People with Different Rare Diseases

People with rare diseases face a number of challenges relating to social support and relationships. Participants in our research found it challenging to form new relationships and existing relationships struggled to adjust to a new diagnosis. Friends and family sometimes made well-meaning but hurtful comments. Instead, participants described receiving more frequent and better quality support from other people with rare diseases in the disease-specific Facebook groups, especially for nurturant and informational support. These groups provide people with a sense of community and belonging they do not feel they are getting elsewhere; people who also had a rare disease were more familiar with their experiences and better able to empathize.

[Although existing rare disease support groups excel at providing many forms of support, there are barriers to facilitating *tangible* support within these communities.]

An interesting new opportunity is to consider how we might extend these existing online rare disease groups to support more *tangible* forms of support. In this chapter, I investigate the possibility of an offer-driven helping network for people with different rare diseases to facilitate exchanges of social, especially tangible, support. I additionally include people with common chronic illnesses in this study to begin exploring the possibility of extending such a network to broader chronic illness populations. There are three important questions we must answer to be able to successfully design such a system:

1. What kind of support do people need? What support can people best provide?
2. Which characteristics do we need to know about two people for them to be interested in connecting with each other for social support?
3. How comfortable are people sharing these characteristics on social platforms?

I begin by summarizing relevant existing research on the mechanisms by which people are drawn to each other and the characteristics that are important when connecting two people, especially in online contexts. I then describe a two-part survey conducted to identify the tasks that may be suitable for a tangible support "helping network" [21, 64], the most important characteristics for matching people with different conditions, and people's information sharing preferences. In part one, I ask people to self-report dimensions on which they prefer to be similar to people in their helping network and about their comfort disclosing information to different network members. In part two, I evaluate our ability to curate profiles of potential helpers. I conclude with a discussion of the role of unconscious bias in how people select each other, the potential to leverage existing social media features for such a system, and the potential privacy implications of embedding such a system into a larger social platform.

This chapter contains material from our CHI 2017 paper [203] (coauthored with Grace Bastin, Leslie S. Liu, Katie Siek, and Kay Connelly). It additionally contains material from two unpublished manuscripts, describing research conducted with Julia Mayer, Alan Clark, Bin Xu, Juan F. Maestre, Katie Siek, and Kay Connelly.

6.1 Related Work

To facilitate introductions between people for the sake of exchanging support, it is necessary to understand the nuances of connecting people to each other. Terveen et al. [320] explain:

> "Recommending people [to each other] fundamentally changes the game. Some amount of personal information about users is necessarily revealed. This makes for an inherently riskier interaction, and thus elevates issues such as privacy, trust, reputation, and interpersonal attraction to much greater importance."

Here, we rely on a particular class of recommender system called "social matching systems" or "social recommender systems." Rather than recommending products like books or movies to people, social matching systems recommend people to people [320]. Unlike other kinds of recommender

systems, people recommender systems need to consider the complexities of social interactions and attraction [320]. Social matching systems are largely based on theories of interpersonal attraction, such as similarity-attraction theory which suggests that people are actively drawn to people like them (*"likeness begets liking"*) [43, 226]. Participants in this dissertation reported gaining significant value from connecting with similar others (people who have the same disease, who have navigated similar challenges, who have been through similar experiences).

Similarity-attraction is known to exist based on status characteristics (age, gender, race, ethnicity, education, religion) [188], value characteristics (thinking in similar ways) [16, 188], as well as based on hobbies [80], preferred activities [345], shared experiences [269], and social habits [95]. Evidence of this tendency of individuals to associate and bond with similar others (called "homophily") has been repeatedly shown to exist in online social networks [23, 24, 53, 321]. Researchers have considered a number of different similarities for facilitating matches between people, including not just similar demographic characteristics [62, 148], but also virtual or physical places visited [137, 233, 319], interests [137, 148, 233], and even language style [148]. Table 6.1 summarizes different characteristics that have been shown in previous work to be useful in connecting people online or are strong sources of homophily.

[Similarity also influences willingness to help one another [65]; when connecting people for social support, similarity is an important factor.]

An alternative to similarity-attraction is "complementarity attraction," which suggests people are drawn to others who complement them, or fill certain gaps ("opposites attract") [92, 216, 252, 356]. Complementarity-matching is most frequently observed in matching systems for job seeking [213], expertise finding [283, 363], or mentor-mentee matching [42] as well as in sharing economy services (e.g., Lyft, Uber, AirBnB) [87, 88, 182]

I leverage this tradeoff between similarity-attraction and complementarity-attraction to match people with rare diseases for tangible support. I hypothesize that having a rare disease would be a strong mutuality for connecting two people (similarity-attraction), while having *different* rare diseases would overcome symptomatic and geographic limitations that might occur among people with the same disease (complementarity-attraction).

Personal Characteristics. These strongly predict interpersonal attraction, but are generally subjective, individually variable, culturally variable, invisible, and challenging to operationalize for social matching technologies [16, 95, 188, 269, 320, 341]. Examples might include:

- Beliefs & Values
- Shared Experiences
- Personality
- Character
- Trustworthiness
- Sense of Humor
- Attractiveness
- Knowledge
- Lifestyle

Proxy Variables. These are items that are easily visible, objective, or categorical surrogates for personal characteristics [112, 188, 320]. These are very commonly used in making judgments about people when other information is unavailable and social matching systems rely heavily on them because they are easily operationalized [317]. An over reliance on these proxies could lead to false assumptions or reinforced biases [30, 343]. Examples include:

Demographics [62, 148]
- Age
- Gender
- Sexual Orientation
- Race or Ethnicity
- Language

Affiliations
- Religion
- Political Party
- Education
- Profession

Lifestyle [110, 111, 201, 282]
- Drinking Habits
- Smoking Habits
- Relationship Status
- Family Structure
- Income

Appearance [110, 111, 201, 282]
- Height
- Weight
- Build
- Eye Color
- Hairstyle

Mutualities & Complementarities for Assessing Potential Matches. These are profile items or online behaviors that are less obviously visible than proxy variables and generally aim to operationalize invisible personal characteristics, providing more dimensions on which to match people or more variables on which to assess a match [80, 345]. These may vary depending on the use case. Examples might include:

Interest-Based Matching [80,94,137,148,220,233,319]
- Hometown
- Places Visited
- Media (Movies, books)
- Venues (restaurants, cafés, bars)
- Sports (playing, cheering for)
- Interest in an upcoming event
- Group membership
- Content of posts

Expertise-Based Matching [56, 137, 148, 169, 174, 225]
- Skills
- Abilities
- Expertise
- Publications
- Certifications
- Years of Experience
- Posts
- Tags

Health-Based Matching [62, 148, 203]
- Group membership
- Symptoms
- Diagnosis
- Insurance
- Medications
- Medical tests
- Dietary restrictions
- Medical facilities

Mutualities for Connecting with Potential Matches. These are items people use to assess how likely it is they could or should actually connect with a person (as opposed to assessing their interest in a person). Examples include:

Filtering Criteria [363]
- Current Location
- Availability
- Busyness

Establishing Trust [90, 323]
- School or workplace
- Mutual friends
- Group membership

Facilitating Introductions [319]
- Mutual friends
- Upcoming events
- School or workplace

Match Moderators. The relevance of a particular mutuality in a given context is influenced by a number of moderators [222]. Examples might include:

- Rarity *[127, 220–222, 257]*
- Strength *[221]*
- Granularity *[222]*
- Familiarity *[180, 265, 342, 350]*

Table 6.1: Overview of different personal characteristics, proxy variables, and mutualities (for assessing and for connecting with potential matches) that affect how people interpret online profiles.

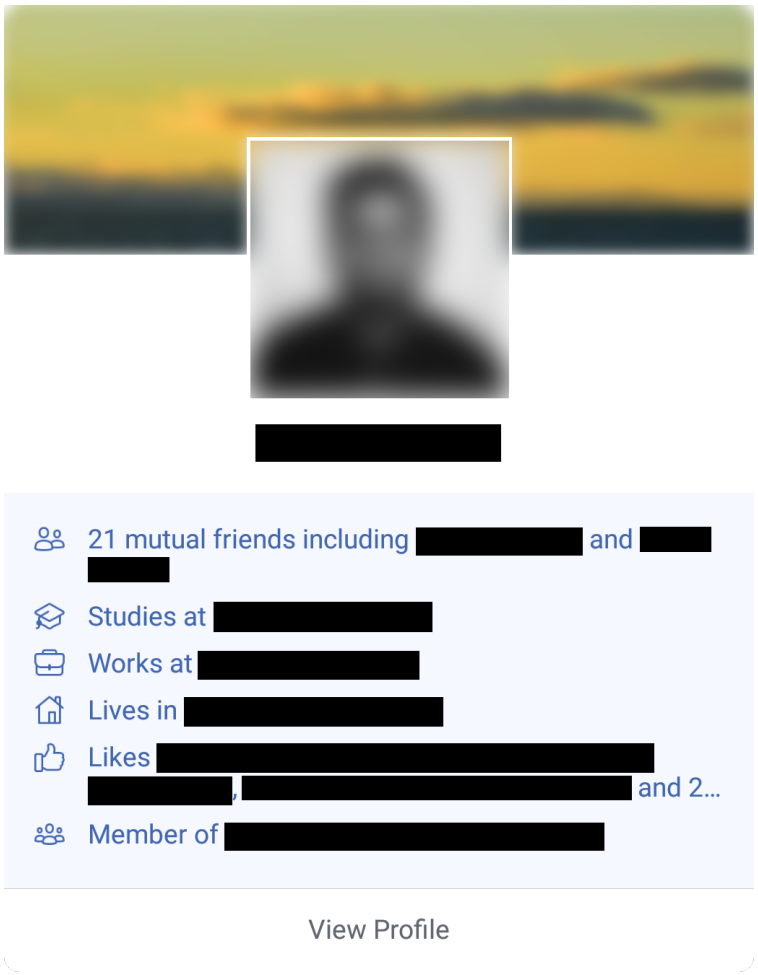


Figure 6.1: Facebook's Discovery People Feature. People can swipe through different profiles and see things they have in common with that person, including mutual friends, events attended, shared affiliations (work or school), hometowns, page likes, and other mutualities.

6.2 Method

To investigate the importance of different mutualities and complementarities in facilitating connections among people with rare diseases in a helping network, we rely on Facebook's "Discover People" feature, shown in Figure 6.1. We can think of Discover People as an example of a social matching system; it uses information from Facebook profiles to show other people based on the similarities between them. Discover People *"can help you discover more about people you have things in common with by browsing through profile cards of people in your community"* [271]. Discover People visibly calls out similarities between people via a box labelled "Things in Common" (shown in Figure 6.1). This aligns with research suggesting perceived similarity is known to be more impor-

tant to the success of a relationship than actual similarity [237] and that observable mutualities more strongly predict attraction [249]. Where existing HCI research has studied social matching systems that connect people to each other for a specific purpose or context (e.g., dating [200, 201], friendships [94, 220, 233, 319], or professional connections [137, 174, 225]), Discover People supports multiple, diverse use cases. It can be accessed through several places on Facebook; some of these entry points lead to categories of people that seem to imply a specific use case (e.g., *people who also work for my company*), while others lead to categories of people for which the use case is less clear (e.g., *my friend's friends*).

> We ground this study in a particular Facebook feature because people with rare diseases are already leveraging Facebook as a place to connect with other people with rare diseases. The "Things in Common" information highlighted as part of the Discover People feature provides a structure with which to present relevant mutualities to help people with rare diseases evaluate possible helpers. Additionally, the open ended nature of Discover People allows for easy extension into new use cases, such as social support.

Of the characteristics explored in previous research, it is important to consider which are most useful for matching people with rare diseases and the privacy implications of sharing this information. We distributed a two-part survey to people with rare diseases and people with common chronic illnesses to understand how they prioritize this information. This study was approved by IU's IRB (Appendix A - #1711196782A001).

6.2.1 Phase One: Survey of Tasks, Privacy, and Helper Preferences

Phase one consisted of a survey (Appendix D) of questions about demographic information, how people prioritize different information in selecting people from online profiles, and comfort levels in sharing information online. To be eligible for the study, potential participants had to be over 18 years old with a rare disease or common chronic illness. We additionally required that participants be Facebook users because many of the survey questions referred to Facebook as an example. We tested this survey internally to identify potential problems and estimate the time it would take to

complete. We then recruited participants from Facebook (including Instagram), Twitter, and Google AdWords using paid advertisements. Details of our recruitment methods are provided in Chapter 9. Participants who completed the survey had the option to be entered into a raffle for a $25 gift card.

The survey began with basic demographics (age, disease information, social network use, gender, country, and education). We then asked participants to *"imagine you are a part of a community of people who help each other out. Every time you help someone else, you earn points that you can trade in for someone else to do a favor for you."* Based on research suggesting people are keen to contribute altruistically and struggle to ask for help (see Chapter 2), we first asked participants what kinds of tasks they could help with, allowing them to select from a check box list. Next, we asked participants, *"You're a great helper and you've accumulated a lot of points. You can now trade in those points for someone to help you out! What kind of tasks would you like to trade in your points for?"* Participants selected from the same check box list, where the possible responses were tangible support topics (cleaning, cooking, computer tasks, driving, etc.) that emerged from our work in Chapter 5. The items were provided in random order. We also provided a "none of the above" option and allowed participants to write in additional suggestions of tasks.

In the next stage of the survey, we asked participants to *"Imagine you're choosing from a list of people willing to help you with one of the tasks you just picked."* We then asked them to choose their first, second, and third choice helper from the following items (presented in random order):

- Someone with similar demographics to me (same age, gender, race, sexual orientation)
- Someone with a similar lifestyle to me (relationship status, children, drinking or smoking, dietary preferences)
- Someone with similar interests (sports, movies, books, hobbies)
- Someone with expertise that complements mine (having special skills or talents)
- Someone with similar health experiences (same disease, symptoms, tests, medications)
- Someone with similar beliefs as me (same religion, political affiliation, etc.)

In the final stage of the survey, we asked participants which of that information (i.e., demographics, lifestyle, beliefs, interests, expertise, and health experiences) they would be comfortable sharing with the general public, with Facebook friends, and with a community of other people with chronic illnesses. They also had an option to indicate they would not be comfortable sharing any of that information.

We received 315 responses to phase one of our survey. Of these responses, 29.97% had at least one rare disease and 71.75% had at least one common chronic illness (11.53% of respondents

had both a rare disease and a common chronic illness. As in Chapter 4, these respondents are included in both categories.) An additional 9.80% of respondents described undiagnosed conditions or conditions where we could not verify whether the condition was rare or common based on the description provided; these responses are excluded from comparisons between common and rare disease respondents.

The plurality of respondents were young (20.46% of respondents were 18–25) and the majority identified as female (62.25% selected the pronouns "She/Her/Hers", 33.14% selected "He/Him/His", 4.03% selected "They/Them/Theirs", and 0.58% preferred something else). We received responses from participants in 35 different countries from all around the world, although we targeted our advertisements only at English speakers. Respondents had a range of education levels; 26.41% had no more than a high school education, 27.89% had completed a Bachelor's degree, 25.22% had completed a Masters or Doctoral degree.

6.2.2 Phase Two: Selecting a Helper

After completing phase one, respondents could opt-in to being contacted for a follow up study; 85.7% of participants agreed to be contacted and 69.2% indicated we could view their public Facebook profile. After a respondent had completed phase one, we waited two weeks (to minimize issues of recall) before sending them a personalized link to a follow up survey.

In this follow up survey, we asked them to select one person they would most like to receive help from from four example Facebook profiles. We used their responses to phase one of the survey as well as data from their public Facebook profiles to determine the content of these example profiles. Of the four profiles, one showed three "Things in Common" the respondent rated as most important in phase one and one showed three "Things in Common" the respondent rated as less important in phase one. One profile showed three random "Things in Common" (i.e., a mix of mutualities that were rated as important and ones rated as unimportant in phase one). We also included one profile with no mutualities (i.e., nothing in common).

We created these profiles manually for each respondent using image editing software. In preparing these profiles, we endeavoured to take various match moderators (described in Table 6.1) into account as much as possible to ensure that the example profiles were grounded in the participants' real lives and were as representative as possible of the kinds of people they might be interested in connecting

with. A limitation of this work is that the amount of information available on participants' public facing Facebook profiles varied and our use of match moderators was limited to the researchers' judgment (as opposed to participant's description of the importance of different page likes or affiliations). These example Facebook profiles contained different pieces of information based on participant data, but used fake names and stock photographs.

Participants who completed phase two received a $5 gift card for their time. We received 71 responses to phase two of our survey and these responses were almost evenly distributed between rare disease (45.07%) and common chronic illness (54.93%) respondents. Of these responses, 70.42% preferred the pronouns "She/Her/Hers", 23.94% preferred "He/Him/His", and 5.63% preferred "They/Them/Theirs".

6.3 Analysis & Results

Our analysis focuses on answering three key questions:

Q1: What kind of support do people need? What support can people best provide?

Q2: Which characteristics do we need to know about two people for them to be interested in connecting with each other for social support?

Q3: How comfortable are people sharing these characteristics on social platforms?

6.3.1 Giving and Receiving Support

To answer Q1, we considered two different independent variables, each with two levels: 1) whether we were asking about *providing or receiving* support and 2) whether the respondent had a *rare or common* disease.

First, we considered the support people with rare diseases were most interested in *providing* and most interested in *receiving* (Figure 6.2). We conducted chi-square tests for each support task and found that people with rare diseases were significantly more interested in providing help with computer tasks ($p < 0.01$, $\phi = 0.38$), cooking ($p < 0.01$, $\phi = 0.24$), medical advice ($p < 0.01$, $\phi = 0.21$), and laundry ($p < 0.01$, $\phi = 0.34$). They were significantly more interested in receiving help with cleaning ($p < 0.05$, $\phi = 0.18$), lifting heavy objects ($p < 0.01$, $\phi = 0.40$), legal advice ($p < 0.01$, $\phi = 0.34$), and financial advice ($p < 0.05$, $\phi = 0.17$). There were no significant differences

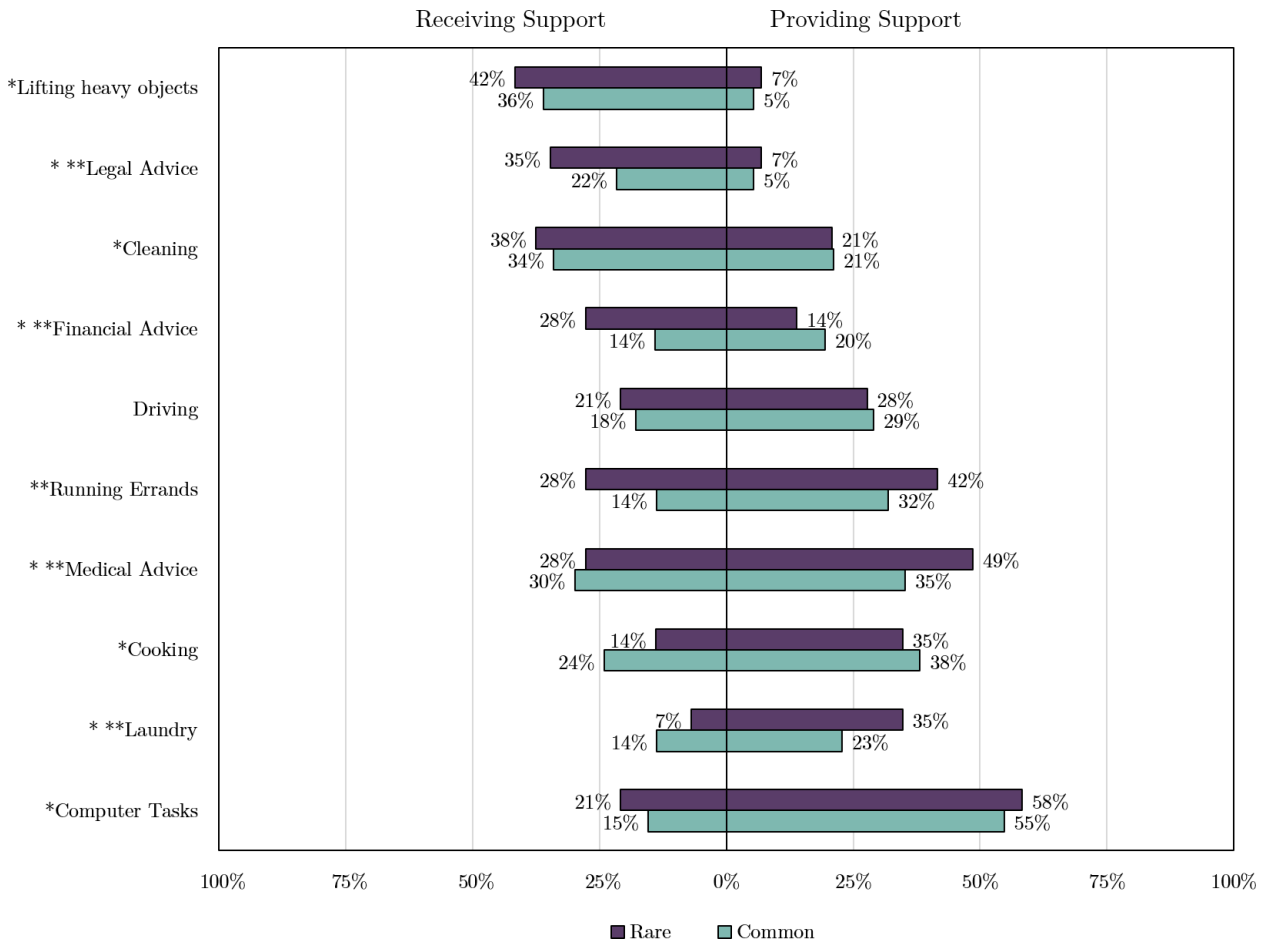


Figure 6.2: Tasks people would be willing to provide help with and tasks they would be interested in receiving help with for rare disease and common chronic illness populations. Single asterisk (*) indicates statistically significant differences between providing and receiving and double asterisk (**) indicates statistically significant differences between rare disease and common chronic illness.

for driving or running errands. This suggests that, with the exception of driving and running errands, there is an imbalance between the number of people with rare diseases offering to help with a given task and the number of people interested in receiving help with it.

> Helping networks for people with rare diseases will need to account for this imbalance in the supply of and demand for different support tasks, possibly by incorporating external sources of support or opening up the community beyond just people with rare diseases.

Next, we considered differences between rare disease and common chronic illness respondents. We conducted chi-square tests for each support task and found that people with rare diseases were

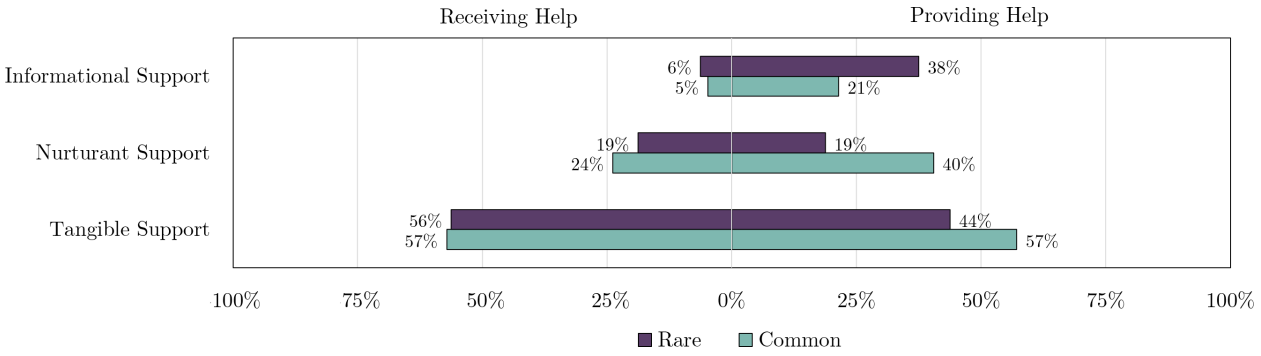


Figure 6.3: Results of coding of optional write-in responses.

significantly more interested in receiving legal advice ($p < 0.05$, $\phi = 0.13$), financial advice ($p < 0.01$, $\phi = 0.15$), and running errands ($p < 0.01$, $\phi = 0.16$) than people with common chronic illnesses. They were also significantly more willing to offer medical advice ($p < 0.05$, $\phi = 0.12$) and laundry ($p < 0.05$, $\phi = 0.11$) than people with common chronic illnesses. There were no tasks for which people with common chronic illnesses were significantly more likely to offer or request support.

Survey respondents also had an option to write in additional suggestions of tasks for which they would be interested in receiving or providing support, and 23.5% of respondents took advantage of this option. Two researchers coded these written responses as being either informational, nurturant, or tangible support (Figure 6.3) as defined by Cutrona and Suhr [76]. We assessed inter-rater reliability using Cohen's Kappa, scoring 0.80.

The majority of write-in responses described tangible support tasks, and people with rare diseases described wanting to *receive* tangible support slightly more often (56.25%) than they described wanting to *provide* tangible support (43.75%). People with common chronic illnesses valued both equally. Respondents described wanting to receive support in the form of tasks such as pet care, child care, gardening/lawn care, and massages/spa treatments.

For the most part, offers of support were quite different from support requests; respondents described being able to provide support with sewing, make up or hair styling, electrical work/welding, 3D printing/electronics, writing, translating different languages, carpentry, civil engineering, and other various skills. Some of these tasks seem broadly applicable and it is immediately obvious what their value to a helping network could be (e.g., hair styling). Other offers take more consideration to identify the value of the skill to a potential support recipient, especially if the support recipient does not possess this skill themselves. It might not be immediately clear to a participant in a

helping network what use cases exist for 3D printing, but people with this expertise may be able to imagine many different ideas (e.g., 3D printing prosthesis [153, 266] or applying civil engineering skills to accessibility initiatives [145, 146]). Helping networks might facilitate some of these projects by encouraging the person with the skill to provide suggestions or ideas of projects– these ideas could also be provided by past support recipients.

People with rare diseases were interested in providing informational support (37.50%) much more often than they were interested in receiving it (6.25%). Offers and requests of informational support typically addressed specific domains:

- *"Remind friends about appointments"*
- *"I also edit CVs and other documents, so could help people in this way"*
- *"locate doctors within Kenya"*
- *"I could assist with providing Workers' Compensation advice. (I was a professional WC Adviser before I had to stop working)"*
- *"study of medical books"*

People with rare diseases were equally interested in receiving and providing nurturant support (18.75% each). Offers and requests of emotional support included responses such as:

- *"Encouraging and being a friendly voice/face"*
- *"I can help them psychologically by giving them advice, listen to their problems and try to fix every thing together"*
- *"Keeping someone company"*
- *"Thought sharing"*
- *"Understand the situation like a 'normal' person"*
- *"help me to be confident"*

Finally, some responses described wanting to receive a lot of help (*"On a rough week it would be everything as I would be in bed"*) or receiving no help (*"Leave me alone"*).

6.3.2 Selecting Helpers

To answer Q2, we consider self-reported preferences for choosing a helper from phase one of this survey (*"Which of these people would be your first/second/third choice to help you out?"*) as well as each respondents' selection of a profile in phase two of the survey.

People with common chronic illnesses and rare diseases alike reported valuing helpers with complementary expertise; 86% of people with rare diseases and 65% of people with common chronic illnesses self-reported that *"Someone with expertise that complements mine (having special skills*

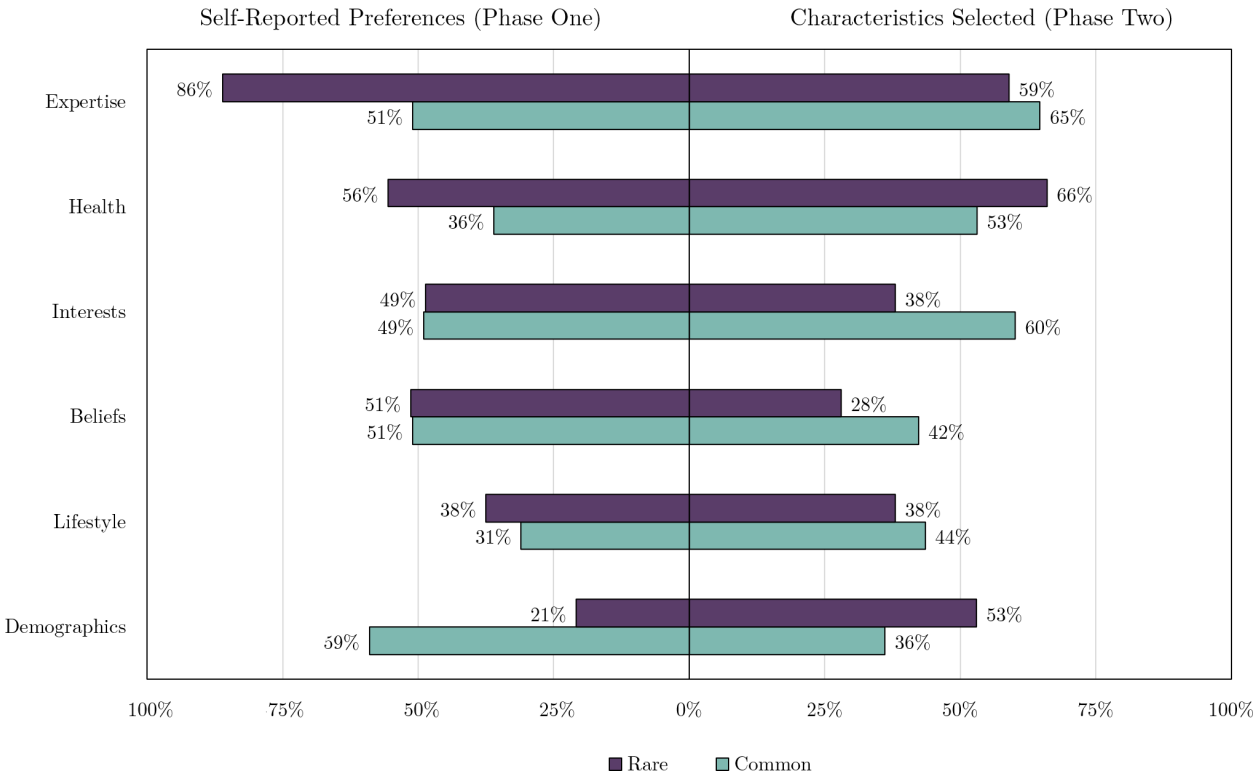


Figure 6.4: Preferences of characteristics of a helper.

or talents)" would be one of their top three choices of helpers (see Figure 6.4). Participants also frequently reported that having similar health experiences (56% rare diseases, 53% common chronic illnesses) or similar interests (38% rare diseases, 44% common chronic illnesses) would make a potential helper more appealing. In phase two of the survey, we provided participants with four profiles to choose from. One profile was someone with whom they shared mutualities or complementarities that they had self-reported (in phase one) as being important. The second profile was someone with whom they shared mutualities or complementarities they had reported as being less important. The third profile was someone with whom they shared three random mutualities, and the fourth was someone with whom they did not share any mutualities (i.e., where the "Things in Common" box was omitted). Few (7%) participants selected the profile of the person with whom they had nothing in common. Mostly participants selected either the person with whom they shared important mutualities (34%) and the person with whom they shared random mutualities (35%).

To understand these differences between what participants said they would do (in phase one) and what they actually did in (phase two), we considered the characteristics of the profile each participant selected. For example, if a respondent selected the random profile and this profile

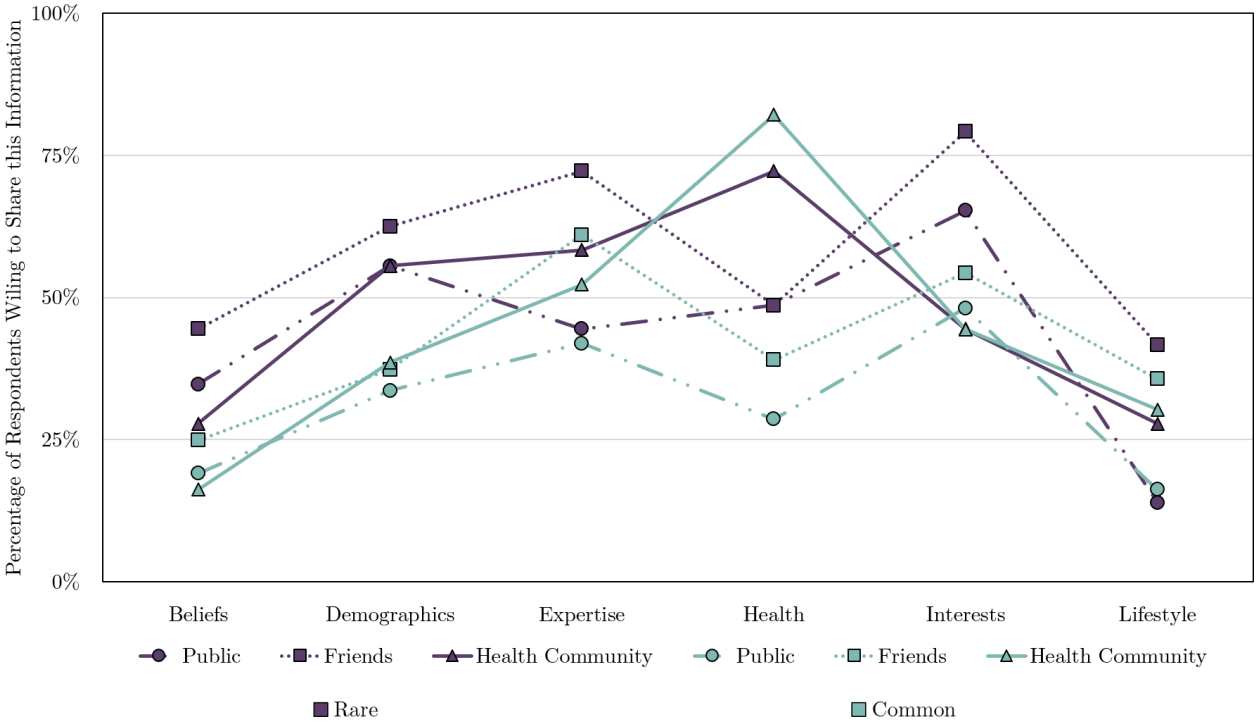


Figure 6.5: Willingness to share information with different audiences.

showed a person with similar beliefs, similar demographics, and complementary expertise then we recorded that participant selected those three characteristics (and did not select any of the the other three characteristics). Here we find that, as expected, participants are drawn to people with complementary expertise, although not to the same extent as predicted; where 86% of people with rare diseases claimed that expertise would be one of their top factors influencing their choice of helper, only 59% of them actually selected a helper with complementary expertise. We saw similar results for beliefs and for interests; more people with rare diseases reported valuing these characteristics than actually selected them in phase two. Characteristics people with rare diseases *were* drawn more towards than predicted were health (66% selected vs. 36% self-reported) and demographics (53% selected vs. 21% reported).

6.3.3 Information Disclosure

To answer Q3, we first considered people's willingness to share information with three different audiences: a public audience, their Facebook friends, or an online health community (Figure 6.5). We found that respondents were generally more willing to share information with their friends than

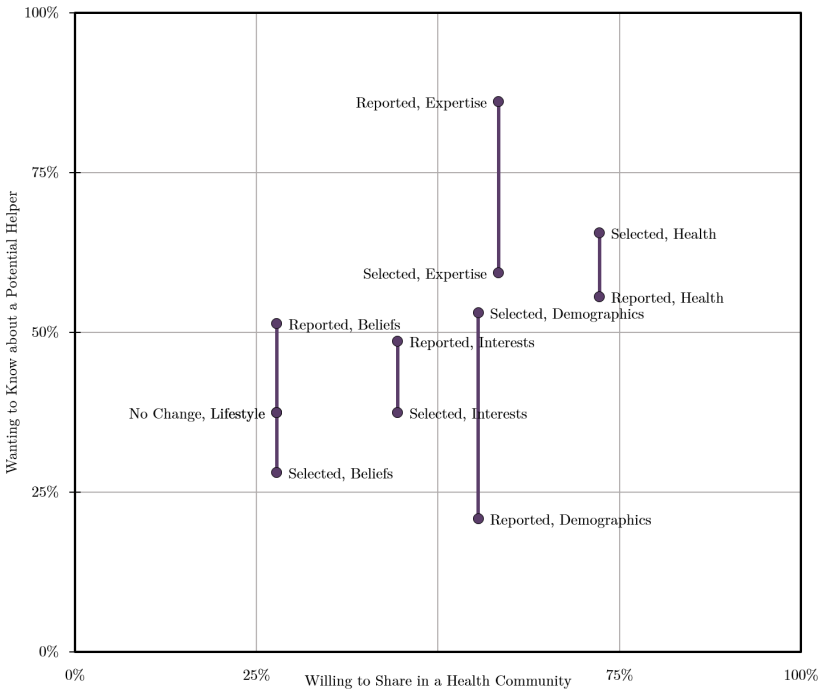


Figure 6.6: Alignment of willingness to share information and the desire to know that information about another person.

with other audiences. The one exception to this was health experiences, which people were more willing to share with members of a health community.

We found that people with rare diseases were more willing to share information than people with common chronic illnesses. An average of 44% of people with rare diseases were willing to share information with a public audience, compared to 31% of people with common chronic illnesses. This was also true for audiences of Facebook friends (58% rare diseases, 42% common chronic illnesses) and health communities (48% rare diseases, 44% common chronic illnesses). We found that certain information like beliefs (28%) and lifestyle (also 28%) were considered more private and people were less willing to share this information, regardless of the audience or disease type. Participants were most willing to share information about interests (56%) and expertise (55%).

We also considered how these information sharing preferences compared to information seeking preferences (Figure 6.6). That is, how does one's willingness to share information compare to one's desire to see this information about someone else? We found that profile items like expertise and health experiences were both desired information and acceptable to share within a health community, even with the differences between self-reported information and actual selection. Thus, these may

be useful items to emphasize in the design of profiles for a helping network. Items such as beliefs and lifestyle were less comfortable to share and less important to see, indicating they may be a lower priority for profile design.

6.4 Discussion

Findings from this study suggest that while there are many support tasks of interest to people with rare diseases, there is an imperfect balance between the supply of providers and the demand from recipients. We have begun to identify areas where people do not behave rationally (i.e., as they say they will) when choosing a helper. In this section, we draw from theories of sociology and social psychology to explain these differences and propose areas of further study. We also uncovered differences in people's willingness to share information on different topics with different audiences. In this section, we discuss the implications of these differences for the design of health communities embedded within larger social platforms. We review existing features of social media platforms that could facilitate support exchanges and reflect on the privacy implications of exchanging support on mainstream social media.

6.4.1 Computer-Mediated Helper Selection

People are interested in understanding experiential similarities and subjective personal characteristics about a person from their online profile, but this can be challenging to assess from the information available. Similarities that *are* better defined or more easily visible (such as gender, race, or age) are useful in matching people, but they may merely be proxies for the other more experiential similarities that people are actually looking for. In this study, we saw that people rely more heavily on demographics in making decisions about helper than they reported that they would, and less on items like beliefs or lifestyle that might be better signals of personal characteristics. Future work might investigate the relevance of these findings to healthy populations.

One explanation is that there is a substantial amount of unconscious bias at work, especially on social media profiles that emphasize photos in their design (including Facebook's Discover People). It may be the case that people are first drawn to profile photos prior to examining the "Things in Common" box; by the time they realize they share mutualities with a person, they may have

already formed an opinion. If the only information we have about a person is what they look like (when we are meeting someone for the first time or see their picture online), then proxy variables are the quickest things people can use to make these snap judgments. This is problematic because it can quickly lead to stereotypes or false assumptions about the person if we have pre-existing biases about certain groups or people or if we are lumping people into a group that does not describe them well [30]. In the case of health communities, an over-reliance on proxy variables can inhibit people's access to support. For example, Wang et al. [343] found:

> "Although personal traits were evident classifiers, the experiences that participants could relate to, empathize, and were knowledgeable of were the underlying similarities that enabled effective exchange of social support. For instance, P1 initially joined a group with similar height and had to shift to another one defined by age (40s), in which she could receive better support."

Interestingly, research has shown how easy it is to manipulate these judgments [25] and providing people with a common group identity can be useful for overcoming initial assumptions [122]. Nier et al. [251] found that if we put a baseball hat of a person's favorite sports team on another person, people previously judged as out-group now are judged as in-group– simply because of the baseball hat showing support for the same team. In the context of social media profiles, this suggests that highlighting other kinds of mutualities beyond just those proxy variables can help overcome some of the stereotyping and biases that occur when we have to make quick decisions about people based on limited information.

[In this case, we might consider "Rare World" as a baseball hat of sorts– respondents with rare diseases prioritized disease similarity more than they said they might, allowing people to see an important mutuality beyond the immediately visible demographic characteristics.]

Future work should also consider not just how people make decisions about who to connect with online, but also the next steps in this interaction. What characteristics make for a strong peer support relationship offline? What characteristics are most relevant for support delivered remotely? How might we design for a supportive *community*, rather than transactional interactions? Similarly, if we are facilitating introductions for the sake of tangible support exchanges that will occur offline,

then trust and safety are important topics. Toch et al. [323] describe how people consider Facebook profiles as *"an honest signal, an identity carrying signal which is very hard to imitate."* The authors describe participants' use of pictures, friends, and comments to evaluate a person's trustworthiness. Donath and boyd [90] explain, *"knowing that someone is connected to people one already knows and trusts is one of the most basic ways of establishing trust with a new relationship."*

6.4.2 Building Helping Networks within Mainstream Social Media Sites

Where people with common chronic illnesses often prefer disease-specific communities that are separate from existing social media sites [158, 261, 305], the research in this dissertation has shown that these independent sites are not actively used by people with rare diseases because they lack the necessary critical mass. Instead, people with rare diseases rely more heavily on existing social networking sites are much more widely used by people with rare diseases, including for health purposes. They provide a place where people with rare diseases can more easily find each other for support. Thus, given that people with rare diseases are already on social media and already using these sites for health discussions, there is value in considering existing features that could be leveraged to create a "helping network".

Existing research has investigated mobilization attempts, *"broadcasted status updates in which people ask questions or request information, favors, or other forms of assistance"* on Facebook [99]. These requests address a range of social support types [100, 335] and might take the form of recommendations/social connections, factual knowledge, social coordination, favor requests, or opinions/polls [98], although the majority of mobilization attempts represent informational support requests [239]. Sites like Facebook are thought to lower *"the barriers for broadcasting requests for (and provisions of) support"* [99], but primarily for support that has a low cost to provide (i.e., requests that can be quickly fulfilled in response to a Facebook status or post, without requiring actions elsewhere on- or offline) [98]. Some of these mobilization attempts might make use of existing features, such as the "Looking for Recommendations" option or "Poll" option (Figure 6.7).

Asking for informational support on an social media is thought to be preferable to using a search engine because people trust their networks [239]. Help seekers can better target people who know them and may be better able to provide personalized support [238]. It also means they have access to a broader network of weak ties [132]. Ellison et al. [98] explain:



(a) Looking for Recommendations (b) Polls

Figure 6.7: Examples of Facebook features explicitly supporting mobilization attempts. Source: newsroom.fb.com

> "This broadcasting feature, a key component of most social networking sites, enables users to distribute content including requests for informational or emotional support, to a potentially large audience with little effort. The status update feature is helpful when seeking information because it enables users to broadcast to a wide set of ties (or even strangers, in the 'public' setting) through a single post, which is especially useful when one cannot identify the best person to fill a particular need."

Ellison et al. [98] argue that broadcasting support requests to a wide audience on social media is a benefit, allowing access to a diverse network of strong and weak ties [99, 123, 132, 181]. This is at odds with the self-presentation challenges associated with chronic illness [250, 261].

> Matching for social support requires additional insight into the conflict between a goal of receiving social support and the goal of impression management, *especially* if it takes place within a larger social network with mixed audiences and collapsed contexts [27].

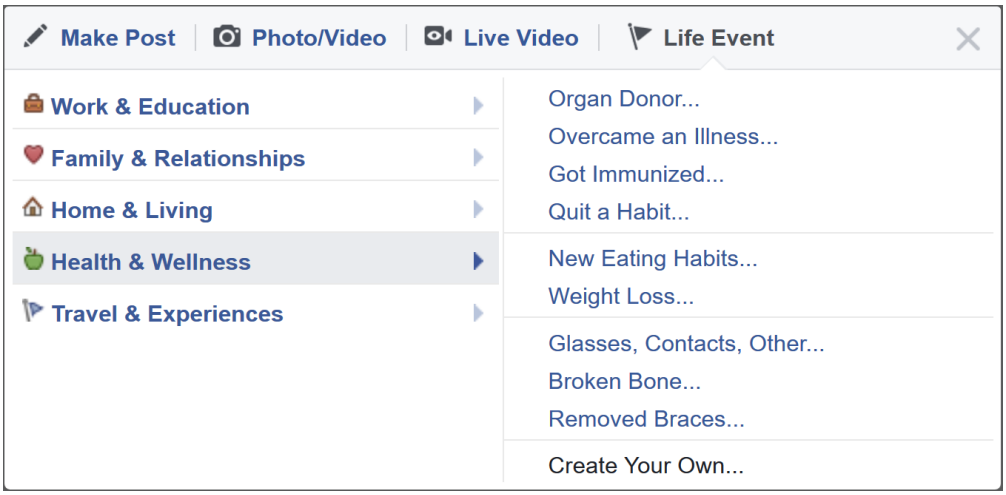


Figure 6.8: Health-related Life Events on Facebook. Source: newsroom.fb.com

Newman et al. [250] explain,

> "the need to express a need for support can run at odds with the need to present oneself as a positive, appealing member of the community. People naturally wanted others to view their health status and activities favorably and also did not want to overload or bore them with inappropriate communications. In addition, we found that people put considerable effort into to building and shaping their networks to more effectively pursue their health-related goals and to reduce the conflicts between those goals and impression management goals."

In the case of rare diseases, this trade off is particularly unclear; participants in our research have described a number of competing priorities, including raising awareness and advocating for their condition, minimizing family members' awareness fatigue, and desiring some sense of privacy or confidentiality. One possible feature of interest is "life events" (Figure 6.8). This feature allows people to self-identify as having been diagnosed with a disease recently. This could potentially be a useful tool for people with rare diseases to (optionally) broadcast to their network that they have a given rare disease, contributing to goals of advocacy and awareness. Declaring this life event could then unlock access to more private disease specific communities where people with rare diseases could share more personal or health-specific information.

For some people, adding a life event to their profile could contribute to their goals of raising awareness about their condition; some participants were very open about their experiences. Other participants, however, might find this information too private to share with Facebook friends or with the public. It is possible people could create an life event with privacy settings set to "Only Me"

that would still unlock access to a more private space. An interesting area of future research would be considering an "Only People Like Me" privacy setting, which would disclose the rare diagnosis only to other people within one's existing social network who also have a rare disease diagnosis; this could be particularly valuable for more stigmitized conditions.

There are also potential concerns about the use of the data by the social media platform (in this case, Facebook). Chronic illness bloggers have responded to the addition of "Health & Wellness" items to the life events feature with some concern:

> "Even though I very publicly fought cancer myself, I was a bit hesitant to enter this information in. I'm not worried about who sees it as much as I'm interested in how the company will use it. Could they use the data to advertise to me directly? Absolutely. But what else is possible with all of that information about one person. Only Facebook truly knows [253]."

This may be less of a barrier-to-use for people with rare diseases who, as we have seen in this research, are more open to sharing information on social media platforms to begin with. There are also concerns, however, about how audiences of Facebook friends and family might react to seeing these kinds of life events. If relationships between people with rare diseases and their social networks are already strained because of an over-emphasis on the disease, regular updates on Facebook may not help.

6.5 Summary

In this chapter, we investigated the possibility of using mainstream social media platforms to facilitate social support exchanges among people with people with chronic illnesses and especially people with rare diseases. We identified three major asymmetries:

1. We described the differences between the types of social support people are most interested in receiving and the support they are most interested in providing.
2. We provided an understanding of how people value different information on social media profiles of peer supporters, and discuss how these values differ from their behavior.
3. We discuss the information people seek about potential peer supporters and their comfort disclosing this same information about themselves to different audiences.

Part III

Considerations for Conducting Rare Disease Research

Conducting research with people with rare diseases is challenging. There are many different rare diseases each of which impacts, by definition, very small numbers of people. These people are geographically distributed around the world. Thus, conducting this research requires careful consideration to a range of questions. How might we ethically and effectively recruit participants for this research? How might we conduct this research remotely? How might we study rare diseases even when they are underrepresented in chronic illness data sets? What additional privacy risks do we need to consider when working with small, sensitive populations? I am not aware of any "handbook" on conducting rare disease research. Rather, I have drawn from best practices on ethics, privacy, and methodology from many different sources. In conducting this research, it has become clear that these recommended practices may not always be suitable for this unique population. I have also developed *new* methods as necessary. Thus, in this part of the dissertation, I provide a detailed and frank discussion of the methodological decisions made for this research, the new approaches developed, and lessons learned throughout this process. I answer the final research question, RQ4 (provided again for reference in Figure 6.9.)

In Chapter 7, I consider approaches to using machine learning with data sets with a relatively large number of people with common chronic illnesses and a relatively small number of people with rare diseases. The challenge here is that standard machine learning approaches can achieve high accuracy simply by predicting all the examples as common chronic illness. If, as in this dissertation, we are interested specifically in those rare disease examples, these results are insufficient. Thus, in this chapter I evaluate a number of different approaches that specifically address imbalance data and compare them against standard machine learning approaches.

In Chapter 8, I describe the Asynchronous Remote Communities (ARC) method. We developed this method to be able to conduct groups-based research with people with rare diseases who are

[RQ1] What are the experiences of people with rare diseases?

(a) How are these experiences similar to the experiences of people with common chronic illnesses?

(b) How are these experiences different from the experiences of people with common chronic illnesses?

(c) How might we support the unique experiences of people with rare diseases through design?

[RQ2] What are the support needs of people with rare diseases?

(a) To what extent are these support needs being met?

(b) What are the sources of support for people with rare diseases?

[RQ3] How might we facilitate tangible support exchanges among people with rare diseases?

(a) What kind of tasks do people need support with? Which tasks could people best provide support with?

(b) Which kinds of characteristics do we need to know about two people for them to be interested in connecting with each other?

(c) How comfortable are people sharing these characteristics on social platforms? How comfortable are they sharing this information within a private community of people with rare diseases?

[RQ4] How might we conduct research with people with rare diseases?

(a) What can we learn about people from self-reported online behavior?

(b) How might we analyze quantitative data that is heavily skewed towards people with common chronic illnesses?

(c) How might we conduct group-based, human-centered research when populations are dispersed?

(d) What privacy and ethics considerations are necessary in conducting research with this sensitive population?

Figure 6.9: Research questions.

geographically dispersed. This method was developed for the ARC study described earlier in this dissertation; in Chapter 8 I provide a detailed account of the ARC method and the analysis of the "success" of the method. ARC has also been adopted and adapted for studies with other populations, including pregnant women and new moms [275] and people living with HIV [210]; I review these adaptations, reflecting on the additional insights gained from these iterations.

In Chapter 9, I discuss considerations of privacy and ethics in rare disease research. First, I consider suggested best practices for ethical recruitment in online research and discuss the challenges of applying these recommended practices to rare disease research. I discuss our experiences in recruiting for the studies described in Part II and evaluate the effectiveness of these approaches. I discuss the tension between the convention of masking participant identities (thus preserving the privacy of people with rare diseases who share sensitive experiences in confidence) and the potential

benefits of naming participants (allowing for greater treatment of people with rare diseases as active collaborators in the research process and contributing to goals of increased awareness). I conclude by exploring ways of anonymizing rare disease survey data to the extent that it could be shared with other researchers to facilitate additional research and evaluating the loss in the utility of the data that occurs through this anonymization process.

This part of the dissertation provides researchers with detailed insights and sometimes frank discussions of the approaches we developed or adapted for this research, with the goal of being useful towards refining these methods further or using them with other sensitive, dispersed, small, or otherwise challenging populations.

“ *Perfectly ordered disorder designed with a helter-skelter magnificence.* ”

—Emily Carr in *Hundreds and Thousands*

Chapter 7

Learning from Imbalanced Data

Survey methodologies, particularly those distributed online, are naturally suited to studying dispersed populations. In Chapters 4 and 6, we used survey methods to draw comparisons between rare disease populations and chronic common illness populations. Here we were faced with the challenge of dealing with skewed data; this problem is called *class imbalance* in machine learning [49]. Given the rarity of the diseases, we often have more responses from people with common illnesses than from people with rare diseases. If we consider people with a common chronic illness as one class and people with a rare disease as another, there is a huge skew towards people with common diseases.

> Simply adopting standard learning techniques can achieve high accuracy by predicting all the examples as the majority class (common chronic illnesses); this is problematic because our main interest in this dissertation is people with rare diseases.

The nature of this class imbalance problem (see Figure 7.1) is that the cost associated with misclassifying the minority class (in this case, rare diseases) is higher than misclassifying the majority class (common chronic illnesses). If we misclassify someone with a rare disease as having a common chronic illness, this furthers their existing frustrations with the medical system, their cost of care, their isolation, and depression. In machine learning terms, the cost matrix does not contain uniform cost values but rather has a higher cost for the instances that are rare diseases but classified as

		Actual Disease Type	
		Rare Disease	Common Illness
Predicted Disease Type	Rare Disease	Correctly Classified Rare Diseases	Missed Common Illness
	Common Illness	Missed Rare Disease	Correctly Classified Common Illnesses

Figure 7.1: Relative costs of errors in identifying people with rare diseases from imbalanced data.

common chronic illnesses than for those that are common chronic illness but are classified as rare. If we assume the class of rare disease as the positive class, then we prefer recall (how many relevant items are selected) rather than precision (how many selected items are relevant).

In this chapter, we explore approaches to dealing with this imbalance problem in rare disease research. We review approaches from existing research, primarily pre-processing methods that adjust the distribution of the data. We then explore Yang et al.'s [362] Soft-FGB in more depth; this approach handles the imbalance as part of the classification process (as opposed to a pre-processing step) and allows us to trade off between precision and recall. We compare the performance of these different methods (pre-processing approaches, Soft-FGB) to more traditional, standard machine learning methods using our survey data sets from Chapters 4 and 6. This work was conducted collaboratively with Shuo Yang, Kim Oakes, Kay Connelly, and Sriraam Natarajan and this chapter contains material from our 2016 CHASE paper [209].

7.1 Analyzing Imbalanced Data in Machine Learning

Machine learning researchers typically handle imbalanced data as a pre-processing step. Most simply, researchers will randomly undersample the majority class (i.e., using less of the common chronic illness examples and using all the rare disease examples, resulting in a more balanced, smaller data set overall) or randomly oversample the minority class (i.e., using more of the rare disease examples, while keeping the common chronic illness examples constant, resulting in a more balanced, larger data set overall). Many researchers have explored ways of doing this over/undersampling more intelligently, as well as the effectiveness of each approach. Japkowicz [166] evaluated both *random* over/undersampling and *focused* over/undersampling, where only examples nearest to the decision boundary are differently sampled. The authors found that all approaches were reasonably effective

and the more sophisticated sampling techniques did not give any clear advantage (in the domain studied). Ling and Li [194] used a combination of oversampling *and* undersampling, finding that the best results are achieved when classes are equally represented. The authors found that this does not necessarily have to be achieved through the combination approach; using both sampling methods did not outperform simply undersampling the majority class. Chawla et al. [50] introduced Synthetic Minority Oversampling Technique (SMOTE), a state of the art oversampling method that generates synthetic examples along the line segments joining the minority examples with their five nearest neighbours.

[Recently, Yang et al. [209,362] developed an approach to learning from imbalanced data by explicitly trading-off between false positives and false negatives. This allows us to incorporate knowledge about the domain (in this case, rare diseases and common chronic illnesses) into the cost matrix.]

Specifically, Yang et al. [362] introduce a penalty term into the objective function of a learning algorithm that allows for the trade-off between the precision and recall to be tuned during the learning process. The learning algorithm Yang et al. [362] employ is called Functional Gradient Boosting (FGB) [86,119,176,177,245,362]. Here, we use y to denote the class variable, $\mathbf{X}$ to be the set of features, x^j to be the j^{th} feature and the suffix i to denote the i^{th} example. Thus $\hat{y}_i$ is the true label for the i^{th} example and $\mathbf{X}_i$ is the set of all features for the i^{th} example. In our case, each survey respondent is an example, their disease type (rare/common) is the class, and their answers to all the survey questions is the feature values.

The goal of many probabilistic models is to learn the distribution $P(y_i|\mathbf{X}_i)$ for all examples i. Given a training set, most methods optimize the loglikelihood over the training set, where loglikelihood is given as $LL = \sum_i log(P(y_i|\mathbf{X}_i))$. Gradient-descent is usually performed on the loglikelihood to find the set of parameters that best model the training data. FGB methods employ a similar approach with two key differences. First, they represent the conditional distributions using a sigmoid function,

$$P(y_i|\mathbf{X}_i) = \frac{e^{\psi(y_i=\hat{y}_i|\mathbf{X}_i)}}{\sum_{y'_i} e^{\psi(y_i=(y'_1|\mathbf{X}_i)}} \tag{7.1}$$

where the denominator is the sum over all the possible label values of the example. Given that our classification task is binary, this sum is performed over the common and rare disease probabilities. Now the loglikelihood can be written as,

$$\log LL = \sum_i [\psi(y_i = \hat{y}_i; \mathbf{X}_i) - \log \sum_{y'_i} \exp\{\psi(y'_i; \mathbf{X}_i)\}] \tag{7.2}$$

The second key difference between an FGB approach and a standard gradient-descent approach is the process of obtaining the gradients. While standard methods differentiate LL with respect to $P(y|\mathbf{X})$ (the parameters of the distribution), FGB methods differentiate LL with respect to ψ (the function that models this conditional distribution). Friedman [119] took this derivative with respect to each training example i. This is an approximation of the true gradient but led to excellent results in many real-tasks [86, 119, 176, 177, 245, 362].

The gradient of equation 7.2 with respect to the potentials (ψ_i) for each example i is:

$$\Delta(y_i) \; = \; I(\hat{y}_i = Rare) - P(y_i = Rare; \mathbf{X}_i), \tag{7.3}$$

where I is an indicator function which returns 1 for rare diseases and 0 for common illnesses.

This gradient depends on how well the current model fits the true label of the example, and then is assigned to each example to generate a regression dataset. If it fits well, the gradient would approach 0 and if it does not, the predicted probability of the example would be far from the true label and hence make the boosting algorithm attach a high weight to that example. Then in the next iteration, we learn a regression function to fit the current regression dataset and add this to the model to improve the probabilistic predictions. As these iterations go on, the probability of all positive examples is pushed towards 1, and all the negative examples towards 0. This process is shown in Figure 7.2.

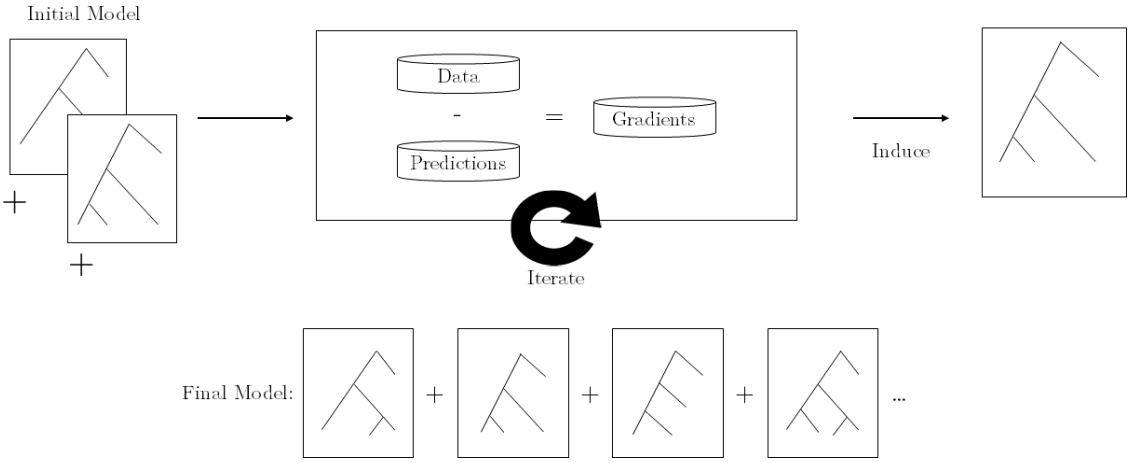


Figure 7.2: Functional gradient boosting process.

While this method is successful, it still treats misclassified positive and negative examples equally. In this dissertation, it is important that we classify rare diseases more correctly; the goal is to achieve a higher recall for rare diseases. Yang et al. [362] achieve this by modifying the objective function to incorporate this cost difference. Specifically, the authors add a term to the objective function so the positive and negative examples can be penalized differently:

$$c(\hat{y}_i, y) = \alpha\, I(\hat{y}_i = 1 \wedge y = 0) \; + \; \beta I(\hat{y}_i = 0 \wedge y = 1),$$

where $\hat{y}_i$ is the true label of the i^{th} instance and y is the predicted label. $c(\hat{y}_i, y) = 0$ when the example has been correctly classified. $c(\hat{y}_i, y) = \alpha$ when it is a rare disease example but classified as common, while $c(\hat{y}_i, y) = \beta$ when it is a common chronic illness example but classified as rare.

The new objective function is now:

$$\log J = \sum_i [\psi(y_i; \mathbf{X}_i) - \log \sum_{y_i'} \exp\{\psi(y_i'; \mathbf{X}_i) + c(\hat{y}_i, y_i')\}] \tag{7.4}$$

Note the difference to the original FGB function in Equation 7.2; this new objective function includes a cost function that allows for different misclassification costs for different labels. In our cases, missing rare diseases is costlier than missing common diseases. By setting the parameters $\alpha > 0$ and $\beta < 0$, the different costs of false positive and false negative examples can be incorporated into the learning process, hence the trade-off between precision and recall can be controlled.

7.2 Experimental Results of Identifying Rare Examples

To evaluate approaches to handling this imbalance problem, we conduct experiments using two data sets: our survey data on uses of technology and sources of support (described in Chapter 4) and our survey data on helper selection and information sharing preferences (described in Chapter 6). Both these data sets are heavily skewed towards common chronic illnesses, despite our deliberate focus on recruiting rare disease respondents. The first data set included 30% rare disease responses and 60% common chronic illness responses. The second data set included 20.83% rare disease responses and 74% common chronic illness responses. Our experiments focus on answering two key questions:

Q1: How effective is a class imbalance approach in detecting rare diseases using self-reported behavioural data?

Q2: How effective is Yang et al.'s [209, 362] method in handling class imbalance as part of the classification process (as opposed to changing the class distribution)?

To answer **Q1**, we compare four standard, widely used supervised classification methods (Naïve Bayes (NB), Logistic Regression (LR), 5-Nearest Neighbors (NN), and Decision Trees (DT)) that are not specifically designed to handle class imbalance against four class imbalance methods (random undersampling, random oversampling, SMOTE [50], and Yang et al. [209,362] cost-sensitive boosted probabilistic classifier).

To answer **Q2**, we compare Yang et al.'s [362] cost-sensitive boosted probabilistic classifier against two random sampling methods, as these methods have been shown to be effective in imbalanced datasets [166, 194]. We additionally compare against SMOTE [50], a state of the art oversampling method that generates synthetic examples along the line segments joining the minority examples with their five nearest neighbours.

In all experiments, we performed 10-fold cross validation, and the test sets were consistent between experiments. The results of these experiments (Table 7.1) are presented as averages of the results from the ten test sets. Standard evaluation metrics include accuracy, Area Under the Receiver Operating Characteristic Curve (AUC-ROC) and F_1 (the harmonic mean of precision and recall). We include these metrics, which measure accuracy with a balanced weight between positive and negative examples, but we focus primarily on evaluation metrics that assign higher weights to higher recall regions.

			Standard Classification Methods				*Class Imbalance Classification Methods*			
			NB	**LR**	**5-NN**	**DT**	**Oversample**	**Undersample**	**SMOTE**	**Soft-FGB**
Data from Chapter 4	*Standard Metrics*	**FPR***	0.185	0.263	0.088	0.188	0.226	0.292	0.214	0.158
		TNR	0.815	0.137	0.912	0.812	0.174	0.108	0.186	0.242
		AUC-ROC	0.149	0.615	0.684	0.612	0.142	0.159	0.685	0.117
		Accuracy	0.113	0.641	0.124	0.690	0.687	0.688	0.690	0.467
		F	0.514	0.401	0.360	0.437	0.491	0.566	0.478	0.523
	Class Imbalance Metrics	**TPR**	0.507	0.413	0.288	0.408	0.511	0.657	0.472	0.972
		FNR*	0.492	0.587	0.112	0.592	0.489	0.343	0.528	0.028
		F_3	0.507	0.400	0.299	0.412	0.504	0.632	0.471	0.825
		F_5	0.507	0.402	0.293	0.410	0.508	0.647	0.472	0.909
Data from Chapter 6	*Standard Metrics*	**FPR***	0.228	0.108	0.191	0.021	0.244	0.222	0.224	0.530
		TNR	0.772	0.892	0.809	0.979	0.756	0.778	0.776	0.469
		AUC-ROC	0.804	0.942	0.796	0.549	0.861	0.868	0.837	0.608
		Accuracy	0.725	0.818	0.693	0.760	0.818	0.816	0.808	0.572
		F	0.488	0.590	0.314	0.051	0.829	0.823	0.770	0.488
	Class Imbalance Metrics	**TPR**	0.569	0.569	0.306	0.028	0.880	0.855	0.861	0.956
		FNR*	0.431	0.431	0.694	0.972	0.120	0.145	0.139	0.044
		F_3	0.551	0.573	0.308	0.031	0.869	0.848	0.841	0.783
		F_5	0.562	0.571	0.307	0.029	0.876	0.852	0.853	0.877

Table 7.1: Experimental results for predicting rare diseases. Green indicates best performance. Asterisk indicates the metric should be minimized (all other metrics should be maximized).

Specifically, we report on two additional metrics: F_3, and F_5, where this F-measure is:

$$F_\beta = (1 + \beta^2)\frac{Precision \cdot Recall}{(\beta^2 \cdot Precision) + Recall}$$

where β is the importance given to recall over precision (i.e. a higher β indicates more emphasis on recall and a smaller β indicates more emphasis on precision). We use F_3 and F_5 to increase the importance of recall over precision. We also report the confusion matrix for each experiment.

> The class imbalance methods generally outperform the standard methods at identifying rare examples (true positive rate, false negative rate, F_3, and F_5). Thus, **Q1** can be answered affirmatively in that class imbalance methods are more effective at identifying people with rare diseases.

There is a slight trade off when using these methods in our ability to identify common examples (see false positive rate, true negative rate, accuracy). We see this trade off as acceptable; we would rather falsely identify someone as having a rare disease who actually has a common chronic illness than miss someone who actually has a rare disease. If we can identify someone who *may* have a rare disease, we can direct them towards appropriate medical and clinical resources to find out with greater certainty.

> Additionally, we can see that Yang et al.'s [362] approach comfortably outperforms other class imbalance methods in many respects. It performs substantially better on every class imbalance evaluation metric (true positive rate, false negative rate, F_3, F_5) without losing too much performance on the standard evaluation metrics. Thus, **Q2** can also be answered affirmatively.

7.3 Summary

The results of these experiments illustrate that it is possible to identify people with rare diseases from several different data sets using a Soft-FGB approach and this approach is preferable not only to standard machine learning approaches but also preferable to methods that handle imbalance as a

pre-processing step. Sampling methods alter the class distribution in the training set, which makes the training set not as reflective of the true test set. It is therefore unsurprising that a method that handles class imbalance in a principled manner would outperform a sampling method. This leads to increased opportunities for rare disease research to take place; there may be future opportunities to learn more about rare diseases using chronic illness data sets even if they contain only a small number of rare examples.

“ *Learn from the mistakes of others.*
You can't live long enough to make them all yourself. ”

— Eleanor Roosevelt

Chapter 8

Asynchronously Conducting Remote, Group-Based Research

Research methods like interviews, focus groups, design workshops, and role-playing are largely intended to be conducted co-located and synchronously with study populations to make significant contributions and impact the designs of future systems. Co-located studies are not always possible for group-based methods when participants have limited travel access or when social, political, or economic climates inhibit a potential participant's ability to assemble. In some cases, researchers may not have access to enough participants in their local areas. Finally, participants may not feel comfortable in a physical study setting, whether being physically present or allowing researchers into their environments. These barriers to co-located research are particularly relevant when conducting research with people with rare diseases; potential participants are geographically distributed and, depending on the specific condition, there may be additional accessibility barriers to assembling.

> Researchers need ways of conducting group-based studies online to ensure perspectives from underserved populations are considered when designing new systems, and improve the scientific merit of our research by expanding sample sizes to reach theoretical saturation or statistical power.

Although previous work in HCI has addressed remote strategies for interviews via instant messaging [337], email and phone [89], and or video chat [151], there has been little discussion in HCI on remote *group-based* methods that allow participants to interact with each other. Some

research in the social sciences has reflected on approaches to web-based focus groups [309,316]; these can be done synchronously with participants attending at the same time via an online platform or asynchronously, with participants discussing via online bulletin boards or forums.

In this chapter, I describe an approach to conducting group-based needs assessment research using Asynchronous Remote Communities (ARC). This approach is builds on existing social science research on asynchronous, web-based focus groups but is augmented with additional activities often used in design research (diaries, scenarios, personas, etc.) to better address the specific goals of HCI research, as well as to allow for data triangulation. We piloted this method with people with rare diseases for the study described in Chapter 5. Where Chapter 5 described the results of that study, this chapter describes the ARC method itself and the activities we used to collect information about the needs of this distributed and remote population.

This chapter relies most heavily on material from our 2017 EAI Endorsed Transactions on Pervasive Health and Technology journal paper [206], written with Ben Jelen, Annu Prabhakar, Lora Oehlberg, Katie Siek, and Kay Connelly. It secondarily contains material from additional CHI and Pervasive Health publications [205, 210, 275], written with Jordan Beck, Ciabhan Connelly, Kay Connelly Lucia Guerra-Reyes, Ben Jelen, Vanessa M. Kleinschmidt, Juan Fernando Maestre, Julia Dunbar, Annu Prabhakar, Katie Siek, and Patrick Shih. Our primary contribution here is a detailed, in depth description of the ARC method such that other researchers may replicate, modify, and improve upon this method.

8.1 Human Centered Research Methods

In this section, we discuss existing approaches to conducting HCI research remotely, and provide a brief overview of some commonly used research methods that have inspired and influenced our own methodological choices.

8.1.1 Remote Methods

Some discussion has occurred on best practices for conducting research with participants remotely. In particular, researchers have proposed strategies for remote interviews, because they are considered to be valuable, but challenging methods. Voida et al. [337] employed instant messaging for conducting

interviews remotely and noted that attention expectations, timing, context, and persistence are all impacted as a result of the text-based method. Dimond et al. [89] extend this work to include email and phone as additional methods of qualitative data collection, noting that synchronous interviews (by phone) tended to produce much longer transcripts than semi-synchronous (instant message) or asynchronous (email) methods, but that they do not necessarily produce more unique ideas. Hillman et al. [151] provided an in-depth discussion of their approach to conducting interviews over video chat, and the challenges they faced.

Focus groups, although often used for needs assessments, have not seen the same level of discussion within sociotechnical research communities about their adaptability for remote populations. Researchers in social science and market research suggested real-time virtual focus group with people participating through online bulletin boards [316], as well as online communities [309]. These discussions do not necessarily take into account the specific goals of needs assessments in HCI, or the types of activities conducted in design research. In our work, we rely on Sweet's [316] guidelines for online focus groups, but augment their method by introducing some additional HCI research methods, translated to an online format.

8.1.2 (Co-located) Group-based Research Methods

Group-based methods add value by allowing researchers to obtain a range of viewpoints in less time. They may mitigate the shortcomings of one-on-one methods (e.g., empowering quieter participants to contribute as they are comfortable) and can engender interaction between participants, enabling them to raise ideas they might not have thought of on their own [187]. Here we provide a brief overview of a selection of these methods; many of them are group-based methods, however we also leveraged some one-on-one methods and adapted them for a online and group settings.

Focus Groups have the benefit of receiving many perspectives at once and allow participants to build on each other's contributions. Focus group participants, however, may be unwilling to discuss sensitive topics in a group setting and talkative/opinionated participants can monopolize conversations [34]. These challenges are overcome by skillful facilitation and careful moderation; a trained focus group moderator and a design researcher might work side by side [187].

Surveys are powerful tools for getting data from larger samples and are often used to describe populations or explain behaviors [12]. Surveys are often used alongside other human centered

research methods, as a way of validating findings across a larger population, confirming the results of qualitative analysis with the existing study population, or obtaining baseline information about an understudied population [108].

Diaries are useful when asking people to reflect on things that change over time, like mood or opinion [3]. They may provide more accurate information, because people are expected to document events as they go, instead of recalling past events [187]. Participants may not adhere to the study expectations (e.g., retroactively filling in data shortly before a study session, known as "parking lot compliance" [310]) which can impact the reliability of the data. There is room for a variety of media in diary studies, such as text, photos, audio, and video (e.g., [2, 33, 39, 47]) captured using a range of tools like paper, online forms, mobile phones, or cameras (e.g., [2, 47, 114, 260]).

Personas & Scenarios give designers a specific person or context to design for, without relying too much on an actual person's idea of how to address the problem. Cooper [66] argues that *"merely being the victim of a particular problem doesn't automatically bestow on one the power to see its solution."* These can be developed *with* participants to discover these scenarios personas [18] or can allow existing findings to be reframed in a way useful to design [364].

8.2 The Asynchronous, Remote, Communities (ARC) Method

In this section, we introduce the ARC method, an approach to conducing human centered needs assessment research remotely. We provided details of this method and consider its effectiveness. In determining "effectiveness," we were looking for useful and honest responses, participation over time by different people, and that the activities be comfortable for participants. We collected,

- comments and posts from the study group,
- metadata (timestamps, likes, "seen by"),
- private Facebook messages and email threads we received from participants,
- survey responses from Typeform [1],
- Messages submitted to Google Voice [2], and
- summaries of discussions in Facebook groups used for recruitment.

[1] http://www.typeform.com/
[2] http://voice.google.com/

Two researchers reviewed all qualitative data and iterated on codes until converging on an inter-rater reliability (Cohen's Kappa) score of 0.78. We examined,

- *conversational flow* (relevance of each comment to the main post or where two versions of similar conversations co-occurred in multiple threads),
- *comments about activities* (feedback about an activity, comments contextualizing participation, apologies for delayed responses, expressions of confusion), and
- *life events influencing participation* (e.g., a relative passing away, being hospitalized, having a job interview).

We used Tableau [3] to visualize the Facebook metadata and responses to the closed-ended survey questions, as well as simple analytics to compare what participants actually did and what they said they did. We also conducted social network analysis to understand the strength of the relationships between participants where we identified emerging sub-communities [144]. We counted interactions between participants (either a participant's comment in response to another's post/comment or a participant liking another's post/comment). These counts determined the weights of the edges connecting the participant nodes. We used Gephi [4] to calculate the modularity (used for detecting clusters or communities in networks) and to visualize these relationships.

8.2.1 Pre-Study Interactions

Recruitment.

We recruited adults ($\geq$ 18 years old) with a rare disease from Facebook support groups. We posted a link to our study information sheet to each group along with our recruitment message. Group members who were interested were instructed to contact the researchers by Facebook message or email. Because we would be conducting the study on Facebook, we recruited only from Facebook. We did not want to create additional privacy risks by encouraging people to share their data on Facebook who were not already doing so. Most groups from which we recruited were groups we had been a part of for over a year. One researcher initially joined these groups to recruit for past research and found members of these communities to be enthusiastic about our presence and eager to be involved in research.

[3] http://www.tableau.com/
[4] http://gephi.github.io/

This researcher was known to group members and had built rapport by sharing results of previous research and contributing to the community in other ways (e.g., by helping to access academic articles of interest located behind paywalls). In these groups, questions about the study were easily satisfied. If a group member had a question about the study, a different group member (usually a participant from a previous study) would jump to the researcher's defense, vouching for the researcher's legitimacy and expressing gratitude for ensuring their condition was represented in research projects.

> ***Lesson #1:*** We recommend taking the time to build a strong rapport with group members before, during, and after the study, following not only best practices for recruiting [38], but also good practices for maintaining those relationships at the conclusion of the study [6].

Informed Consent.

We used email or private Facebook messages to send the informed consent document to anyone expressing interest in the study. Potential participants were instructed to review the document, print it, sign it, scan or photograph the signature page, and return it to the research team by email or private Facebook message.

> We required a physical signature, instead of a typed response or digital signature, to ensure participants took the informed consent process seriously, particularly in the absence of a physically present researcher to walk through the document with them. Unfortunately, this was a barrier for many participants.

We distributed 53 consent documents and 14 were returned. Many people who did not return the informed consent document (as well as some that did) struggled with the process of completing this document via printing, signing, and digitally sending it to the research team, despite not having communicated any concerns about the study itself to the research team. Some of these difficulties were technical (*"I might need to do a screen shot if I can't get the scanner to work, but is [tomorrow] okay?"*) and some were physical (*"I will have to wait for my dad to get off of work so he can print is and scan it for me. Sorry for the inconvenience, but if you can bare with me, I am in! I need*

something to do and I want to help...can I just email you that I give you my permission?"). We were able to enroll an appropriate number of participants to ensure there were enough people to actively engage in discussions without being so large that participants could not reasonably get to know each other. However, we recognize that requiring a physical signature may have limited participants by physical and technological ability.

For in-person studies, participants are not always interested in taking time to read the informed consent sheet and would be happy to sign without reading. As ethical researchers, it is our responsibility to talk through the information with participants to ensure they understand what they are consenting to. When conducting studies remotely, we do not have the same opportunity to make in-the-moment assessments of how much attention the participant has given to the document and how much s/he understood, but this does not make *informed* consent any less critical [40].

> ***Lesson #2:*** We recommend investigating alternative methods of consenting electronically. One idea would be to seek consent via Facebook chat, mimicking how a researcher might walk through a paper consent form with a participant in person, pausing after each section for questions. Alternatively, require participants to pass a short quiz on the highlights of the consent document. Participants who do not pass the quiz should be contacted by a researcher to discuss the contents of the document and ensure they understand.

8.2.2 Procedure Overview

We created a private Facebook group for this study and invited people who completed the informed consent process. We chose Facebook as the platform on which we conducted this research because it is actively used by people with rare diseases as a way of connecting with each other, (more than online platforms specifically for rare diseases (see Chapters 3 and 4). We conducted the study over the course of 22 weeks, beginning with a 5 day period of introductions to help participants get to know each other and become comfortable in the group environment. We then introduced 11 activities (summarized as A1–A11 in Table 8.1) over the course of the remaining 21 weeks.

#	Name	Activity	Shared?	Duration	G/R	Tool	Media
A1	Diary	Participants tracked interactions they had with other people about their disease. They documented who they talked to, how they communicated, what they discussed, and how they felt about it.	Directly	24 hrs	R	Choice	Text
A2	Circles	Participants used household objects to illustrate how comfortable they were sharing information with different people by drawing circles with themselves at the center and placing people at different distances from the center.	Directly	Once	G	Other	Other
A3	Questions	Participants made a list of questions they wished their friends/family would ask them about their disease.	Directly	Once	R	Group	Text
A4	Problems	Participants ranked a list of problems (provided by the researchers) by the relevance to them personally.	Directly	Once	R	Group	Text
A5	Photo Elicitation	Participants took photos representing two main themes from A4 and commented on each others' photos.	Anon.	3 days	R	Phone	Photo
A6	Solutions	From the problems in A4, participants discussed strategies or solutions they had used to address these problems.	Directly	Once	G	Group	Text
A7	Mad Lib	Participants created a mad lib, detailing the more humorous aspects of conversations they have surrounding their conditions.	Directly	Once	G	Group	Text
A8	Movie Script	Participants wrote a script for a movie about their life, taking place between them and a friend or family member.	Directly	Once	G	Group	Text
A9	Rant Line	We set up a "Rant Line," a Google Voice number that participants could call, text, or send photos to anytime they needed or wanted to rant about something.	No	3 days	R	Phone	Other
A10	Personas	We created and posted two personas and asked participants to critique them and discuss how representative they were of their own lives.	Directly	Once	G	Group	Text
A11	Survey	Participants completed a survey to debrief their experiences in the study.	No	Once	R	Website	Text

Table 8.1: Activities used in the study. **Shared:** Responses were shared *directly* to the group, collected by researchers and *anonymized* before being shared, or were *not* shared with the group at all. **Duration:** Period of time over which the activity occurred. **Generative/Recall (G/R):** Activity involved *recalling* some past experience or *generating* new material abstracted from past experiences. **Tool:** Activities required participants to use specific tools such as the Facebook *group*, a *phone*, or a *website* while others allowed participants their *choice* of tool. **Media:** Activities were submitted as *text* or as *photographs*.

We also sometimes posted research updates and participants frequently started new conversations on their own. At the request of one participant, we tagged activity posts (starting from the second activity) with [ACTIVITY] so participants could tell the difference between posts that were "official" research tasks and "less important" posts from researchers or other participants.

8.2.3 Activities

In this section, we describe participants engagement with and reactions to the various study activities (in depth descriptions of each activity are provided in Appendix E). Table 8.1 categorizes each activity by:

- *Shared.* Was the activity response shared with other study participants directly, anonymized first by the researcher, or not shared at all?
- *Duration of Activity.* Was this a one time activity or did it take place over time?
- *Generative vs. Recall.* Did the activity involve recalling past experiences or generating new artifacts or opinions?
- *Tool.* What tools were used to complete each activity?
- *Media type.* Was the activity text, drawing, photo, or voice based?

Categories of Activities

We categorized activities by how responses were *shared*. For many activities, participants were instructed to post directly to the group. In A5, the submissions were sent to the researcher privately and then anonymized before being shared with the group. In A9 and A11, the submissions were made anonymously and directly to the researcher and were not shared with the group at all. We saw more likes and comments when participants posted directly to the group ($\bar{x} = 11.13$ comments/posts) than when they responded in an anonymous or semi-anonymous way ($\bar{x} = 5.75$ comments/posts).

> We speculate that seeing others contributions may help to remind participants to submit their own (by showing up in their news feed more often) or inspire participants who are unsure of what to submit.

We categorized activities by *duration* (completed in one sitting or over the course of several hours/days). There was more engagement in activities that took place over time ($\bar{x} = 17$ com-

ments/posts) than for one-time activities ($\bar{x} = 15.6$ comments/posts). Comments on one-time activities were often confined to a single thread, whereas discussions on longer term activities took place over several threads, with participants adding their own new material as they came up with it. In both cases, participants continued to respond to each other even after the activity ended.

We also categorized activities by whether they required *recalling* personal information or *generating* new material. We saw more comments when participants recalled information ($\bar{x} = 18.2$ comments/posts) than when they had to be creative ($\bar{x} = 10.2$ comments/posts). When asked to recall information, they distributed their responses over several comments, and built off each others' experiences. There was very little of this when activities were generative; they typically only posted the results of their efforts and did not discuss each others' contributions. One participant found this discouraging and stated (in reference to A8), *"Iwanted to know what others thought of my script and was a bit hurt when I didn't get any feedback."* In A5, part of the activity was to comment and discuss each others' submissions, which did lead to more feedback.

> ***Lesson #3:*** We recommend encouraging participants to build on each other's creative contributions. This can be challenging on social media if participants are not observing each other as the work is being created, and may need to be encouraged explicitly.

Additionally, we reflect on the differences in *tools* used to complete activities. We saw that activities that only required the Facebook group to complete received more comments and likes ($\bar{x} = 11.57$ comments/posts) than activities requiring some other tool, such as a phone, website, or physical objects ($\bar{x} = 6.50$ comments/posts). In the case of A1, where participants had the choice of how they kept track of data, the results were somewhere in between ($\bar{x} = 10.00$ comments/posts).

Finally, we compare differences in *media*. We saw that activities requiring only textual responses had more activity ($\bar{x} = 10.88$ comments/posts) than activities requiring other media ($\bar{x} = 6.67$ comments/posts).

Activity Preferences

Participants eagerly volunteered feedback about activities throughout the study and in the debrief survey (A11). They looked favorably on most activities; almost all activities had a median rating of 6 on a 7-point scale (1 = "*Hated it*!", 7 = "*Loved it*!"). Participants enjoyed creative activities (*"It*

was fun to be able to engage my creative side" (A2)).

A1 (*median* = 5, *modes* = 4, 5, 6) and A8 (*median* = 5, *mode* = 5) were not as popular. Some participants described being confused by the creative activities: *"I just couldn't understand what was wanted, despite the examples."* They also seemed uninspired sometimes: *"I really don't understand what to do here, [Researcher's Name]. I'm big on taking photos but I don't get how I can take a photo of someone not caring or understanding"* (A5). We posted abstract examples or sample responses from other domains to help provide a sense of what a response could be without leading too much. We also provided clarification and answered a number of questions. Especially for A5 and A7, these examples and clarifications were insufficient, as there was still a lot of questions. Instead, participants seemed to do better when they were following the lead of a fellow participant; in the case of A7, once one or two participants had posted mad libs, many others followed. In A5, participants submitted photos to the researcher rather than to the group, so participants did not get to see each other's contributions until much later.

> ***Lesson #4:*** We recommend having participants post directly to the group, even work in progress. Seeing other participant's contributions can give inspiration to people who are hesitant to contribute creatively. This also increases participants' awareness of group activity (through notifications or news feed items).

After completing A1, one participant remarked, *"Never spoke to a soul in person...Right now being homebound because of the illness, my life is pathetically limited."* Survey responses (A11) confirmed this sentiment: *"I am homebound and have very few interactions with others. The activity pointed that out to me yet again."* and *"Just hit home how often I have to talk to the doctors and how often I don't talk to other people in a day"* and *"kind of overwhelms or can depress me as I rarely see actual people."* This activity drew attention to negative aspects of life that were uncomfortable to reflect on.

> ***Lesson #5:*** We advise exercising caution when selecting activities; conducting research asynchronously and online means researchers do not have the same chance to assess how a method is going as it is happening. By the time researchers are aware of discomfort, it may be too late to adjust the method.

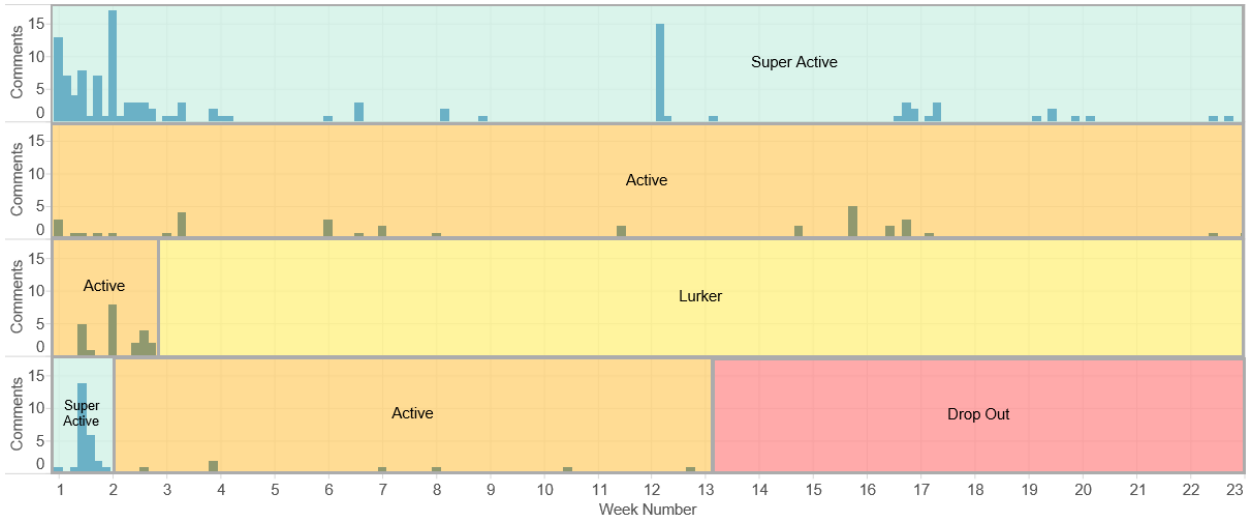


Figure 8.1: Comments throughout the study with examples of engagement levels

Levels of Engagement

We observed four different levels of engagement which we labeled super active, active, lurking, and dropped out. Super active meant commenting, liking, and posting frequently. Active participation meant contributing steadily, but not to the same volume as super active participants. Participants in a state of active participation contributed mostly by commenting rather than liking posts; they responded to posts by researchers but were less likely to initiate new discussions or engage in conversations with each other. Lurking meant seeing posts but not engaging with them in any way. Finally, dropped out meant the participant had actually left the group. Participants transitioned between these states. No participant became more active; some participants maintained a steady level of activity, while others became less active over the course of the study (Figure 8.1).

Engagement Over Time

We observed a burst of activity in the first three weeks as participants got to know each other. There were an average of 16.1 comments per day during the first 3 weeks ($\sigma = 14.0$) but only 1.3 comments per day for the remainder of the study ($\sigma = 3.4$). Participation peaked on Saturdays and slowly decreased throughout the week until Friday, even though many participants were unemployed.

Lesson #6: We recommend taking time to understand potential participants' Facebook behaviour to inform decisions about when and how often to post, as well as the overall study duration.

Participants typically responded to new activities within 24 hours (Figure 8.2), unless the activity took place over several days (e.g., A9). When participants did not respond within 24 hours, this was a sign that something was wrong; sometimes they missed notifications about new activities (*"sorry I didn't see this pop up!"*), had to take a break from the study (*"I'm just back in [city] today after 10 days in [another city] I am catching up."*), or forgot to do an activity they had seen (*"I completely forgot about the rant line. But I sure could have used it this morning."*). We tried four approaches to encouraging participation:

1. **Reminding participants of activities.** When posting a new activity, we reminded participants about the last activity. We saw a brief increase in responses after reminders.
2. **Pinning the current activity.** For part two of A5 (sharing photos with the group and asking participants to comment), participants could not find the photos within the group. We tried pinning the album to the top of the group, but this was an ineffective strategy and participants were still confused.
3. **Tagging participants in posts.** After seeing a reminder, some participants asked for clarification on which activity the reminder referred to. We tagged participants who expressed confusion in the comment section of the relevant post. This was more effective than pinning posts to the top of the group because participants could easily click the notification.
4. **Commenting on posts.** For activities where participants were uncharacteristically quiet, we commented on the post to provide clarification (in case of confusion) and to increase the likelihood the post would show up in their newsfeed (in case they had missed a notification). This strategy was effective; there was an increase in participation following the comment.

Despite these attempts to encourage participation, participants still mentioned in A11 that they were unaware of activities. It is challenging to work within the restrictions of Facebook to ensure posts are brought to a participant's attention.

Lesson #7: Researchers should be prepared to adopt multiple approaches to ensure activities are seen.

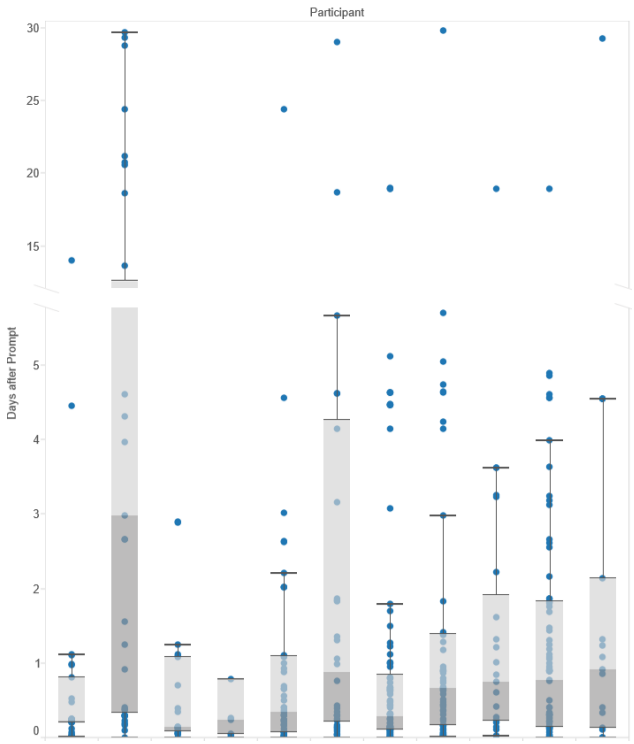


Figure 8.2: Days until each participant's first comment on a post. Two participants are omitted–one spent the entire study lurking and the other participant dropped out early in the study.

8.2.4 Overall Engagement

We observed that several people revisited posts long after they had been posted initially or would do several activities at once (Figure 8.2). This is similar to findings from other diary studies showing that people will complete several tasks at once, often shortly before a meeting with a research team [310].

> ***Lesson #8:*** We discourage having activities that build on one another where the sequence is important because participants complete activities out of order.

8.2.5 Relationships between Participants

Social vs. Activity Posts

Participants had mixed views on posts by other participants not directly related to research. We allowed social conversations outside of research activities because we felt that socially connected participants would be more comfortable being honest and sharing their activity responses with the

group. However, it was confusing to some participants when social threads were mixed in with specific research activities. Especially in the beginning of the study, this caused people to wonder if responses were "required" or not. One participant commented, *"...it's mostly been socializing at this point. I was going to chime in when it got more research-oriented."* Another requested, *"when you post a question in the group, can you do it as an admin or something? it is confusing ... and I don't want to miss a serious question."*

Most participants enjoyed this aspect of the study; participants rated the social aspect of the study higher than any other activity ($scale\ 1-7$, $median = 7$, $mode = 7$): *"It was good getting to know others in the group. I really liked that most were so open and friendly."* We note that the A11 survey would not have been completed by participants who Dropped Out; we suspect that the overwhelming amount of social activity by people of a different demographic may have been a factor contributing to their decision to leave the study.

A few participants expressed concerns about negativity and found it depressing to hear others' stories: *"... I also found a lot of negativity and some members being trapped and/or defined by their disease so that put me off."* Yet they saw value in having a place to express themselves: *"... if others are like me, they don't have unfettered ability to do that in our family."*

> The social support provided in ARC study groups is an opportunity we have to provide a real benefit to participants [124, 259]. This is especially valuable in this particular population, for whom social support is not otherwise readily available.

Lesson #9: We recommend providing opportunities for socialization between participants that are separate from formal study activities. Although this may not appeal to everyone, it can help screen for participants who are not interested in this aspect of the study and would likely become frustrated later if social conversations continue throughout the study.

Sub-Communities

Our modularity analysis showed that, aside from two participants who were Lurking or Dropped Out for the entire study, there were two sub-communities in this study; one with two rare diseases

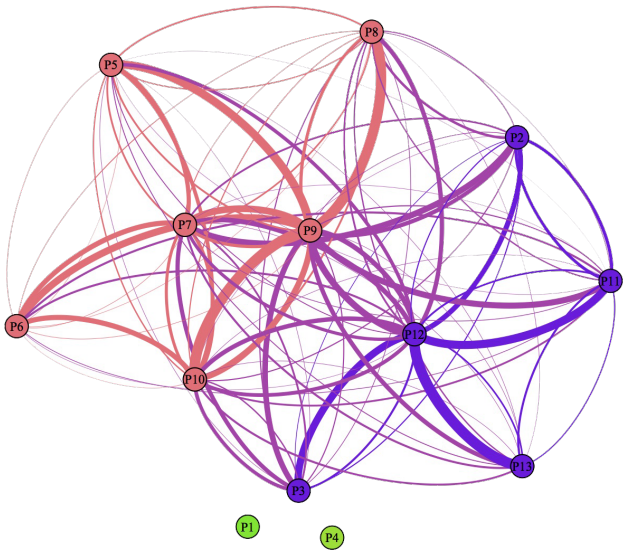


Figure 8.3: Participant sub-communities in the Facebook group. We observed two clusters; one with three diseases and one with two diseases.

and one with three rare diseases (Figure 8.3). Participants with the same conditions were recruited from the same Facebook groups, so it is likely they had an existing familiarity with each other before beginning the study. This was especially true in the case of Super Active participants who, we observed, tend to be prominent figures in their other Facebook groups as well. It is also likely that participants related to people with the same disease better.

We did not observe clusters by other demographics, however age and gender appeared to be factors in determining activity levels. Both men who enrolled in the study ultimately Dropped Out, and almost all of the women under 40 years old were Lurkers.

8.2.6 Researcher Perspective

Moderating

An online group differs from an in-person group in the weight and permanence of each post. In person, comments may be easily ignored if they are deemed irrelevant, uninteresting, or rude. Some others may simply not hear the comment in the first place. Online, each comment is given equal weight in the conversation. At first, this might indicate that a participant who is normally soft spoken and easily dismissed could contribute more to a conversation happening online than in person. In reality however, we found that conversations became dominated not by the weight of a

particular post but by the volume of posts from certain participants (the Super Active participants). It was easy to forget about Lurker participants. Several participants rarely contributed — one participant did not contribute the entire study, despite remaining in the group the whole time.

Inappropriate comments are easily written off in-person, but linger online. Removing a post or deleting a comment is a much more deliberate action than redirecting an in-person conversation. We deleted one post where a participant explicitly asked the group to donate money towards medical and living expenses. We removed the post and contacted the participant to politely explain our concerns. S/he seemed understanding, stating *"i understand.Life just sucks right now and I nee all the help I can get. My apologies."*, but dropped out of the group shortly after.

The majority of the posting in the group was done by one researcher to make things easier for participants and to build rapport, but there were three additional researchers in the group who could notice and quickly address a problem should one arise. We considered using a secondary Facebook account to maintain boundaries between research and social uses of Facebook, but ultimately chose to use our personal Facebook accounts partly to ensure that we were notified of new posts and could intervene quickly. We also wanted to engender trust with participants; we felt moderators should be portrayed as themselves [41] and not as a group account for the whole team, as if an occult hand were conducting the study. I had been using my personal Facebook account for well over a year to build rapport in rare disease groups.

Data Analysis

One of the benefits of conducting this study on Facebook was the amount of data available at the end of the study. The challenge was the overhead involved in compiling it all, especially as the data came from a number of different sources. We were not aware of any tools that met our needs for exporting all of the data we needed in a form useful for analysis. We resorted to a lot of manual gathering and coding; in a longer study or larger group it would be worth further investigating methods of doing this automatically. The time spent managing this data was substantial. In this case, the benefits of using a well-established social networking platform that participants were already familiar with outweighed the time required to collect this data.

Lesson #10: We recommend being mindful of the number of input mechanisms (e.g., Google Voice, email, survey platforms, etc.). Structuring activities to capture data from a range of different sources adds to the richness of the data but means data will be distributed in different locations and need to be collected and organized. More input mechanisms means more overhead.

8.2.7 Post-Study

At the end of the study, we left the Facebook group intact so participants could continue to connect with each other. We told them we would post updates about the research, but no further research would take place inside of the group. We informed participants that they could receive updates about the research by email if they opted to leave the group after the conclusion of the study. We continue to inform participants, as well as the Facebook groups we recruited from, about the status of our rare disease research. After the last official study activity, many participants remained in the group, although activity was limited and has decreased over time.

We provided participants with a draft copy of each significant new publication emerging from research they participated in to ensure they are comfortable with the content. This mitigates misinterpreting or misrepresenting comments and ensures participants have final say over how their data is used.

8.3 Extensions of ARC

Prabhakar et al. [275] adapted ARC for their research with pregnant and new mothers, applying many of the lessons from our initial study with people with rare diseases. Aside from varying in the population studied, they also ran their study for a shorter period of time (eight weeks as opposed to 22), with multiple (three) different Facebook groups, with a larger sample size overall ($n = 48$). The authors added several new activities, including a poll, several validated instruments, Ask Me Anything (participants asked and answered questions to the group), Advice Columnist (participants gave advice to a fictional character), and sharing one's Google search history with the goal of triangulating data. Maestre et al. [210] also adapted ARC for research with people living with HIV. Their eight week study additionally included activities typically found later in the design process,

including co-design of solutions and evaluating existing prototypes. They additionally focused very specifically on using different activities to triangulate their insights, exploring places where similar themes emerge from multiple activities or places where participants express conflicting ideas.

Prabhakar et al. [275] and Maestre et al. [210] each reflected on the existing lessons we provided as a result of our study with people with rare diseases. In some cases, these lessons led to successes in their studies and in other cases the authors identified additional considerations for these lessons. Both authors also introduce additional lessons from their own experiences using ARC. Table 8.2 summarizes the lessons from these three studies.

Our first recommendation (Lesson 1) was to build a strong rapport with groups used for recruitment. Neither Prabhakar et al. [275] or Maestre et al. [210] had existing relationships with recruitment groups; Maestre et al. [210] volunteered 50 hours at a local HIV support center to better understand the community before beginning recruitment for the study. Maestre et al. [210] also reported being successful in answering questions about their informed consent document via Facebook chat (Lesson 2), although they required physically signed and scanned documents (as in our study). Prabhakar et al. [275] allowed participants to consent via email. Future research is needed to continue investigating methods of obtain informed consent electronically.

Both sets of authors saw success from encouraging participants to engage with each other (Lesson 3) and in encouraging participants to share directly to the group (Lesson 4). They also provided options for participants who were uncomfortable posting to the group directly. Maestre et al. [210] note *"our participants had the option to send their submissions via e-mail."* Neither set of authors reported any major conflicts during the study (Lesson 5).

Prabhakar et al. [275] built on Lesson 6 by distributing a poll at the beginning of the study to assess preferred times for posting activities, although Maestre et al. [210] point out that this is not necessarily a helpful solution if participants have different opinions and live in many different time zones. Rather, they focus their efforts on adopting multiple strategies to ensuring activities were seen (Lesson 7). Prabhakar et al. [275] asked participants to "favorite" the study group to increase the likelihood of participants being notified or seeing posts from the group in their news feed and Maestre et al. [210] pinned the activity to the top of the group each week and set a "deadline" for each activity, after which the authors posted a reminder and sometimes sent private messages to participants to follow up.

#	**Lesson from MacLeod et al. [205] (1–11), Prabhakar et al. [275] (12–17), and Maestre et al. [210] (18–22)**
1	Building a strong rapport with members of groups used for recruiting before, during, and after the study.
2	Investigating alternative methods of consenting electronically.
3	Encouraging participants to provide feedback and build on each other's creative contributions.
4	Encouraging participants to post directly to the group, even submissions still in progress. Seeing other participant's contributions can give inspiration to people.
5	Exercising caution when planning the study and selecting activities; conducting research asynchronously and online means researchers do not have the same chance to assess how a method is going as it is happening.
6	Taking time to understand potential participant's Facebook behavior to inform decisions about when and how often to post and the overall study duration.
7	Researchers should be prepared to adopt multiple approaches to ensure activities are seen by participants.
8	Discouraging activities that build on one another where sequence is important.
9	Providing opportunities for socialization between participants that are separate from formal study activities.
10	Giving careful consideration to the makeup of the group when recruiting, targeting either a highly homogeneous population or targeting several homogeneous groups.
11	Being mindful of the number of input mechanisms. Each additional input mechanism introduces additional overhead.
12	Being mindful of the number of steps needed to complete an activity when selecting activities for the ARC method.
13	Consideration of participant's technology preferences, while developing activities.
14	Find balance between the ease of use and the usefulness of the data collection activities while engaging participants.
15	Adapt commonly used data capturing mechanisms used in FtF focus groups to use in online, asynchronous research methods.
16	Careful consideration of activity selection. When possible, we must provide meaningful and helpful interventions.
17	Researchers should be proactive about strategies to make data collection and organization easier.
18	Make support group administrators understand, approve, and be part of the study so that they can vouch for the intention, safety, and potential benefits of the study.
19	Post activities and reminders on the same days every week.
20	Researchers should use their personal accounts when studying stigmatized populations with the ARC method on a social media platform like Facebook. Protect participants' identities by warning them about confidentiality risks and setting boundaries in the sharing of information.
21	Learn about the type of language to use when communicating with the stigmatized before using ARC.
22	Be prepared to clarify miscommunication and answer questions as soon as possible to avoid unnecessary distractions and misunderstandings among participants.

Table 8.2: Lessons learned from three iterations of ARC.

Neither study included activities where sequence was crucial (Lesson 8), and both sets of authors similarly saw where participants would complete activities in bulk or out of order (see Stone et al.'s [310] concept of "parking lot compliance"). Instead, Prabhakar et al. [275] and Maestre et al. [210] both focus on triangulation, where their activities do not directly follow from one another but address similar themes from different angles.

Through multiple studies, we observed participants' interest in socializing with each other can vary greatly by population and by group composition (Lesson 9). Participants in Prabhakar et al.'s [275] study expressed very little interest in getting to know one another and primarily engaged in the specific study activities. Participants in Maestre et al.'s [210] study on the other hand were highly enthusiastic about meeting one another and requested that the group continue after the official end of the study so they could stay in touch. Further research could investigate means of creating community among participant such that people feel comfortable sharing. Researchers might also consider participants' interest in group connections at the beginning of the study.

Prabhakar et al. [275] broke their study into three smaller study groups that were more homogoenous by role (recent mothers, first-time pregnancies, and experienced pregnancies) and report being successful with this approach (although we note that there was little engagement outside of the formal study activities). Maestre et al. [210] report on a single study group of highly multicultural participants and saw success with this approach as well. Similarly, although we made this recommendation (Lesson 10) initially based on findings that participants in our study were drawn most often to participants with similar conditions and demographics, we also saw that participants expressed valuing the opportunity to get to know participants with other rare diseases because they had similar experiences.

> Homeogeony in group composition may be less important than finding common ground. Future research might consider helping participants find this common ground (such as using Facebook's Things in Common feature) to build stronger connections within a cohort.

Both studies relied on multiple sources of information and ways of submitting data (Lesson 11). Prabhakar et al. [275] reflect on the relative value of different activity types, considering the insight gained against the effort required, finding most activities to be generally worth the effort.

8.4 Discussion

8.4.1 Making Space to Reflect on Methods in HCI

HCI researchers regularly apply a range of methods to address different topics or questions [197]. When existing methods or practices are insufficient in some way or a new research opportunity is identified, they sometimes develop new methods or adapt existing methods for new contexts or settings [85]. Dickson and Stolterman [85] argue that new (design) methods are rarely developed in a user-centered way, nor are they tested or evaluated as such. In many cases these methods are used or tested only a small number of times [85, 286, 287, 314] before being *"set aside as a point of analysis, discussion, and a starting point for future research"* [85]. One possible reason for this is that new methods may emerge out of a need for a new approach to a specific problem rather than a deliberate decision to create a new method that is useful to other HCI researchers and practitioners. Moreover, seeing the broader relevance of a new approach to different research questions can be challenging if the new approach is contextualized and discussed in relation to a specific problem.

We argue there is value in engaging in a more substantive examination of a research method, as we have done in this chapter and in publications [206, 210, 275]. These extensions have revealed opportunities to revise or expand on lessons learned in our initial study with people with rare diseases. It reveals the fluidity with which the ARC method can be adapted to study different populations. We suggest, re-purposing an argument from Carroll and Rosson [45], that if we make our use of research methods more *"explicit[, then] we will better be able to manage and learn"* from each others' experiences and build a richer understanding of method development and use, which is important for conducting more effective inquiry.

Still, there is an outstanding question about the *validity* of new methods in HCI and qualitative research. These types of discussions (i.e., those about validity of qualitative research) are much more common in the social sciences where researchers have proposed ways of determining validity of research outcomes [71, 82, 218, 327]. Creswell and Miller [71] note that validity criteria might depend on *"the particular methodological design"* and *"the paradigmatic assumptions invoked by the research."* Taxonomies of validity consider things like the rigor and sincerity of the work [327], making a substantive contribution to our understanding of social and cultural life [82], having

potential for impact for its participants [82], and providing *"experience-near"* accounts that are fair to participants. It is thus important for qualitative researchers to articulate the grounds on which their validity claims rest.

These approaches to assessing validity are valuable for examining qualitative research outcomes. However, there are other strategies for improving validity of data throughout the research process. For example, triangulation involves using multiple sources of data to produce understanding; researchers often assume that this will result in some convergent meaning or "truth" [193] but this is increasingly controversial because *"it loses the context through which alternative meanings are derived, and [. . .] assumes some underlying reality to be converged upon."* Instead, many researchers consider triangulation to be valuable in eliciting divergent accounts to produce an understanding that is *"rich, robust, comprehensive, and well-developed"* [284]. There are four kinds of triangulation [83, 268]:

1. **Methods Triangulation:** considering findings from different data collection methods (typically a blend of qualitative and quantitative methods);
2. **Source Triangulation:** considering findings from different data sources within the same method (e.g., different points in time, public vs. private settings, etc.);
3. **Analyst Triangulation:** considering findings from multiple analysts reviews of the data to provide checks on selective perception and illuminate blind spots,; and
4. **Theory/perspective Triangulation:** considering multiple theoretical perspectives from which to review the data.

In particular, researchers use this triangulation to search for contradictions (consistency checking). Unlike in quantitative research, in qualitative research these outliers are critical to our understanding. They can tell us our initial interpretation or themes may not be correct or there may be some mechanism in between that helps to reconcile these differences [46].

8.4.2 Towards a Definition of ARC Research

Researchers in the HCI community sometimes describe themselves (in their publications) as having been "inspired by" a particular method or having borrowed parts of it without strictly adhering to the complete set of norms or rules of the method. In some cases, this is a way to signal to the

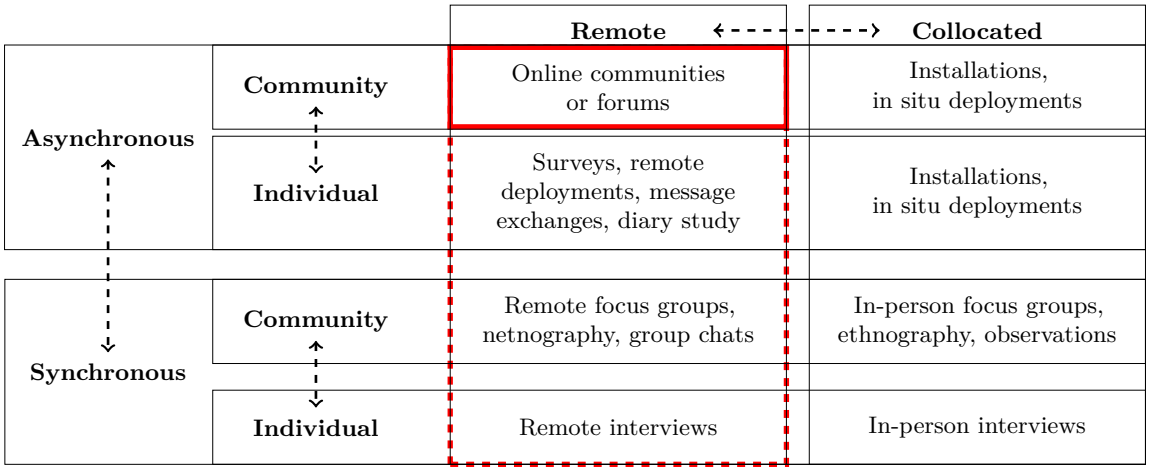


Figure 8.4: Examples of Research Methods in HCI. ARC requires, at minimum, an asynchronous, remote, community (solid red outline). ARC may also draw from synchronous or individual methods (dashed red outline).

audience that the authors should not be held accountable to those norms and rules or that a set of rules do not exist. This distancing from rules has the benefit of allowing for adaptations or tailoring of methods to new research questions, populations, or practical constraints. Thus, researchers can iterate on our approaches towards conducting better research overall. The evolving interpretation of rules also allows for conversations to take place around different applications or interpretations of methods; it may be that not every method has one single, agreed upon definition. The downfall here is that it is challenging to evaluate the validity or rigor of these adaptations; if the norms or rules of the original method do not apply, which norms or rules do? This flexibility can just as easily become an excuse if the method is poorly understood or executed.

With every iteration of ARC we, as a community, gain additional insight into best practices or strategies for effective application of this method. Part of what makes ARC an appealing option for researchers conducting studies with hard-to-reach populations is its flexibility. We have described ARC as being suitable for groups that have *"limited travel access,"* when *"social, political, or economic climates inhibit a potential participant's ability to assemble,"* when *"researchers may not have access to enough participants locally,"* or when "*participants may not feel comfortable in a physical study setting*" [206]. These kinds of potentially sensitive situations require great care in ensuring activities are appropriate for participants and the study is well designed. Flexibility in applying ARC in different ways can help support this, and not every lesson will be relevant to every population.

And yet, there is still value in considering what *actually* is ARC? Which parts of this method are flexible? At what point does it become a different method entirely? We begin most simply by considering the three dimensions in the method's title (asynchronous, remote, and communities) and their antonyms (synchronous, co-located, individual). Figure 8.4 provides examples of methods fitting the intersection of these categories. We constrain ourselves to only methods where a researcher is actively involved throughout the study and data is collected in response to some prompt or task given by the researcher. We exclude methods that analyze existing data (e.g., analyzing pre-existing social media posts). Realistically, many HCI research studies will move between these boundaries or use multiple methods (e.g., having participants take photos as an asynchronous, remote, individual activity, but meet with them in a co-located setting for a follow up interview).

From this framework we argue that, at a minimum, ARC requires an asynchronous remote community, but also has the potential to include elements of synchronous and/or individual research (outlined in red in Figure 8.4). For example, all three ARC studies made use of surveys as part of the study and participants completed these independently. Prabhakar et al. [275] also introduced a synchronous component when they conducted remote interviews with the participants in their ARC study. Although we are not aware of an ARC study that has made use of any synchronous, remote, community methods, it is not difficult to imagine a case where a researcher might include a synchronous, group-based activity. This framework contributes to a definition of ARC as a method or approach to conducting research. However, it does not prescribe:

- *Choice of platform.* ARC has historically been conducted in secret Facebook groups; an ARC study conducted on a different platform might look quite different from the studies conducted to date. Researchers might decide on a different platform for reasons of data ownership, privacy, or anonymity. There may also be features of these other platforms that appeal to researchers (perhaps features that simplify the analysis or allow for different kinds of activities to be included) or to participants (we conducted ARC on Facebook because people with rare diseases are active Facebook users.)

- *Activities and their frequency.* Researchers might choose different kinds of activities depending on which stage of the design process they are in, their population's access to different resources, the research questions they have in mind, etc. They may choose to incorporate elements of

individual and/or of synchronous research.

- *Study length and sample size.* These parameters depend on the goals of the study, activity type, and the population being studied. Ultimately, researchers might want to collect enough data to reach study goals and choose a sample that is representative of the population.

- *Participant groupings and group size.* Our analysis of study groupings originally suggested that participants were drawn to to others with similar conditions and demographics, but additional analysis (in Chapter 5 as well as in [210, 275]) suggested there was value in allowing participants to connect with a diverse community. Maestre et al.'s [210] was more international than the other two studies, but was still conducted entirely in English; future ARC studies might decide to make use of the automatic translation available on platforms like Facebook to further emphasize this type of diversity.

Of course, with the flexibility to make these choices comes a responsibility to do so in a way that is suitable for the participants or study population. A more public facing group might be appropriate for groups who strongly seek advocacy or transparency, but would not have been appropriate for Maestre et al.'s [210] work with a more stigmatized group. Using smart phones to take pictures to share with the group can lead to rich data within the group, but might not work well for people without access to that technology. Heterogeneous groups can lead to richer insights and help participants get to know someone they might not meet otherwise, but they could also lead to disagreements over differences.

> We strongly recommend that researchers spend time getting to know their study population before attempting an ARC study.

This might take the form of a pre-study using more traditional methods like interviews, observations. It might also involve working with a community partner or someone who is familiar with the study population. The bottom line is that researchers should be familiar with the population so they can make appropriate decisions about the elements on which ARC is flexible.

8.5 Summary

This is an early step in understanding how group-based research can be conducted using a common social platform, like Facebook. Significant additional research is necessary better understand the full potential and limitations of such an approach.

Our findings and reflections are influenced by our decision to conduct this study on Facebook. In a few activities, participants missed notifications or were not aware we had posted an activity. The mechanics of Facebook's algorithms are unclear, so we are not confident in our assessment of why this happens or how to prevent it. Additionally, "seen by" tracks whether someone acknowledged a notification but not necessarily that they actually read the post. We use counts of how many participants have "seen" a notification as an extra measure but acknowledge that this cannot be fully relied upon to determine whether someone actually read the post. The order of activities could also impact participation, especially as participation in longer studies tends to decline over time [115,167]. Using a different order of activities would illustrate how participants react to different types of activities, regardless of when they appear in a study.

We are also, so far, unaware of any studies comparing in-person and remote methodologies. It is possible that participants would have felt just as distressed when some of the activities made them think about the limitations of their social life in an in-person study, or it is possible that the remote nature gave them more time to reflect and think. Having laid the groundwork for how to conduct a study on Facebook, a future study could compare local and remote methods within a single population.

We hope that this method not only helps the HCI community conduct studies with hard-to-reach or atypical populations, but that it may also help balance out the benefits derived from such endeavors between researchers and participants.

“ *But let no person say what they would or would not do,*
since we are not judges for ourselves until circumstances call us to act. ”

—Abigail Adams, in a letter to John Adams, May 1784

Chapter 9

Small Data Privacy Protection & Ethical Rare Disease Research

Digital technologies and social networks have created opportunities for conducting research with populations that are otherwise hard to reach. This means beginning to address the dearth of research that has historically been a source of frustration for people with rare diseases [15,160,273,279]. With these new opportunities however come new risks and tradeoffs: What are the ethics of recruiting participants from various sources online? What does it mean to have truly *informed* consent when conducting research remotely? How can we protect participant identities when the rarity of their conditions makes them more easily identifiable? How do we balance people with rare diseases' desire for research and advocacy with their desire for privacy and dignity?

In this chapter, I answer none of these questions. Rather, I describe my experiences in conducting research for this dissertation, discussing the dilemmas we faced, decisions we made, and reflections on areas of future research. I see this chapter as contributing to an ongoing conversation on ethical research with sensitive populations, rather than a conclusive statement on best practices. This chapter contains material from several workshop papers from CHI 2015 [207] (with Kim Oakes, Kay Connelly, and Katie Siek), Pervasive Health 2016 [204] (with Maia Jacobs, Katie Siek, Kay Connelly, and Elizabeth D. Mynatt), and WISH 2017 [202] (with Jacob Abbott and Sameer Patil).

9.1 Ethics of Recruitment in Rare Disease Research

A key challenge with many human centered research projects is recruiting participants. Many of these challenges are amplified when working with people with rare diseases because they are not only challenging to recruit locally (i.e., they are geographically dispersed) but also because they are a more sensitive population requiring additional attention to issues of ethics and privacy. Existing medical informatics researchers have, where possible, turned to online sources to recruit people with rare diseases, suggesting that *"Patient-run, disease-specific communities were a major asset in recruiting. Partnering with these organizations at the outset of a study could facilitate improved access to participants"* [295]. Researchers report that social media is responsible for 84% [295] to 91% [60] of recruitment referrals in studies where multiple recruitment approaches were used.

In recruiting for the interview study described in Chapter 3, we found that most groups (including the moderators and group members) were amenable to having a researcher in the group and our recruitment efforts led to a far greater response rate than anticipated. Participants seemed interested in spreading awareness of their conditions, and were highly motivated to participate in research. Similarly, participants seemed to feel a responsibility to participate in research about their condition, since they are aware of how little research exists. Many actively sought to participate in clinical trials, partly to gain access to treatments that would otherwise be inaccessible to them, and partly to increase population sizes for existing clinical trials, thus ensuring more and better research available to future generations. Finally, we believe that many people with rare diseases were happy to have the chance to share their story with someone who would listen.

In the ARC study described in Chapters 5 and 8, our recruitment experience was more challenging. There was noticeably more discussion about credentials, IRB approval, study procedures, and benefits/risks of participation. Some group members were disappointed to learn that the research would not lead to outcomes directly used by physicians to improve the treatment of their health condition; they viewed our presence in the group as an attempt to profit without providing anything in return. HCI research can be viewed by group members as less of a priority than medical research or as taking too long to produce meaningful and directly applicable results.

It is important to consider best practices for working with these patient-run, disease-specific communities in a way that does not disrupt the communities or cause additional risk to community members.

Bruckman [38] suggests that when recruiting from online communities, *"it is important not to jump right in, but to 'lurk' quite a bit first. [...] People respond better to someone who has been part of the group (at least for a little while) and seems to understand the community."* Bruckman [38] also acknowledges that *"It is easy to say, 'if you're studying a talent site, start singing'."* This was a key challenge for our research– it would be absurd to suggest "if you're studying rare diseases, acquire a rare disease." Thus, to recruit via these online communities researchers end up participating in groups where they do not belong. Some groups have guidelines specifically banning outsiders in their communities; many groups are intended *only* for people who actually have the rare diseases. Even groups where it is not *required* that members have a rare disease, there is an *assumption* that this is primarily the case. Group members may not be aware of the presence of researchers in the group and may behave make assumptions about the privacy of their information that may not be true. Even though we did not collect or analyze this data directly, it was still visible to us and the group members might not have realized this. Future designs of online communities might better support this kind of research by making insights about the membership of a group more readily available or visible to group members, such as through visualizations of the types of people present in the group or notifications to members about new additions. Researchers might also consider ways of making themselves visible or known to group members up front, for example through an introductory post to the group clearly stating their goals of participation and allowing group members to ask questions or express concerns. Bruckman [38] further explains, *"I generally ask my students not to study sites with vulnerable populations unless it is necessary [...] It is advisable, if possible, to stick with something less sensitive where there are lower consequences to disturbing site activity accidentally."*

Bruckman's [38] recommendation to avoid studying vulnerable populations is at odds with research suggesting a need for rare disease research and that people with rare diseases are enthusiastic about contributing to this research, so long as it is is well aligned with their goals (see Chapter 1).

Source	Advertisement	CPR
Chapter 6	Facebook, Twitter, Google	$5.23
Whitaker et al. [347]	Facebook	$14.41
	Television	$1,094.27
	Radio	$635.92
	Print	$811.99
	Email	$37.77

Table 9.1: Cost per Respondent (CPR) for the study discussed in Chapter 6 compared to the median CPR for recruitment approaches from Whitaker et al.'s [347] review.

Source	CTR	CPC
Facebook	1.76%	$0.04
Instagram	2.93%	$0.12
Messenger	1.37%	$0.04
Audience Network	2.82%	$0.03
Twitter	0.81%	$0.48
Google AdWords	8.47%	$0.10
Google Search Partners	0.49%	$0.09
Google Display Network	1.55%	$0.04

Table 9.2: Results (Click through Rate (CTR) and Cost per Click (CPC)) of recruiting via advertisements on popular social platforms

This is similar to experiences described by other rare disease researchers who have made comments to the effect of, *"In our experience, patient group moderators, charities and medical departments are extremely happy to support such work provided it has a sound academic rationale, has been suitably ethically-reviewed and may potentially benefit patients."* [79] Future research might consider ways of engaging more deeply with people with rare diseases as true collaborators throughout every stage of the research process [332] to ensure there are true benefits for people with rare diseases.

To mitigate some of the challenges of recruiting through online communities while still leveraging the success of social media recruitment approaches, we investigated the possibility of targeted, paid online advertisements for the final study of this dissertation (Chapter 6). This approach was a success: we were able to recruit the necessary number of participants for our research and the cost of this recruitment approach was on par with (or even more affordable than) previous research (Table 9.1). We did, however, have varying experiences on different platforms (Table 9.2).

On Facebook (including the main Facebook platform as well as Instagram, Messenger, and an external audience network), we found it difficult to target people with rare diseases with advertisements. Advertisers can target users who are "similar to" followers of certain Facebook pages. We were able to target audiences similar to followers of larger organizations like the National Organization of Rare Diseases (NORD) or Rare Disease Day, but were not able to target audiences of smaller organizations like the Canadian Organization for Rare Disorders (CORD) or the European Organization for Rare Diseases (EURORDIS). We also were not able to target audiences of specific rare disease awareness pages or support groups. Thus, many of our keywords

for targeting advertisements on Facebook focus on health and illness as a whole (e.g., Neuroscience, Immune system, Human musculoskeletal system), rather than rare diseases. We we were also able to target pages like The Mighty, a blog about chronic illness experiences. Although, we did not investigate the recruitment source for specific respondents, one might expect that our Facebook advertisements led to a mix of common chronic illness and rare disease respondents, with a skew towards the more common examples; future research might specifically investigate this hypothesis.

Twitter and Google allowed for more granular targeting of less popular hashtags, search terms, and keywords. On Twitter, we targeted users based on hashtags of rare diseases in general (e.g., #raredisease, #orphandrugs, #nord), hashtags of specific rare diseases (e.g., #adrenoleukodystrophy, #hereditaryspasticparaplegia, #idiopathicgastroparesis), or for chronic illness communities more broadly (e.g., #spoonieproblems, #lifewithachronicillness, #chronicpain). On Google AdWords, we used a similar set of keywords to Twitter (but without the hashtags). Here, we were better able to specifically target rare diseases rather than a general interest in chronic illness.

Advertising tools like Twitter and Google are effective solutions for recruiting participants without needing to join disease-specific groups. However, there is also a cost here; not joining these groups means missing out on opportunities to observe the norms of these communities. Bruckman [38] similarly argues for the value of observation, stating *"You will not appreciate the nature of the site until you have spent a non trivial amount of time there."* Unerti et al. [332] similarly argue that there are a number of benefits to working closely with a community, such as in a CBPR approach, including *"more relevant research, wider impact, better fit between interventions and target beneficiaries, more effective recruitment and retention of diverse populations, improved internal validity, more rapid translation of research into action, and development of people."*

> Future research might consider what CBPR could mean when the "community" is a virtual community. Recruiting via advertisements was effective, but there is additional value to be gained by understanding how to work with these communities in ways that are mutually beneficial.

Additionally, there is a concern with social media recruitment (discussed as a limitation in Part II) that the resulting participants may not be representative of the greater population. Whitaker et al.'s [347] systematic review of the use of Facebook as a recruiting technique for health research

found that young, white, educated, women tend to be over represented in these studies (compared to national data). This is true of the studies in this dissertation as well, however, further investigation shows that over-representation of these same demographics is also present in traditional recruitment techniques [36, 130, 306, 361]. That is, unrepresentative samples are a problem not limited to social media recruitment. Future research could consider ways of supporting researchers in more representative or purposeful sampling.

9.2 The Ethics of Masking & Naming in Patient Narratives

Qualitative researchers generally adhere to a "convention of confidentiality" [13] to prevent distress, humiliation, harm, or other negative consequences for participants [13, 97]. It is common practice in qualitative research to use quotes from participants and include tables summarizing participant demographics. Although it is unusual to see direct identifiers (e.g., real names) included in qualitative publications, contextual identifiers may still make it possible to identify participants.

Deductive disclosure occurs when characteristics of study participants make them potentially identifiable in research publications or presentations [170, 324]. Sieber et al. [301] use research about teachers as an example, suggesting that if the researcher names the school district where the research took place, a person with knowledge of that school district may be able to identify the participants based on traits like age, gender, years with the school district, etc. This deductive disclosure was a concern for us throughout this research, and we generally provide less demographic information about participants than a reader might like to have for the sake of protecting participant identities and minimizing deductive disclosure risks.

Researchers sometimes have to make uncomfortable tradeoffs between an absolute commitment to privacy and a responsibility for making respondents' lessons known [129, 170, 344]. We need to take responsibility for determining which parts of a participant's contributions present a privacy concern [**?**, **?**, 324] and account for this by changing details in quotations [170], altering non-essential details to make the participant unrecognizable [344], or creating new personas that are composites of several participants without directly representing any real individual. However, participants have been known to strongly object to having their words or characteristics altered in reports [67]. These approaches also risk rendering the data useless for answering the research questions or distorting the

original meaning of the data [**?**]. It can also make it difficult for reviewers and readers to understand or asses the validity of the work [**?**]. Bruckman [37] explains:

> "This disguising also erects barriers to the social practice of science. In an open scientific community, individuals ideally publish results sufficiently detailed for others to attempt to duplicate those results and affirm or question the findings. This idealized model from the physical sciences is always hard to replicate in social sciences, but even harder when the act of protecting subjects adds substantial new barriers to follow-up inquiry by others."

Jerolmack (in a conversation with Murphy [243]) argues technologies like Google and social media have made it even easier to identify people by performing keyword searches, and the practice of using pseudonyms offers *"at best, an illusory promise to protect confidentiality"* [243]. He suggests there may actually be value in providing participants the choice of *not* being de-identified:

> "There was very little that I could offer my participants for all the time they gave me, but they viewed seeing their name in print as intrinsically rewarding. When I handed out copies of [my book], most quickly thumbed the pages looking for their name and some excitedly took photos of the printed pages they appeared on and texted them to friends and family. This has convinced me that, at least some of the time, naming may be more ethical than masking."

Bruckman [37] makes a similar argument about content creators online, discussing the tradeoff between giving *"the right of Internet users to receive credit for their creative and intellectual work"* and *"the need to protect vulnerable human subjects in research studies."* In our work with rare diseases, we rarely run into issues of content creation and attribution of intellectual property, but there is still a tradeoff here between participants' goals of advocacy and increased research and their goals of privacy and dignity. We also found that participants eagerly and excitedly read through our draft paper submissions, identifying their own quotes and sharing the papers with their network. In some cases, even people online who were not actually participants in the research in question left comments remarking that they saw elements of their own experiences and stories in the work, almost as though they *had* participated in the research.

One possible approach for future research might be to allow participants to choose their own alias or pseudonym (e.g., [103]). This approach engages participants directly in a conversation about how they are represented and empowers them to communicate something about themselves through

a name, while still providing some level of anonymity. It also makes it easy for them to identify themselves to in publications and disclose this to others if they choose.

Guillemin et al. [135] argue that procedural ethics (required ethics procedures within organizations) are helpful for encouraging researchers to think about ethical issues but are largely a formality that do not appropriately address the specific ethical dilemmas arising in qualitative research. A standard part of procedural ethics is some confidentiality agreement at the beginning of the study; this is crucial to acquiring informed consent and building rapport with participants [73]. However, these discussions take place before the participant has actually disclosed any information, and confidentiality is rarely an ongoing conversation [170].

Kaiser [170] suggest dealing with deductive disclosure risks by *"making respondents better informed of the use of data"* and encourages *"discussing the specifics of audience and confidentiality after data collection,"* noting that many participants are driven by a desire to help others [20, 48, 93, 162]. The author states that, *"discussing the use of their data can help them to grasp the outcomes of their participation"* [170]. Member checking [193] (providing the interpretation and report to the informants to check the authenticity of the work) is one approach that can address this deductive disclosure risk, as well as enhance validity of research by allowing participants to point out inaccuracies [219]. Others have pointed out concerns with this approach, warning that participants may not like the way they are portrayed [67, 97, 185], and researchers might write differently from the start because they know their work will be seen by their informants [156].

> In this dissertation, we conducted member checks on our qualitative research, but still leaned heavily towards masking rather than naming. Future research might reflect on procedures for engaging in ongoing discussions about confidentiality and data use and ways of ensuring research is useful and empowering to participants (e.g., for advocacy initiatives).

9.3 Reidentification Risk in Rare Disease Survey Research

Especially in the case of rare disease data, making anonymized data publicly available can be extremely beneficial and lead to further advancements in research that might not be possible otherwise.

> Rare disease data is not easily anonymized; simply removing directly identifying information is insufficient because only a small number of attributes are needed to re-identify an individual from such a data set, particularly in conjunction with the name of the (rare) disease.

For example, ALS is a disease that, although widely known because of the recent ALS ice bucket challenge [179, 308, 333, 351], impacts less than 0.01% of Americans. Using only gender, age, and country of an individual, we can narrow down to close to approximately 1,000 people in the US. For people living in a smaller country, we could narrow down even further to as few as 10-15 people.

There are numerous examples of health data having been re-identified by linking "quasi-identifiers" (i.e., features that are not directly identifying, but can be combined with other quasi-identifiers to create or uncover a unique identifier) to other public data sets (e.g. [96, 315, 359]). One example was in work by Sweeney et al. [315]; the authors were able to link a database of health insurance claims (with direct identifiers removed) to a voter registration list, using quasi-identifiers (date of birth, zip code, and gender) to re-identify individuals listed in both databases. Similarly, Malin and Sweeney [212] were able to re-identify people based on genomic datasets using disease genes. Solomon et al. [307] demonstrated the likelihood of re-identification of behavioral/social science health data given certain kinds of attacks [244], finding that almost any attribute could be used to make a participant unique, either by itself or combined with at least one other attribute.

To combat these kinds of privacy threats, Sweeney introduced k-anonymity [315]. A dataset can be said to have k-anonymity protection if *"the information for each person contained in the release cannot be distinguished from at least k-1 individuals whose information also appears in the release"* [315]. The goal is to be able to release a privacy preserving version of a dataset *"with scientific guarantees that the individuals who are the subjects of the data cannot be re-identified while the data remain practically useful"* [315]. Typically, k-anonymity is achieved by *suppressing*

(replacing values of the attribute with an asterisk) or *generalizing* (replacing the attribute with a broader category). Although optimal k-anonymity is considered to be NP-hard [228], a range of heuristic methods (e.g. [19]) and tools (e.g., [276]) exist that can yield effective results. In this section, I explore k-anonymity as a potential approach to anonymizing rare disease data.

9.3.1 Method

To analyze the tradeoff between privacy and utility in sharing rare disease data, we rely on our survey data from Chapter 4. This data consists of information about self-reported online behavior and perception of health care providers by people with rare diseases and common chronic illnesses. From this data set, we consider age, gender, country of residence, martial status, employment status, education, and disease name to be quasi-identifiers, as these have been used for re-identification in previous research [315]. In Chapter 4, our goal was to predict the disease type of a given individual (i.e., whether someone has a rare disease or a common chronic illness). We made decisions about how the data should be discretized including removing disease names and relying solely on the labels "one or more rare diseases," or "one or more common chronic illnesses." We similarly made decisions about how best to discretize features such as age, years of experiencing symptoms, and years since diagnosis.

All these decisions were made based on the specific use case and goals of the research. This use case was motivated by the research questions in this dissertation not necessarily representative of the ways *other* researchers might use this data. To investigate alternative use cases, we asked a small panel of machine learning researchers ($n = 5$) to suggest possible use cases or prediction tasks based on a copy of the survey and the raw response data. All of these researchers were internal to our institution and added to our IRB as non-interacting personnel so that they could access this data (the data does not contain any direct identifiers, but does contain quasi-identifiers). The researchers were given freedom to select features, resample, and/or discretize the data as they saw fit to complete their proposed task.

We then used ARX [276] (an open source tool for anonymizing data) to generate a k-anonymized version of this original data set. We begin with $k = 2$ (i.e., an individual in the data set cannot be distinguished from at least one other individual in the data set) to assess the changes in utility when the most basic protection is applied.

[That is, if we apply even a minimal amount of additional protection to the data, can it still be used for rare disease research?]

The result of this 2-anonymization is a data set in which age, country, and martial status are suppressed entirely. Disease names are generalized and replaced with a designation of whether the person has a common chronic illness, a rare disease, or both. All other quasi-identifiers (gender, employment status, education) remain unchanged.

We use this anonymized data set to replicate our analysis from Chapter 4 (predicting disease type). We also asked the panel of machine learning researchers to replicate their analysis. Because the anonymization process involves suppressing and generalizing variables, the researchers sometimes had to use different pre-processing approaches from their initial approach. The prediction task, classifiers, and evaluation metric all remained consistent between the initial analysis (on the sensitive data) and the final analysis (on the anonymized data).

For each of these use cases, we compared the performance of the original data set against the anonymized version. The difference in utility relative to the performance of the original data is calculated as:

$$\Delta = \frac{Protected - Original}{Original} * 100$$

9.3.2 Results

Our experiments focus on two key questions:

Q1 How much utility is lost by using an anonymized version of the data to predict disease?

Q2 How much utility is lost for use cases suggested by other researchers?

Replication (Predicting Disease Type)

To answer **Q1**, we compare the results of predicting disease type from the anonymized version of this data against our results from Chapter 4; we use the same classifiers and evaluation metrics as in Chapter 4. Table 9.3 shows the results of replicating the analysis with the original data set and with the anonymized version. We saw previously that Soft-FGB achieves the best results for all three metrics. Here we observe a noticeable (27.16%) decrease in the true positive rate between the

	True Positive Rate			F_3			F_5		
	Orig.	**Prot.**	Δ	**Orig.**	**Prot.**	Δ	**Orig.**	**Prot.**	Δ
NB	0.507	0.524	3.35%	0.507	0.528	4.14%	0.507	0.525	3.55%
LR	0.413	0.456	10.41%	0.400	0.448	12.00%	0.402	0.453	12.69%
5-NN	0.288	0.262	-9.03%	0.299	0.279	-6.69%	0.293	0.268	-8.53%
DT	0.408	0.262	-35.78%	0.412	0.271	-34.22%	0.410	0.265	-35.37%
Oversample	0.511	0.454	-11.15%	0.504	0.471	-6.55%	0.508	0.460	-9.45%
Undersample	0.657	0.553	-15.83%	0.632	0.566	-10.44%	0.647	0.558	-13.76%
SMOTE	0.472	0.360	-23.73%	0.471	0.382	-18.90%	0.472	0.368	-22.03%
Soft-FGB	0.972	0.708	-27.16%	0.825	0.795	-3.64%	0.909	0.793	-12.76%

Table 9.3: Results of replicating Chapter 4's analysis using the original data (Orig.) and protected data (Prot.). We report on the relative difference (Δ) between these two versions of the data for three evaluation metrics: True positive rate and two weighted F measures (F_3 and F_5).

original and anonymized versions of the data set. However, the reduced true positive rate on the anonymized data (0.708) is still better than the performance of the other classifiers on the original data (0.262–0.657) (see Table 9.3). Importantly, the decrease in performance is the smallest when we examine the F_3 score (3.64%), suggesting that the anonymization hurts recall more than precision. This is problematic as it was recall that we were aiming to maximize in our goal of being able to identify the rare disease examples (more so than the common chronic illness ones); the utility of the anonymized data depends, at least in part, on the evaluation metric.

Researcher-Generated Use Cases

To answer **Q2**, we asked five machine learning researchers to suggest use cases or tasks suitable for the data. Each researcher was responsible for decisions around pre-processing, classifier choice, and evaluation metrics. We compared the performance of the original (sensitive) data against the anonymized data for each use case suggested by the researchers.

Two researchers aimed to predict **disease type** (rare or common disease), taking approaches that were different from our earlier approach as well as different from each other. The first researcher used a random forest classifier with 5-fold cross validation, comparing the original data to the anonymized data. They further aimed to make this prediction with the demographic information removed, using the original data set and the anonymized data set. The second researcher used a Linear Support Vector Machine (LSVM) and nested-cross-validation. As in our work in Chapter 4, this researcher chose to remove disease name from the original data set stating, *"that felt like cheating."* For

Task	Classifier	Orig.	Anon.	Δ
Disease type	Random Forrest	0.828	0.738	-10.87%
	Linear Support Vector Machine	*0.520	*0.519	-0.19%
Severity of symptoms	Naïve Bayes	0.359	0.359	0.00%
	2-Nearest Neighbors	0.308	0.306	-0.65%
	5-Nearest Neighbors	0.303	0.324	6.93%
	15-Nearest Neighbors	0.326	0.306	-6.13%
	Random Forrest	0.332	0.292	-12.05%
Social media use	Random Forrest	0.511	0.474	-7.24%
Helpful support sources	Decision Trees	0.521	0.445	-14.59%
	Naïve Bayes	0.503	0.533	5.96%
	Logistic Regression	0.545	0.447	-17.98%
Number of specialists	Random Forrest	0.471	0.474	0.64%
Sharing information	Naïve Bayes	0.628	0.632	0.64%
	2-Nearest Neighbors	0.620	0.620	0.00%
	5-Nearest Neighbors	0.607	0.598	-1.48%
	15-Nearest Neighbors	0.602	0.639	6.15%
Disease name	Bayesian Network	0.377	0.00	-100.00%
	Bagging	0.375	0.00	-100.00%
	Boosting	0.359	0.00	-100.00%

Table 9.4: Results of using original (sensitive) and k-anonymized data for researcher-generated prediction tasks. Asterisks indicate AUC-PR; all other researchers report accuracy.

evaluation metrics, this researcher chose Area Under Precision-Recall Curve (AUC-PR), averaged over 30 trials. The difference in performance between the original and anonymized data for the LSVM approach is virtually nonexistent. Comparing this finding against the results of the random forest approach and our baseline analysis (Table 9.3) suggests that a LSVM approach to this task is more robust to the loss of data. The utility of anonymized data may depend on the choice of classifier.

Two researchers aimed to be able to predict the self-reported **severity of symptoms.** In particular, one researcher wanted to understand the importance of the disease type in making this prediction. This researcher first used the original and anonymized data sets with all variables included, then re-ran the analyses on the anonymized data set with additional variables removed (all demographic information including disease type removed in the first iteration and all demographic information except disease type removed in the second iteration). This researcher chose to use Naïve Bayes and k-Nearest Neighbors ($k = 2, 5, 15$). Based on these results, the first researcher concluded

that disease type is an important attribute to predict quality of life when using an algorithm like Naïve Bayes. The other researcher aimed to make the prediction without demographic information (i.e., based purely on disease information, technology use, and perception of health care providers). This researcher used a random forest approach with 5-fold cross validation. Both researchers chose accuracy as the evaluation metric. In this task, the many of the changes in performance are less noticeable or even non-existent– most of these differences could be easily attributed to expected statistical deviations. Using the anonymized data would likely be acceptable for this task.

Other use cases proposed by the researches including predicting **use of social media** as a source of health information, the most helpful **sources of support**, the **number of specialists** seen by a respondent, and whether or not the respondent **shares information** about medical treatments online. There is variation in the change in performance depending on which classifier was chosen, but for each of these use cases there is at least one classifier where the change in utility of the data is negligible. This suggests that these tasks could be accomplished by researchers who only have access to the anonymized data.

One researcher had intended to predict the **disease name** of the individuals in the data set. For individuals with multiple chronic conditions, the researcher created duplicate entries such that each example has exactly one disease. They included only those cases where there were at least two instances of the disease in the data set (removing conditions that occurred only once). The researcher used Bayesian Networks, bagging, and boosting to make this classification initially. Unsurprisingly, it is not possible to complete this task with the anonymized data, given that disease name is one of the suppressed variables in the anonymized data.

9.3.3 Discussion

Certain tasks (e.g., understanding the impact of disease type on quality of life and predicting online sharing behavior) do not demonstrate much loss in utility. Researchers could likely complete these tasks using the anonymized data instead of the sensitive data, preserving the privacy of people with rare diseases. Other tasks (e.g., predicting disease type or most helpful sources of support) are more challenging and depend on the classifier/evaluation metrics used and how influential the protected features are in making the classifications. In these cases, having the original data would be preferable for improving results but this introduces potential risk to the people in the data set. What

is not clear is whether the protected data is sufficient to draw *some* conclusions. That is, if the only choices were using the protected data or not conducting the analysis at all, is the performance of the protected data still high enough that the analysis is worth doing? This is an interesting question for future work; extensions of this work might engage researchers with specialized knowledge of the domain (rare disease) rather than specialized knowledge of analysis.

Finally, tasks like diagnosing one's disease are impossible to complete without the original data because the target of the prediction (disease name) is one of the variables that is entirely suppressed in the anonymized version (the only remaining information is whether the disease in question is common or rare). If researchers would like to undertake this kind of prediction task, they would need to enter into a special agreement with those responsible for the data. An alternative for future research may be to explore different taxonomies or ways of clustering diseases such that instead of two categories of disease (rare vs. common) there could be many categories.

Our goal with this work was to explore the possibility of being able to release a single protected version of a rare disease data set that meets some *minimal* privacy preserving criteria (in this case, 2-anonymity such that individuals in the data set cannot be distinguished from at least one other individual in the data). Our findings have shown that this 2-anonymity protected data set is not universally useful; higher values of k would only lead to greater generalization and suppression of data, thus reducing the performance even further. It may be possible to maintain utility at higher values of k in a larger data set, but rare disease data sets, by definition, tend to be small.

> Designing tools for privacy-preserving data sharing that take into account community input will be critical to enabling additional research, while protecting the privacy of research subjects.

In most cases, researchers collect data, perform some pre-processing, then make it available in some repository for others to use. This might be appropriate in cases of non-human subjects data or low risk contexts, but it may not be appropriate in the case of behavioral health data (especially rare diseases). It may be possible to create anonymized versions that are better tailored to specific use cases, but we are hesitant to release multiple differently protected versions of the same data set in case of attacks. It is valuable to consider as many use cases as possible prior to

anonymizing and releasing the data. Tools for privacy preserving data sharing should consider community input on the uses of data. One option here is to release a description of the data or a list of the variables/features without releasing the actual data itself. Community members could submit use cases or questions they were interested in answering from the data. This was the case in Wagner et al.'s [339] Device Analyzer project, where the authors solicited input from researchers about *"questions that the research community would like to answer using our dataset."*

It might not be possible to *completely* assess the usefulness or information gain from each specific feature without actually having access to the data, but if the researchers know as much as possible about the use cases they can better generate a single anonymized version that takes these tasks into consideration. In our research, there were several use cases where the change in performance is negligible or tolerable; understanding the potential use cases allows us to assess the need for sharing the sensitive data via special agreement or merely providing a protected version.

9.4 Summary

In this dissertation, we have endeavoured to work with people with rare diseases as ethically as possible, working closely with our IRB and relying on documented best practices where available. In this chapter, we explored some outstanding questions about recruiting people with rare diseases, reporting on results of research, and sharing data to facilitate additional research. Our hope is that researchers will continue to reflect on these challenges in the future, towards creating effective tools and practices that balance risks and benefits in remote research with sensitive populations.

“ *I am one of those irritating people,*
who hang on to the door-knob after they say good-bye,
and will neither come back nor go,
always remembering something else which must be said... ”

—Nellie L. McClung in *The Stream Runs Fast*

Conclusion

In this dissertation, I make two types of contributions [357]: I provide an empirical contribution to understanding people with rare diseases and I provide a methodological contribution to conducting research with unique populations.

9.5 Understanding People with Rare Diseases

The primary contribution of the research in this dissertation is empirical research that tells us about people [357]. Specifically, this research provides a profile of people with rare diseases.

> This dissertation provides researchers and practitioners with an in-depth understanding of the experiences of people with rare diseases and of how these experiences are similar to or different from the experiences of people with common chronic illnesses that have been the focus of existing literature in HCI and other sociotechnical research domains. This understanding of these experiences can inform the design and development of novel sociotechnical systems to support and empower people with rare diseases.

We identified many social experiences in common across a range of different rare diseases regardless of the specific symptoms. We learned that having a rare disease necessitates wearing many different hats; people with rare diseases have to become researchers, advocates, lobbyists, fundraisers, record keepers, and peer supporters—all in addition to their roles as friends, parents, spouses, students,

Rare Disease	**Common Chronic Illness**
Informational Support	
Equally likely to look online for health information.	
More likely to use a social network to look for health information.	
	More likely to use mobile applications and search for health information on a mobile device.
More likely to subscribe to updates about health or medical topics.	
Equally likely to watch videos online about health or medical issues.	
Equally likely to track health symptoms or health indicators.	
	More likely to track wellness metrics (weight, diet, exercise, etc.)
More likely to need legal and financial advice.	
More likely to offer medical advice.	
More likely to be involved with advocacy and awareness raising initiatives.	
Nurturant Support	
Equally likely to read about someone else's health experiences and go online to find others with similar health conditions.	
More likely to share health information online, especially on social media.	
More likely to use mainstream social networks to build community (starting and joining groups, following peers' health updates, memorializing others)	
Tangible Support	
Equally likely to require help with many tangible support tasks (driving, laundry, lifting heavy objects, cleaning, cooking, and computing.)	
More likely to offer help with with laundry.	
More likely to require help running errands.	
More likely to engage in fund-raising activities online.	

Table 9.5: Similarities and differences in social support between people with rare diseases and people with common chronic illnesses.

working professionals, etc.— simply to manage their condition. The intensity of this experience is challenging to grasp; being diagnosed with a condition very few people have heard of, let alone understand, permeates many aspects of one's life. The additional work of advocating for one's self with clinicians, keeping up with research on one's condition, managing information from many different specialists, fundraising or lobbying for better access to medications, and educating other newly diagnosed patients quickly becomes a full time job and it is understandable this could become an overwhelming force shaping one's identity and sense of self, as we saw in our research [203, 208]. **Findings in Chapter 3 contribute to the community by providing a profile of the social experiences and support needs of people with rare diseases.**

Many participants in this dissertation reported that this Rare World experience negatively impacted their relationship with their friends and family members; future research from the perspective of this nuclear caregiving network is imperative. It is possible that, because these friends and family members are not going through the same experience, it is not as much a piece of their identity. They may face awareness fatigue from confronting these diseases so often; they may be overburdened and fatigued as caregivers. They may be in disbelief or denial. They may be unaware that their comments are unhelpful or underestimate the support needs to begin with.

The experiences of people with rare diseases are unique from those of people with common chronic illnesses (summarized Table 9.5). **This dissertation contributes a discussion of similarities and differences between rare and common chronic illnesses.** In the absence of adequate support from friends and family, people with rare diseases turn to online communities, typically disease-specific support groups within larger social media platforms such as Facebook – more so than people with common chronic illnesses. They use these communities to trade information about their disease, to support each other emotionally, and to feel a part of a community. **In understanding these behaviors, we contribute an approach to to identifying people with rare diseases from self-reported online behavior.**

The current design of these online communities, however, does not adequately support more tangible forms of support, such as running errands, doing housework, or performing strenuous physical tasks. It is more challenging for people with rare diseases to support each other with physical, in-person tasks if there are few people nearby with the same disease or if everyone needs the same help because they have similar symptomatic limitations.

	Information Disclosure	Desired Outcome
General Public	*Health:* General information about the condition. *Whole Person:* Basic information (including demographics, interests, and expertise).	*Informational Support:* General awareness about and interest in the condition, towards additional research and better access to treatment.
Friends & Family	*Health:* Information about the condition and a limited amount of information about the personal experience with it. *Whole Person:* Personal information about the individual (including not only demographics, interests, and expertise, but also beliefs and lifestyle information).	*Nurtuant support:* Empathy and validation. Engagement with non-health topics and activities, but patience and forgiveness when disease identity is overwhelming. *Tangible support:* Minimal tangible support, but heavily subject to barriers of guilt over unrepayable favours.
Rare World	*Health:* Information about the specific condition and a limited amount of information about the personal experience with it. *Whole Person:* Contextually relevant information about the individual (demographics and expertise).	*Tangible support:* Exchanged by matching people with things in common but complementary expertise. *Social support:* Discussions of the shared social experiences of living with a rare disease, broadening perspective.
Disease-Specific Communities	*Health:* Extensive information about the personal experience with the condition. *Whole Person:* Contextually relevant information about the individual (demographics and expertise).	*Informational support:* Patient expertise and guidance towards new research. *Nurtuant support:* Discussions of health experiences and a sense of community from being part of a group of similar others.

Table 9.6: Information disclosure preferences and desired outcome for different audiences.

This research contributes a proposal for the design of offer-driven helping networks to facilitate exchanges of social support among people with different rare diseases, using mainstream social media platforms. A person with a rare disease would offer help to a person with a different rare disease in exchange for credit they could use to cash in on an offer presented to them in the future. **We provide an exploration of the application of social matching techniques to understand how people choose helpers online and to connect people with rare diseases via existing social networks.** We consider the privacy implications of sharing rare disease data online, finding that that people with rare diseases were more willing to share information about their health with online health communities or private groups of similar others than they were willing to share this with broader audiences (Table 9.6). Information about interests, demographics, beliefs, and lifestyle was more appropriate for audiences of Facebook friends.

9.5.1 Future Work

In this research, I discussed participants' own perspectives on their conditions, lives, and relationships. I proposed an approach to facilitating support exchange among people with different rare diseases to empower them to help and support each other. This is one of many different possibilities for addressing the needs of people with rare diseases. Future work might work with friends and family members of people with rare diseases to understand their perspectives on these same relationships. What barriers to support exist from the perspective of the friends and family members? What successes or existing strategies might we leverage to provide better support?

Other populations of future study might include people who are undiagnosed. In this dissertation, I have drawn comparisons between rare and common chronic illness populations, but all these participants have *known what condition they have.* A rare disease diagnose requires a significant amount of confidence advocating for oneself with medical professionals, resources travel to specialists and experts, and literacy to conduct research about the condition– what is the rare world experience like for people who are not able to get diagnosed? What do social experiences look like earlier in the journey? In Chapter 4, I investigated information and support seeking behaviors online– how might these behaviors be different if the respondent did not have a specific disease name to use when searching for information or support groups? How do support needs evolve over time?

Similarly, our machine learning models were able to correctly classify whether a respondent had a rare disease or a common chronic illness– how well does that model extend to people who do not know what condition they have? Do the two categories of people still behave sufficiently differently even before they are diagnosed? There is an ethical dilemma here as well: if we were able to identify undiagnosed people who potentially have a rare disease from their online behavior, what do we do with this information? Some existing research has considered early detection of diseases from search engine data [263, 264, 290, 349] where people are actively seeking answers about a health query; this is fundamentally different from detecting disease categories on mainstream social media sites that people visit for a wide range of reasons. Do people want to know this information? What if the condition is untreatable or poorly understood? How might we subtly nudge people towards support resources or appropriate professionals? How might we help people find a network of similar others without disclosing private information to unwanted audiences?

There are also a number of open research questions dealing with the design of support exchange networks. The research in this dissertation explored values and behaviors around selecting a helper from online profiles. Future work might consider the mutualities or characteristics that are most important not just for selecting from a set of profiles, but for ensuring successful interactions. Are there certain mutualities or characteristics that are important for connecting offline? Are there mutualities that are more relevant for support delivered remotely? Which characteristics lead to successful long term relationships? In a helping network for people with rare diseases, a sense of *community* is important for addressing network support needs of people with rare diseases who may be otherwise isolated. How might we design such a system to be more community oriented and encourage long term relationship building, rather than merely transactional interactions? What might feedback, reviewing, or reporting mechanisms look like for such a system? This may be an area where we can learn from the strong history of patient activism to engage with people with rare diseases as key shapers and moderators of these communities.

9.6 Conducting Research with Unique Populations

> A secondary contribution of this dissertation is a methodological contribution [357]. In this dissertation, I provide methods and approaches for conducting research with small, dispersed, sensitive populations.

Conducting research with people with rare diseases as well as people with common chronic illnesses means that survey data is often skewed towards the more common examples. **We provide an an investigation of approaches to analyzing imbalanced data to better understand people with rare diseases within larger data sets.** Additionally, people with rare diseases are often distributed geographically around the world. **We introduce researchers to the Asynchronous Remote Communities (ARC) method for conducting groups-based research remotely.** Finally, there is great potential for conducting additional research with people with rare diseases if data can be anonymized and shared, but there are also significant re-identification risks associated with rare disease data. **In Chapter 9 we contribute an evaluation of the potential for releasing privacy-preserving versions of rare disease data to facilitate additional research.**

9.6.1 Future Work

This dissertation provides reflections on our experiences in conducting research with this population but, as I describe in Part III, we were not always successful and there were many areas where additional research is necessary to be able to ethically and effectively work with people with rare diseases or other unique populations.

First, I argue that there is significant future research necessary to explore ways of conducting informed consent in a computer mediated way. If participants are merely instructed to read an informed consent document and indicate their consent, researchers do not have the same opportunity to assess how well a potential participant truly understands and participants may miss out on opportunities to ask questions. Future work might investigate ways of verifying comprehension in asynchronously (e.g., in a survey) or synchronously (e.g., in a video interview) obtained consent.

Similarly, the research community should reflect on best practices for collaborating with participants throughout the research process to engage in ongoing conversations about the use of participant data (rather than merely at the initial point of informed consent) but also ensure that the research is truly well-aligned with participants needs and goals. What might CBPR look like when the community is a virtual one? How might we collaborate remotely with rare disease stakeholders as research partners, given that people with rare diseases are very interested not just in participating in research but also in shaping research directions (see Chapter 1)?

There are also several open questions about the future of the ARC method. Our development of ARC was entirely informed by our research goals in working with people with rare diseases, including the decision to conduct the study on Facebook. As other researchers adopt ARC for their own research questions and for different populations (e.g., [210, 275]), we consider what ARC might be if it were conducted on another platform. What other features of these platforms might facilitate different kinds of activities? How would the study be different if participants were not limited to their real names? How might we integrate synchronous elements into an ARC study? What benefits of in-person research do we lose in conducting research remotely?

9.7 Summary

Overall, we conclude that people with rare diseases expect to receive different kinds of support from different social relationships and are willing to share the information that is relevant and necessary for achieving these goals (see Figure 6.5). My research has suggested that disease-specific communities on mainstream social media platforms are an effective way for people with rare diseases to provide and receive informational and nurturant support. It is my hope that the insights presented in this dissertation will provide designers with the necessary research to not only expand these systems to also consider tangible support (as I have proposed within) but also to consider ways of improving existing practices around online health communities, particularly those couched within larger social networks. I have drawn attention to the challenges of balancing multiple audiences with different support goals. Additionally, in considering areas where people with rare diseases are similar to or different from people with common chronic illnesses, I open up the possibility to extend this research to additional populations in the future. In investigating social support in rare disease and common chronic illness communities, I have provided a foundation on which we can build novel sociotechnical systems to connect people who may be otherwise isolated and provide them with the support they need.

Appendix A

Institutional Review Board Approvals

All studies in this dissertation were approved by Indiana University's Institutional Review Board. This appendix includes the approval notices for these studies.

INDIANA UNIVERSITY

OFFICE OF THE VICE PRESIDENT FOR RESEARCH

Office of Research Compliance

To: Katherine Connelly
INFORMATICS

Haley MacLeod
INFORMATICS

From:



Chair - IRB-IUB
Human Subjects Office
Office of Research Compliance – Indiana University

Date: May 27, 2016

RE: **NOTICE OF EXPEDITED APPROVAL - RENEWAL WITH AMENDMENT**

Protocol Title: Sensemaking Practices of People with Rare Diseases

Study #: 1405068949R001

Funding Agency/Sponsor: None

Review Level: Expedited

Status: Approved | Submitted to IRB

Study Approval Date: May 24, 2016

Study Expiration Date: May 23, 2018

The Indiana University Institutional Review Board (IRB) IRB00000222 | IRB-IUB recently reviewed the renewal with amendment associated with the above-referenced protocol. In compliance with (as applicable) 21 C.F.R. § 56.109 (e) and 46 C.F.R. § 46.109 (d), this letter serves as written notification of the IRB's determination.

The study is approved under Expedited Category (6) Category 6: Collection of data from voice, video, digital, or image recordings made for research purposes. **(7)** Category 7: Research on individual or group characteristics or behavior (including, but not limited to, research on perception, cognition, motivation, identity, language, communication, cultural beliefs or practices, and social behavior) or research employing survey, interview, oral history, focus group, program evaluation, human factors evaluation, or quality assurance methodologies. (NOTE: Some research in this category may be exempt from the HHS regulations for the protection of human subjects. 45 CFR 46.101(b)(2) and (b)(3). This listing refers only to research that is not exempt.)**, with the following determinations, as applicable:**

Approval of this study is based on your agreement to abide by the policies and procedures of the Indiana University Human Research Protection Program and does not replace any other approvals that may be required. Relevant policies and procedures governing Human Subject Research can be found at: http://researchadmin.iu.edu/HumanSubjects/hs_policies.html.

As a reminder, IRB approval is required prior to implementing any changes or amendments in the protocol, regardless of how minor, except to eliminate immediate hazards to subjects. No changes to the informed consent document may be made without prior IRB approval.

If you submitted and/or are required to provide participants with an informed consent document, please ensure you are using the most recent version of the document to consent subjects.

The approval period is noted above. Failure to receive notification from the Human Subjects Office will not relieve you of your responsibility to ensure compliance with Federal Regulations regarding annual review [as applicable, 21 C.F.R. § 56.109(f) and 45 C.F.R. § 46.109(e)].

You should retain a copy of this letter and all associated approved study documents for your records. Please refer to the assigned study number and exact study title in future correspondence with our office. Additional information is available on our website at http://researchadmin.iu.edu/HumanSubjects/.

If your source of funding changes, you must submit an amendment to update your study documents immediately.

If you have any questions or require further information, please contact the Human Subjects Office via email at irb@iu.edu or via phone at (317)274-8289 (Indianapolis) or (812) 856-4242 (Bloomington).

You are invited, as part of ORA's ongoing program of quality improvement, to **participate in a short survey** to assess your experience and satisfaction with the IRB related to this approval. We estimate it will take you approximately **5 minutes to complete the survey.** The survey is housed on a Microsoft SharePoint secure site which requires CAS authentication. This survey is being administered by REEP; please contact us at reep@iu.edu if you have any questions or require additional information. Simply click on the link below, or cut and paste the entire URL into your browser to access the survey: https://www.sharepoint.iu.edu/sites/iu-ora/survey/Lists/Compliance/IRB_Survey/NewForm.aspx.

/enclosures

INDIANA UNIVERSITY

OFFICE OF THE VICE PRESIDENT FOR RESEARCH

Office of Research Compliance

To: Katherine Connelly
INFORMATICS

Haley MacLeod
INFORMATICS

Kimberly Oakes
INFORMATICS

From: Kenneth C. Mann

Human Subjects Office
Office of Research Compliance – Indiana University

Date: November 20, 2014

RE: **NOTICE OF EXEMPTION - AMENDMENT**

Protocol Title: Technology Use by People with Chronic Illness

Study #: 1410636078A001

Funding Agency/Sponsor: None

Status: Exemption Granted | Exempt

Study Approval Date: November 20, 2014

The Indiana University Institutional Review Board (IRB) EXE000001 | Exempt recently reviewed the above-referenced amendment on November 17, 2014. In compliance with 46 C.F.R. § 46.109 (d), this letter serves as written notification of exempt determination.

The amendment is accepted under 45 C.F.R. § 46.101 (b), paragraph(s) (2) Category 2: Surveys/Interviews/Standardized Educational Tests/Observation of Public BehaviorResearch involving the use of educational tests (cognitive, diagnostic, aptitude, achievement), survey procedures, interview procedures or observation of public behavior if: i) information obtained is recorded in such a manner that human subjects cannot be identified, directly or through identifiers linked to the subjects; or ii) any disclosure of the human subjects responses outside the research would not reasonably place the subjects at risk of criminal or civil liability or be damaging to the subjects financial standing, employability or reputation.

Acceptance of this amendment is based on your agreement to abide by the policies and procedures of the Indiana University Human Research Protection Program and does not replace any other approvals that may be required. Relevant policies and procedures governing Human Subject Research can be found at: http://researchadmin.iu.edu/HumanSubjects/hs_guidance.html.

The Exempt determination is valid indefinitely unless changes in the project may impact the study design as originally submitted. Please check with the Human Subjects Office to determine if any additional review may be needed.

You should retain a copy of this letter and all associated approved study documents for your records. Please refer to the assigned study number and exact study title in future correspondence with our office. Additional information is available on our website at http://researchadmin.iu.edu/HumanSubjects/.

If your source of funding changes, you must submit an amendment to update your study documents immediately.

If you have any questions or require further information, please contact the Human Subjects Office via email at irb@iu.edu or via phone at (317)274-8289 (Indianapolis) or (812) 856-4242 (Bloomington).

You are invited, as part of ORA's ongoing program of quality improvement, to **participate in a short survey** to assess your experience and satisfaction with the IRB related to this approval. We estimate it will take you approximately **5 minutes to complete the survey.** The survey is housed on a Microsoft SharePoint secure site which requires CAS authentication. This survey is being administered by REEP; please contact us at reep@iu.edu if you have any questions or require additional information. Simply click on the link below, or cut and paste the entire URL into your browser to access the survey: https://www.sharepoint.iu.edu/sites/iu-ora/survey/Lists/Compliance/IRB_Survey/NewForm.aspx.

/enclosures

INDIANA UNIVERSITY

OFFICE OF THE VICE PRESIDENT FOR RESEARCH

Office of Research Compliance

To: Katherine Connelly
INFORMATICS

Haley MacLeod
INFORMATICS

From:



Chair - IRB-IUB
Human Subjects Office
Office of Research Compliance – Indiana University

Date: February 23, 2017

RE: **NOTICE OF EXPEDITED APPROVAL - RENEWAL WITH AMENDMENT**

Protocol Title: Designing with People with Rare Diseases

Study #: 1412065553R001

Funding Agency/Sponsor: N/A

Review Level: Expedited

Status: Approved | Submitted to IRB

Study Approval Date: February 23, 2017

Study Expiration Date: February 22, 2019

The Indiana University Institutional Review Board (IRB) IRB00000222 | IRB-IUB recently reviewed the renewal with amendment associated with the above-referenced protocol. In compliance with (as applicable) 21 C.F.R. § 56.109 (e) and 46 C.F.R. § 46.109 (d), this letter serves as written notification of the IRB's determination.

The study is approved under Expedited Category (7) Category 7: Research on individual or group characteristics or behavior (including, but not limited to, research on perception, cognition, motivation, identity, language, communication, cultural beliefs or practices, and social behavior) or research employing survey, interview, oral history, focus group, program evaluation, human factors evaluation, or quality assurance methodologies. (NOTE: Some research in this category may be exempt from the HHS regulations for the protection of human subjects. 45 CFR 46.101(b)(2) and (b)(3). This listing refers only to research that is not exempt.)**, with the following determinations, as applicable:**

Approval of this study is based on your agreement to abide by the policies and procedures of the Indiana University Human Research Protection Program and does not replace any other approvals that may be required. Relevant policies and procedures governing Human Subject Research can be found at: http://researchcompliance.iu.edu/hso/hs_guidance.html.

As a reminder, IRB approval is required prior to implementing any changes or amendments in the protocol, regardless of how minor, except to eliminate immediate hazards to subjects. No changes to the informed consent document may be made without prior IRB approval.

If you submitted and/or are required to provide participants with an informed consent document, please ensure you are using the most recent version of the document to consent subjects.

The approval period is noted above. Failure to receive notification from the Human Subjects Office will not relieve you of your responsibility to ensure compliance with Federal Regulations regarding annual review [as applicable, 21 C.F.R. § 56.109(f) and 45 C.F.R. § 46.109(e)].

You should retain a copy of this letter and all associated approved study documents for your records. Please refer to the assigned study number and exact study title in future correspondence with our office. Additional information is available on our website at at http://researchcompliance.iu.edu/hso/.

If your source of funding changes, you must submit an amendment to update your study documents immediately.

If you have any questions or require further information, please contact the Human Subjects Office via email at irb@iu.edu or via phone at (317)274-8289 (Indianapolis) or (812) 856-4242 (Bloomington).

INDIANA UNIVERSITY

OFFICE OF THE VICE PRESIDENT FOR RESEARCH

Office of Research Compliance

To: Katherine Connelly
DEPARTMENT OF INFORMATICS

Haley MacLeod
DEPARTMENT OF INFORMATICS

From:

Human Subjects Office
Office of Research Compliance – Indiana University

Date: January 03, 2018

RE: **NOTICE OF EXEMPTION - AMENDMENT**

Protocol Title: Selecting online profiles in a helping network

Study #: 1711196782A001

Funding Agency/Sponsor: None

Status: Exemption Granted | Exempt

Study Approval Date: January 03, 2018

The Indiana University Institutional Review Board (IRB) EXE000001 | Exempt recently reviewed the above-referenced protocol. In compliance with (as applicable) 45 CFR 46.109 (d) and the IU Standard Operating Procedures (SOPs) for Research Involving Human Subjects, this letter serves as written notification of exempt determination.

Under 45 CFR 46.101(b) or the SOPs, as applicable, the amendment is accepted as Exempt (710) Category 7 Flex: Research that is not federally funded or otherwise federally regulated and is on individual or group characteristics or behavior (including, but not limited to, research on perception, cognition, motivation, identity, language, communication, cultural beliefs or practices, and social behavior) or employs survey, interview, oral history, focus group, program evaluation, human factors evaluation, or quality assurance methodologies., with the following determinations:

Acceptance of this amendment is based on your agreement to abide by the policies and procedures of the Indiana University Human Research Protection Program and does not replace any other approvals that may be required. Relevant policies and procedures governing Human Subjects Research can be found at: http://researchcompliance.iu.edu/hso/hs_guidance.html.

The Exempt determination is valid indefinitely. Substantive changes to approved exempt research must be requested and approved prior to their initiation. Investigators may request proposed changes by submitting an amendment through the KC IRB system. The changes are reviewed to ensure that they do not affect the exempt status of the research. Please check with the Human Subjects Office to determine if any additional review may be needed.

You should retain a copy of this letter and all associated approved study documents for your records. Please refer to the assigned study number and exact study title in future correspondence with our office. Additional information is available on our website at at http://researchcompliance.iu.edu/hso/.

If your source of funding changes, you must submit an amendment to update your study documents immediately.

If you have any questions or require further information, please contact the Human Subjects Office via email at irb@iu.edu or by phone at 317-274-8289 (Indianapolis) or 812-856-4242 (Bloomington).

Appendix B

Thematic Interview Guide

This appendix includes the thematic interview guide used for the interview study described in Chapter 3.

Background

- Demographics – age, gender, country
- What condition do you have?
- What can you tell me about the condition?
- How long have you had it?

Contextual Questions

- Tell me about your experience with this condition?
- Walk me through a day when...
  - good day,
  - bad day,
  - seeing your doctor
  - understanding,
  - not understanding,

Living with the condition

- How did you feel when you were first diagnosed?
- How does having this condition impact your day to day life?

Support

- What medical support do you get for this condition? Are you satisfied with it? Why/why not?
- What do you bring to your medical appointments?
- Do you receive support from your family or friends? What is that like?
- Do you receive support from anywhere else? Where? What is that like?
- What do they help you with?

Technology – paper, tv, computers, etc.

- What, if any, technology to you use to support your condition?
- How/why do you use it?
- Have you used other technology in the past?
- If you could have a magic tool, that would do anything, what would it do?

Sensemaking

- How well do you feel you understand your condition?
- How well do you feel you are able to manage it?
- Has that changed since you were diagnosed at all?
- Do you track any data about your condition?
- What do you do if you have a question or concern about your condition? How effective is that?

Appendix C

Survey on Technology Use & Information Seeking

This appendix contains the survey instrument discussed in Chapters 4 and 7.

Use of Technology by People with Chronic Illnesses

You are invited to participate in a research study of the use of technology by people with chronic illness. We ask that you read this information and ask any questions you may have before agreeing to be in the study.

The study is being conducted by Haley MacLeod, Kim Oakes, and Kay Connelly in the School of Informatics and Computing.

The purpose of this study is to understand the use of technology by people with rare and common chronic diseases, as well as how people perceive their doctors. We anticipate this survey will take approximately 15 minutes to complete.

Efforts will be made to keep your personal information confidential. We cannot guarantee absolute confidentiality. Your personal information may be disclosed if required by law. Your identity will be held in confidence in reports in which the study may be published.

Organizations that may inspect and/or copy your research records for quality assurance and data analysis include groups such as the study investigator and her research associates, the Indiana University Institutional Review Board or its designees, and (as allowed by law) state or federal agencies, specifically the Office for Human Research Protections (OHRP) who may need to access your research records.

You will not receive payment for taking part in this study.

For questions about the study contact the researcher Haley MacLeod at hemacleo@indiana.edu or 812-369-0463. For questions about your rights as a research participant or to discuss problems, complaints or concerns about a research study, or to obtain information, or offer input, contact the IU Human Subjects Office at (812) 856-4242 or (800) 696-2949.

Taking part in this study is voluntary. You may choose not to take part or may leave the study at any time. Leaving the study will not result in any penalty or loss of benefits to which you are entitled. Your decision whether or not to participate in this study will not affect your current or future relations with Indiana University.

In consideration of all of the above, I give my consent to participate in this research study.

Demographic Information

1. **Age:**

2. **Gender:**
   Mark only one oval.
   - Male
   - Female
   - Other

3. **Country of Residence:**

4. **Marital Status:**
Mark only one oval.

- Married
- Living with a partner
- Divorced
- Separated
- Widowed
- Single
- Other: _____

5. **Education**
Mark only one oval.

- Less than grade 8
- Some high school
- Completed high school
- Technical/trade/vocational school
- Some college/university
- Completed college/university
- Some post-graduate education
- Completed post-graduate education

6. **Employment**
Mark only one oval.

- Full time
- Part time
- Retired
- Student
- Disabled
- Not employed for pay

Chronic Disease Information

7. **Disease Name**

_____

8. **How many years has it been since you first started experiencing symptoms?**

_____

9. **How many years has it been since you were diagnosed?**

_____

10. **How severely do your symptoms impact your life?**
Mark only one oval.

	1	2	3	4	5	
No impact on my life	◯	◯	◯	◯	◯	Extreme impact on my life

Technology Use

11. **How often do you use the Internet?**
Mark only one oval.

- ◯ Several times per day
- ◯ About once a day
- ◯ Several times a week
- ◯ Every few weeks
- ◯ Less often
- ◯ Never

12. **Do you own any of the following technologies? (Check all that apply)**
Check all that apply.

- ☐ Desktop computer
- ☐ Laptop computer
- ☐ Cell phone
- ☐ e-Reader
- ☐ MP3 player
- ☐ Game console
- ☐ Tablet

13. **On your cell phone, do you have any applications that help you track or manage your health?**
Mark only one oval.

- ◯ Yes
- ◯ No
- ◯ I don't have a cell phone

14. **Do you ever use your cell phone to look up health or medical information?**
Mark only one oval.

- ◯ Yes
- ◯ No
- ◯ I don't have a cell phone

15. **Have you ever looked online for information about any of the following? (Check all that apply)**
Check all that apply.

- ☐ Information about a specific disease or medical problem
- ☐ Information about any other health issue
- ☐ Information about memory loss
- ☐ Information about medical test results
- ☐ Information related to health insurance
- ☐ Information about environmental health hazards
- ☐ Information about hospitals or other medical facilities
- ☐ Information about doctors or other health professionals
- ☐ Information about a certain medical treatment or procedure
- ☐ Information about managing chronic pain
- ☐ Information about drug safety or recalls

16. **Have you ever done any of the following? (Check all that apply)**
Check all that apply.

- ☐ Gone online to find others who might have health concerns similar to yours
- ☐ Signed up to receive email updates or alerts about health or medical issues
- ☐ Tracked other health indicators or symptoms online
- ☐ Read someone else's commentary or experience about health or medical issues on an online group, website, or blog
- ☐ Watched an online video about health or medical issues
- ☐ Tracked your weight, diet or exercise routine online

17. **Have you posted comments, questions or information about your health or medical issues on any of the following? (Check all that apply)**
Check all that apply.

- ☐ In a health related online discussion not specific to your condition
- ☐ On a website of any kind
- ☐ On YouTube or other video sharing site
- ☐ On a blog
- ☐ In an online discussion specific to your condition
- ☐ On Facebook, Twitter or another social networking site

18. **Have you ever used a social networking site to do any of the following? (Check all that apply)**
Check all that apply.

- [ ] Start or join a health-related group
- [ ] Get health information
- [ ] Remember or memorialize others who suffered from a certain health condition
- [ ] Raise money or draw attention to a health related issue or cause
- [ ] Follow your friends' personal health experiences or health updates

19. **Have you been helped by following medical advice or health information found on the internet?**
Mark only one oval.

- ◯ Yes, major help
- ◯ Yes, moderate help
- ◯ Yes, minor help
- ◯ No
- ◯ No, minor harm
- ◯ No, moderate harm
- ◯ No, serious harm

20. **Have you ever done any of the following online? (Check all that apply)**
Check all that apply.

- [ ] Posted a review online of a hospital
- [ ] Consulted online reviews of particular drugs or medical treatments
- [ ] Consulted online rankings or reviews of hospitals or other medical facilities
- [ ] Consulted online rankings or reviews of doctors or other providers
- [ ] Posted your experience with a particular drug or medical treatment online
- [ ] Posted a review online of a doctor

Healthcare Professionals

21. **Do you have a personal or family doctor that you rely on if you need medical care?**
Mark only one oval.

- ◯ Yes
- ◯ No

22. **How many specialists did you see in the last two years?**
Mark only one oval.

- 0
- 1-2
- 3-5
- More than 5

Think about the doctor or healthcare professional that you get most of your medical care from.

23. **How helpful is this person in giving you an accurate medical diagnosis?**
Mark only one oval.

	1	2	3	4	5	
Very Helpful						Very Unhelpful

24. **How helpful is this person in providing emotional support?**
Mark only one oval.

	1	2	3	4	5	
Very Helpful						Very Unhelpful

25. **How helpful is this person in providing the medical or health information that you need?**
Mark only one oval.

	1	2	3	4	5	
Very Helpful						Very Unhelpful

26. **How helpful is this person in finding effective treatment strategies for you?**
Mark only one oval.

	1	2	3	4	5	
Very Helpful						Very Unhelpful

27. **How helpful is this person in coordinating your overall health care?**
Mark only one oval.

	1	2	3	4	5	
Very Helpful						Very Unhelpful

Overall, who is most helpful...

28. **...when you need an accurate medical diagnosis?**
Mark only one oval.

- Online sources
- Fellow patients
- Health professionals
- Friends and family

29. **...when you need emotional support in dealing with a health issue?**
Mark only one oval.

- Fellow patients
- Health professionals
- Online sources
- Friends and family

30. **...when you need practical advice for coping with day to day health situations?**
Mark only one oval.

- Friends and family
- Health professionals
- Fellow patients
- Online sources

31. **...when you need information about alternative treatments?**
Mark only one oval.

- Friends and family
- Online sources
- Fellow patients
- Health professionals

32. **...when you need information about prescription drugs?**
Mark only one oval.

- Friends and family
- Online sources
- Fellow patients
- Health professionals

33. **...when you need a quick remedy for an everyday health issue?**
Mark only one oval.

- Friends and family
- Health professionals
- Fellow patients
- Online sources

34. **...when you need a recommendation for a doctor or specialist?**
Mark only one oval.

- Online sources
- Health professionals
- Friends and family
- Fellow patients

35. **...when you need a recommendation for a hospital or other medical facility?**
Mark only one oval.

- Fellow patients
- Friends and family
- Health professionals
- Online sources

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Appendix D

Survey on Helper Selection& Information Sharing

This appendix contains the survey instrument discussed in Chapters 6 and 7.



We are looking for adults (18+) with a chronic illness who use Facebook to participate in a research study about helping other people. It should take approximately 10-15 minutes to complete the survey.

This project is being conducted by researchers at Indiana University under the direction of Haley MacLeod, Kay Connelly, and Katie Siek in the School of Informatics, Computing, and Engineering. The **purpose of this research** is to understand the kinds of tasks people need help with and the kinds of tasks people can help other people with. We also aim to understand how people make decisions when asked to choose a helper based on an online profile. This helps us to build better online communities.

Study procedures. We will begin by asking you some demographic questions about yourself. Next, we will ask you to tell us about different tasks you might like help with sometimes and which ones you think you could help other people with. We will also ask what information you think is most useful if you were choosing a helper based on an online profile as well as what information you would be

comfortable sharing on your own profile. Finally, you will have the option to provide your contact information if you wish to be entered into a draw for an Amazon gift card or if you would like to be contacted for a follow up study.

Taking part in this research study is voluntary. You may remain anonymous if you wish. You may choose not to provide contact information. You may choose not to take part in the study or may choose to leave the study at any time. There will be no consequences if you decide not to participate or if you leave the study part way through.

Benefits. We don’t expect you to receive any direct benefit from taking part in this study, but we hope to learn things which will help scientists in the future.

Risks. Since this study only includes collection of information about you, the only risk to you is a possible loss of confidentiality. We will do everything possible to protect your information, but we cannot guarantee absolute confidentiality. Your personal information may be disclosed if required by law.

Compensation. As a token of our appreciation for your participation, you may enroll yourself in a drawing to win a $25 Amazon gift certificate (1 in 100 chance of winning).

If you have any **questions** about the study, you can contact Haley MacLeod at hemacleo@indiana.edu.

This study has been approved by Indiana University's Institutional Review Board (Study #1711196782). For questions about your rights as a research participant, to discuss problems, complaints, or concerns about a research study, or to obtain information or to offer input, please contact the IU Human Subjects Office at irb@iu.edu.

I agree to participate

1 How old are you? *

○ Less than 18 ◉ 18-25 ○ 26-30 ○ 31-35 ○ 36-40 ○ 41-45 ○ 46-50
○ 51-55 ○ 56-60 ○ 61-65 ○ 66-70 ○ 71-75 ○ Older than 75

2 Do you have any long term or chronic health conditions? *

◉ Yes, I have at least one chronic condition ○ No, I don't have any chronic conditions

3 Which of the following describes your chronic illness(es)?

☐ I have a common chronic illness ☐ I have a rare disease ☐ I have an undiagnosed condition
☐ I'm not sure if my disease is rare or common

4 What is the name of your chronic condition? If you have multiple chronic conditions, you can list them here. If you have an diagnosed condition, please briefly describe your symptoms. *

5 Do you use any of these social networks? *

☐ Other ☐ LinkedIn ☐ YouTube ☑ Facebook ☐ Snapchat ☐ Twitter
☐ Patients Like Me ☐ Instagram ☐ Pinterest ☐ Tumblr ☐ Reddit

6 Which pronouns would you want someone to use if they were talking about you?

○ She/Her/Hers ○ He/Him/His ○ They/Them/Theirs ○ Something else

7 Which country do you think of as home?

8 What is the highest level of education you have completed?

- ○ Less than high school
- ○ High school or equivalent
- ○ Trade/technical/vocational training
- ○ Associate degree
- ○ Bachelor's degree
- ○ Master's degree
- ○ Professional degree
- ○ Doctoral degree

9 For the next part of this survey, please imagine you a part of a community of people who help each other out. Every time you help someone else, you earn points that you can trade in for someone else to do a favor for you.

a. What kinds of tasks would you be able to help someone else with? *

- ☐ Moving heavy items
- ☐ Cooking
- ☐ Cleaning
- ☐ Legal advice
- ☐ Computer tasks
- ☐ Medical or scientific advice
- ☐ Financial advice
- ☐ Picking things up from the store
- ☐ I wouldn't be able to help with any of these things
- ☐ Driving
- ☐ Laundry

b. What other things could you help with? Are there any special skills you have that would be helpful to other people?

c. You're a great helper and you've accumulated a lot of points. You can now trade in those points for someone to help you out! What kind of tasks would you like to trade in

your points for? *

☐ Moving heavy items ☐ I wouldn't want help with any of these things ☐ Computer tasks
☐ Medical or scientific advice ☐ Cooking ☐ Legal advice ☐ Picking things up from the store
☐ Financial advice ☐ Laundry ☐ Cleaning ☐ Driving

d. What else would you want to trade in your points for? Imagine you're having a rough week - what kind of things would you like someone else to take care of for you?

e. What is the **most important** thing you would want to learn about a person to decide if you would choose them as your helper?

○ Other ○ Understand their personality and values ○ See what we have in common
○ See what they look like (pictures) ○ See what they've been up to recently
○ Get basic information (hometown, current city, work) ○ Learn about their friends
○ Decide if they are trustworthy ○ Learn about their hobbies and interests

---

10 Now, imagine you're choosing from a list of people willing to help you with one of the tasks you just picked.

a. Which of these people would be your **first choice** to help you out? *

◉ Someone with similar demographics to me (same age, gender, race, sexual orientation, etc.)
○ Someone with similar health experiences as me (same disease, symptoms, tests, medications, etc.)
○ Someone with expertise that complements mine (having special skills or talents)
○ Someone with a similar lifestyle to me (relationship status, children, drinking or smoking, dietary preferences)

- ○ Someone with similar beliefs as me (same religion, political affiliation, etc.)
- ○ Someone with similar interests to me (sports, movies, books, hobbies)

b. Which of these people would be your **second choice** to help you out? *

- ◉ Someone with similar beliefs as me (same religion, political affiliation, etc.)
- ○ Someone with similar health experiences as me (same disease, symptoms, tests, medications, etc.)
- ○ Someone with similar interests to me (sports, movies, books, hobbies)
- ○ Someone with a similar lifestyle to me (relationship status, children, drinking or smoking, dietary preferences)
- ○ Someone with expertise that complements mine (having special skills or talents)

c. Which of these people would be your **third choice** to help you out? *

- ○ Someone with a similar lifestyle to me (relationship status, children, drinking or smoking, dietary preferences)
- ◉ Someone with similar interests to me (sports, movies, books, hobbies)
- ○ Someone with expertise that complements mine (having special skills or talents)
- ○ Someone with similar health experiences as me (same disease, symptoms, tests, medications, etc.)

---

11 Which of the following information would you be comfortable **sharing publicly** on an online profile (e.g., a Facebook profile) so anyone could see it? *

- ☐ Information about my beliefs (religions, political affiliation, etc.)
- ☐ Information about my lifestyle (relationship status, children, drinking or smoking, dietary preferences, etc.)
- ☐ Information about my demographics (age, gender, race, sexual orientation, etc.)
- ☐ I wouldn't be comfortable sharing any of these things
- ☐ Information about my health experiences (disease, symptoms, tests, medications, etc.)

- [ ] Information about my interests (sports, movies, books, hobbies)
- [ ] Information about my expertise (skills, abilities, talents, education, etc.)

---

12 Which of the following information would you be comfortable **sharing with your Facebook friends** via an online profile? *

- [ ] Information about my demographics (age, gender, race, sexual orientation, etc.)
- [ ] Information about my lifestyle (relationship status, children, drinking or smoking, dietary preferences, etc.)
- [ ] Information about my beliefs (religions, political affiliation, etc.)
- [ ] Information about my interests (sports, movies, books, hobbies)
- [ ] Information about my health experiences (disease, symptoms, tests, medications, etc.)
- [ ] Information about my expertise (skills, abilities, talents, education, etc.)
- [ ] I wouldn't be comfortable sharing any of these things

---

13 Which of the following information would you be comfortable **sharing with other people with a chronic illness** via an online profile (e.g., as part of a secret Facebook group or private forum)? *

- [ ] Information about my interests (sports, movies, books, hobbies)
- [ ] Information about my lifestyle (relationship status, children, drinking or smoking, dietary preferences, etc.)
- [ ] Information about my demographics (age, gender, race, sexual orientation, etc.)
- [ ] Information about my expertise (skills, abilities, talents, education, etc.)
- [ ] Information about my beliefs (religions, political affiliation, etc.)
- [ ] Information about my health experiences (disease, symptoms, tests, medications, etc.)
- [ ] I wouldn't be comfortable sharing any of these things

14 You're almost done! We just have a few more questions...

a. Can we contact you for a follow up survey?

◉ Yes ○ No

b. What email address should we use to contact you?

c. Can we look at your Facebook profile? We will only look at parts of your profile that are publicly accessible and no identifying information will be used in publications or presentations that come out of this research.

◉ Yes ○ No

d. What email address or phone number do you use to log into Facebook? We will only use this information to find your Facebook profile (using the search bar on Facebook).

e. Would you like to be entered into a draw for an Amazon gift card?

◉ Yes ○ No

f. What email address should we send your gift card to if you win the draw?

Submit

Appendix E

ARC Study Activities

A1 - Diary. Kientz et al.'s study [178] of visually impaired people locating lost items is a good diary method example. Participants recorded a diary entry each time they lost an object. They were given the option of using a mobile application, calling a phone number to leave a voice mail, or sending the entry by email. Inspired by this method, we asked participants to keep a diary for 24 hours of everyone they interacted with about their condition. The instructions are provided in Figure E.1.

> Activity time! For 24 hours, I'd like you to keep track of interactions you have with other people about your disease. Make a note of:
> - who you talked to (could be people on Facebook, your friends or family, medical providers, etc.),
> - how you talked to them (online, on the phone, in person, etc.)
> - a brief description of what you talked about and how you felt about it.
>
> After 24 hours is up, post your notes as a comment on this post (as much as you are comfortable).
>
> Thanks!

Figure E.1: Diary activity prompt.

A2 - Circles. Moncur et al. [234] conducted a focus group with mothers where they asked participants to use post-it notes to write down different types of information they gave out when their baby was in the hospital. They had participants organize these post-it notes on a wall grouped by social network hierarchy layers [150]. We adapted this method by asking participants to

draw circles representing this social network hierarchy and use household objects to represent the information distance of various people from themselves (at the center of the circles). The specific instructions for this activity are provided in Figure E.2.

[ACTIVITY] For today's activity, you're going to need a paper and pen, and some small colourful objects - these could be buttons, paper clips, candies, whatever you've got lying around.

What I'd like you to do is draw some circles inside of some other circles. Imagine that you are in the center circle and that the people you know are represented by the small objects. Place these small objects in different layers of the circle based on how much information you think it would appropriate for them to receive about your condition or your health. You can use the small object to represent a specific individual or a type of individual.

I'm including an example here of one I did, imagining more of a social scenario than a health one since I don't actually have a rare disease. Basically blue is friends, green is family, pink is professors or bosses, and purple is more distance acquaintances. Closest to the center are my three closest friends. The three closes green pins represent my three immediate family members. After that the pins start to represent groups of people (e.g. friends from Indiana, friends from back home, extended family in general, etc.).

Please post a picture of what you come up with to the group. You can label it or describe it in the comments. Thanks!

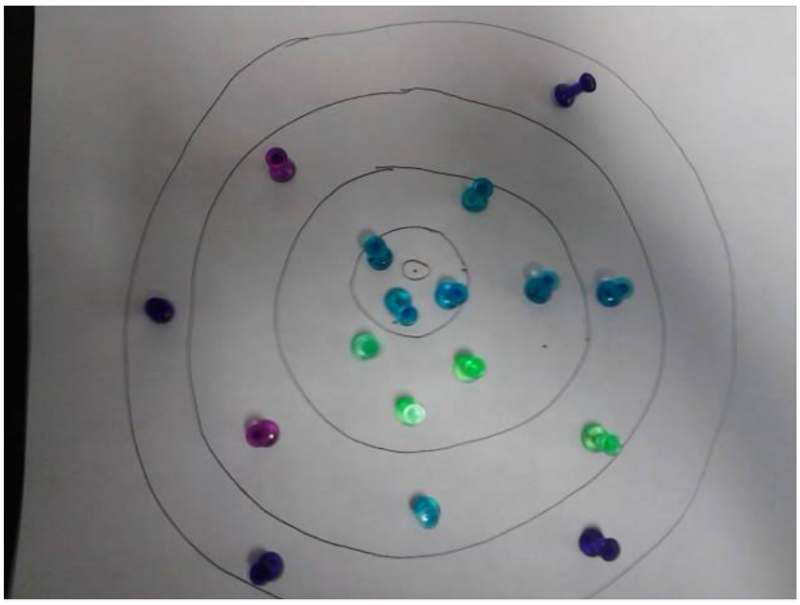


Figure E.2: Circles activity prompt.

A3 - Questions. Building from Moncur et al. [235], we asked participants to submit questions they wished people would ask about their condition. Moncur et al. [235] were interested in understanding the types of information people wanted to know about members of their network; in our study we were interested in the perspective of the person with the rare disease so we reframed this activity to ask about the information a participant *would want someone else to know*, rather than information they would want to know about someone else. The specific instructions are provided in Figure E.3.

[ACTIVITY] Thanks to everyone who did the last activity! It sounds like there's a difference between what you want people to know and what they want to know. I was reading a paper (you can find it here if you're curious http://socialdigital.dundee.ac.uk/…/2009-TITB-SpecialIssue….) where they tried to figure out how much information people wanted to know about someone else's health. There's a list of questions/scenarios that they use (picture below), but these are in a bit of a different context. I'm hoping that you can help me come up with a list of questions that would more relevant to having a rare disease. Essentially, what are things that you wish your friends/family would ask you? Post any questions you come up with in the comments.

TABLE II
MODEL OF REPORT CONTENT BY HIERARCHY LAYER AND GENDER

INFORMATION ITEM	SUPPORT CLIQUE		SYMP. GROUP		CLAN	
	M	*F*	*M*	*F*	*M*	*F*
Which hospital is XXX in?	E	E	N	E	N	N
How can I contact XXX?	E	E	N	E	N	N
Is there anything that XXX wants me to do in particular?	E	E	N	E	N	N
What's wrong with XXX? (short summary)	E	E	N	N	N	N
What are the long-term effects of this illness?	E	E	N	N	N	N
How long is XXX likely to be in hospital?	N	E	N	N	N	N
What practical help does XXX need outside of hospital (e.g. - help with home or work commitments while XXX is in hospital and cannot carry out normal activities)?	E	E	N	N	--	N
What are the hospital visiting times?	E	E	N	N	--	N
How can I help with how XXX is feeling emotionally?	E	E	N	N	--	N
Any change in XXX's condition since they went into hospital?	E	E	N	N	--	N
How is XXX coping emotionally?	E	E	N	N	--	N
Does XXX need surgery?	E	E	N	N	--	N
Does XXX need help with breathing?	N	E	N	N	--	N
Is there a risk that I could have caught Sarrenitis from XXX?	N	N	N	N	--	N
What's wrong with XXX? (detailed explanation)	N	E	N	N	--	--
What does XXX want me to tell other friends and relatives?	N	E	N	N	--	--
What practical help does XXX need in hospital?	N	N	--	N	--	--
What drugs is XXX being given?	N	N	--	N	--	--
Is Sarrenitis an inherited condition?	N	N	--	N	--	--

E= Essential; N= Nice to know; -- = Exclude

Figure E.3: Questions activity prompt.

A4 - Problems. In this activity, we synthesized our findings so far from posts and comments in this study as well as from earlier studies (Chapter 3). We conducted preliminary analysis and synthesized findings into a list of 15 issues raised by participants and asked them to rank order these by importance as well as discuss the accuracy of the list. The instructions are provided in Figure E.4.

[ACTIVITY] I've put together a list of all the problems I've seen in your comments. Please rank them in order of how much you agree that they are problems for you personally (most problematic at the top, not at all problematic at the bottom). And if there's anything missing from the list, you can add it in too. Here's the list in no particular order:
1) Practical, routine tasks
2) People not understanding
3) People not caring
4) People not believing me
5) People not following through with promises
6) People not remembering
7) Not being able to keep up with friends
8) People being negative towards me
9) Feeling isolated
10) Feeling abnormal
11) Doctors not knowing what the disease is
12) Doctors not believing me
13) Not enough research taking place
14) Ruining relationships
15) Financial support

Figure E.4: Problems activity prompt.

A5 - Photo Elicitation. Le Dantec et al.'s [189] study made use of photo elicitation to capture perceptions of homeless people on technology. They met with participants three times over three weeks. In the first meeting, they provided participants with a disposable camera and instructions on what to document with the camera. These instructions were things like, *"Take photos of places or situations where you needed help"* or *"Take photos of of [sic] the things you use: telephones, buses, radios, televisions."* In the second meeting, they collected the cameras. In the third and final meeting, they conducted an interview about the resulting images. In our study, we asked participants to use their phone cameras to document certain themes (emerging from results of A4) and submit these photos to the researcher. We intended to post these to the group in one batch to

avoid identifying who took which picture and preserve privacy. Instead of follow up interviews, we asked participants to tag and comment on the photos. The instructions for the activity as well as the follow up activity are provided in Figures E.5 and E.6.

[ACTIVITY] We went through your answers to the ranking question from a couple weeks ago, and I'm including the chart below so you can see what's up. Each of the bars is one of the items from the list, and you can see that the 2nd and 3rd items (people not caring and people not understanding) are pretty consistently ranked as being important (they are more compact and have lower numbers).

For the next three days, I'd like you to take pictures that represent these ideas of caring and understanding. What do you want people to care about? what do you want people to understand? how do you try to accomplish this? what are the barriers? Save these pictures until the end of the three days, and then send them to me privately all at once (either via Facebook message or by email at hemacleo@indiana.edu). Afterwards, I will put an album together for the group and there will be a follow up activity, so if there is anything sensitive that you don't want the rest of the group to see, please let me know.

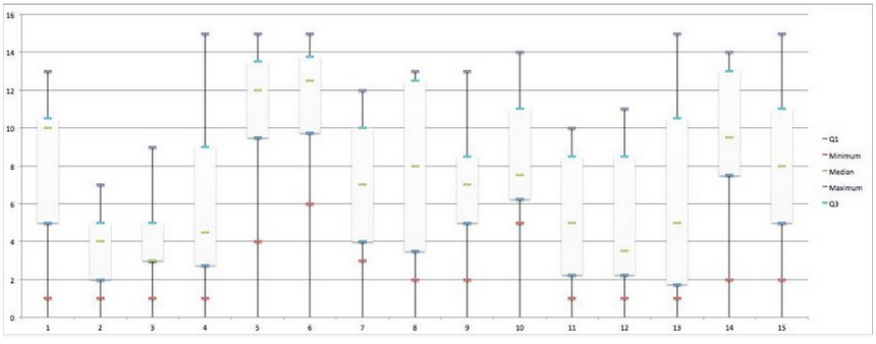


Figure E.5: Photo elicitation activity prompt.

Hi all, I've added the photos below that you submitted - if you didn't submit any photos but are feeling inspired after seeing these, you can still send me some and I will add them 🙂

Some of these have explanations of why they were submitted right on them, and others are less obvious. For each, photo I'd like you to come up with a few (say 3-5) keywords that you think describe how these relate to the theme (people not caring and people not understanding). You can add additional comments if you'd like.

Thanks!

Figure E.6: Photo elicitation follow up.

A6 - Solutions. In A6, we asked participants to discuss possible solutions to the challenges outlined in A4. The instructions were:

> [ACTIVITY] Think about your top two or three answers to my last post (the list of challenges) - what strategies do you use or try to use to address these? Which strategies have been the most successful or helpful?

Figure E.7: Solutions activity prompt.

A7 - Mad Lib. Inspired by Bauer et al.'s [18] use of "DesignLibs" (mad libs for design), we wanted to work with participants to construct scenarios (as in [44]) that described their experiences. We came across [334], which contained an amusing mad lib for people with diabetes that is "Written by People You've Met Who Don't Understand Diabetes". Given that people not understanding was a recurring theme in the first part of this study, we asked participants to adapt this diabetes example to their own experiences with a rare disease.



Figure E.8: Mad lib activity prompt & example mad lib.

A8 - Movie Script. As an alternative approach to generate scenarios with participants, we framed this activity as the creation of a movie script. We asked participants to write a script from a scene from a movie about their life. The instructions were:

[ACTIVITY] Instead of photos, let's try a writing exercise. Imagine you are a writing the script for a movie about your life. Write out a scene that takes place between you and a friend or family member (you can choose which one), keeping in mind the themes of caring/understanding. You have two options 1) Write a scene of how things ACTUALLY are, or 2) Write a scene of how you WISH things were, (or if you're feeling ambitious, one of each!)

Figure E.9: Movie script activity prompt.

A9 - Rant Line. Grimes et al. [133] introduced EatWell, a system allowing people to leave voice memories about their attempts at healthy eating, as well as listen to voice memories that others created. Palen et al. [260] similarly leverage voicemail as a way of conducing diary studies, discussing the advantages of this technique over other diary collection methods including time intensiveness, mobility, and richer descriptions. In our study, we set up a Google Voice account to which participants could call in and leave messages like in [133, 260]. We also gave participants the option of sending in text messages or photo messages. Our hope was that this would help them share their thoughts with us as they were happening or were relevant, rather than having to remember them later. The instructions were:

Thanks everyone for all your contributions! There are only THREE more activities left.

For the next activity, I've set up a "Rant Line" - the number for it is 812-297-8790. You can call it or send texts or photos anytime you feel like you need/want to rant about something. Keep that in mind for the rest of the week and over the weekend.

And if you haven't already done the madlib activity, please do so when you get a chance 🙂.

Figure E.10: Rant line activity prompt.

A10 - Personas. Cooper [66] advocated for personas as a tool to guide designers. Here, we use personas as a way of verifying and adjusting our understanding of the participants in our study. We prepared two personas, representing synthesized and abstracted versions of the participants in our studies. We presented these personas to the group for feedback. The instructions are provided in Figure E.11 and the personas are provided in Figure E.12.

[ACTIVITY] Second last activity! I'm adding pictures below of some personas, loosely based on your contributions to the group. Personas are tools designers use to help guide their ideas. Your job is to critique them: do these apply to you? what's missing? what's wrong? Bonus points if you want to submit or describer your own!
(Blurb here on personas if interested: https://en.wikipedia.org/wiki/Persona_%28user_experience%29)
(Also, the diseases below are completely fictional, just for the record)

Figure E.11: Personas activity prompt.

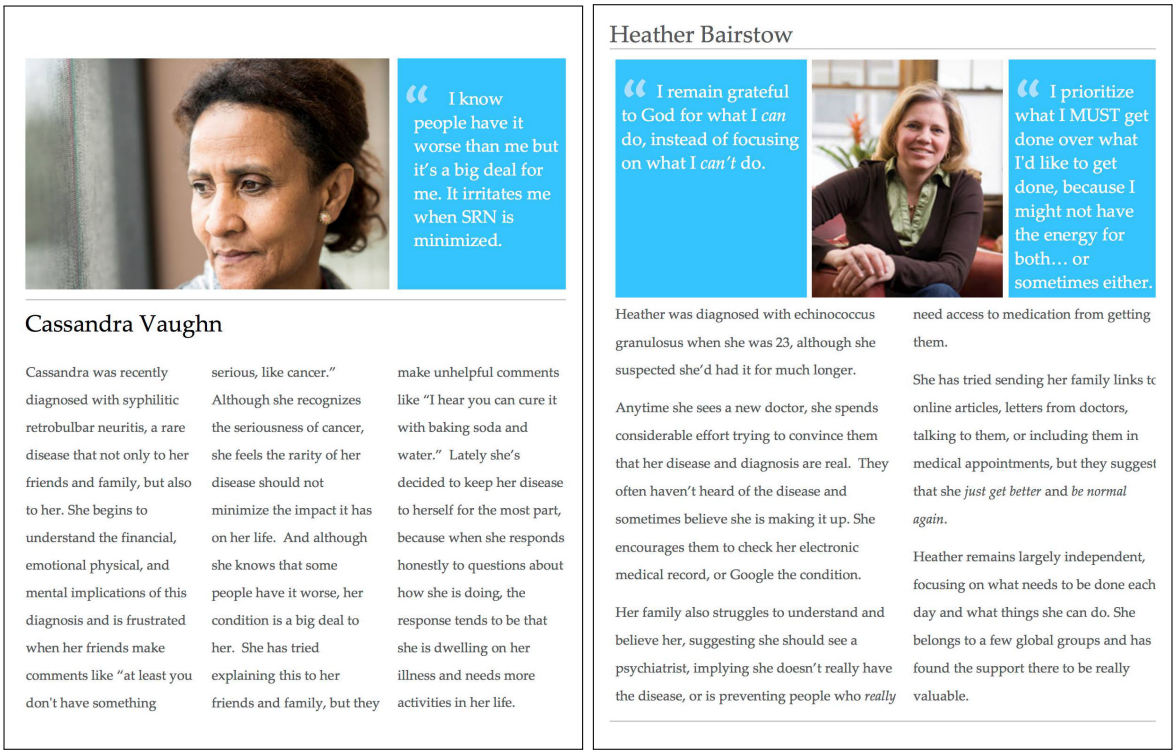


Figure E.12: Personas provided in ARC study.

A11 - Survey. At the end of the study, we asked participants to complete a survey. In our case, this was done to evaluate our methodological approach and understand participants' experiences in the study. We include it in our discussion here because using a survey as part of an ARC study could also be done to gain different types or insights during a study, or to confirm findings with participants. The instructions were:

[ACTIVITY] Last activity! This one is pretty important, so I'm going to tag you all in it to be sure you see it 🙂

I need feedback from you about your experience in this group. Has it been fun or miserable? Easy or hard? Totally ridiculous? I want to hear about it!

Here's a link quick survey with some specific questions I'd like you to answer: https://haley23.typeform.com/to/KQ1P1n The survey will be completely anonymous - I won't be able to tell who said what unless you tell me.

You can also comment here with your feedback if you want to discuss it with the group as a whole. I would also love it if as many of you as possible would be willing to do a short follow up interview, by phone or Skype/GoogleHangout/Facetime - whatever is easiest for you. If you're willing to do this, please comment below or send me a message directly and we can set up a time.

Once the survey is done, I will post a message explaining what happens next on my end, what you can expect from me going forward, and what will happen to the group.

Figure E.13: Survey activity prompt.

-

Appendix F

List of Rare Diseases

ALS "Amyotrophic lateral sclerosis (ALS) is one of a group of disorders known as motor neuron diseases. It is characterized by the progressive degeneration and eventual death of nerve cells (motor neurons) in the brain, brainstem and spinal cord that facilitate communication between the nervous system and voluntary muscles of the body. Ordinarily, motor neurons in the brain (upper motor neurons) sent messages to motor neurons in the spinal cord (lower motor neurons) and then to various muscles. ALS affects both the upper and lower motor neurons, so that the transmission of messages is interrupted, and muscles gradually weaken and waste away. As a result, the ability to initiate and control voluntary movement is lost. Ultimately, ALS leads to respiratory failure because affected individuals lose the ability to control muscles in the chest and diaphragm. ALS is often called Lou Gehrig's disease."[1]

Chiari Malformations "Chiari malformations are a group of complex brain abnormalities that affect the area in lower posterior skull where the brain and spinal cord connect. The underlying anatomy of Chiari malformations is thought to be present at birth (congenital), although in many cases they may not become clinically apparent until adulthood. [...] Part of the cerebellum (known as the cerebellar tonsils) may protrude (herniate) through the foramen

[1] https://rarediseases.org/rare-diseases/amyotrophic-lateral-sclerosis/

magnum, which is the normal opening found in the occipital bone at the base of the skull. The tonsils may thus interfere with the flow of cerebrospinal fluid (CSF) to and from the skull and spinal canal, potentially leading to accumulation of cerebral spinal fluid in the subarachnoid spaces of the brain and spine. A Chiari malformation can also cause pressure on the brain and produce hydrocephalus (pressure due to excessive cerebrospinal fluid accumulation in the brain) and the spinal cord, potentially causing a wide variety of symptoms."[2]

Congenital Anosmia "Congenital anosmia is a condition in which people are born with a lifelong inability to smell. It may occur as an isolated abnormality (no additional symptoms) or be associated with a specific genetic disorder (such as Kallmann Syndrome or congenital insensitivity to pain). Isolated congenital anosmia is usually sporadic, although some familial cases have been reported. In most cases of isolated congenital anosmia, the genetic cause in unknown. Unfortunately, there is currently no cure or treatment for congenital anosmia."[3]

Ehlers-Danlos Syndromes "The Ehlers-Danlos syndromes (EDS) are a group of related disorders caused by different genetic defects in collagen. Collagen is one of the major structural components of the body. Collagen is a tough, fibrous, protein, and serves as a building block essential in both strengthening connective tissue (e.g. bones) and providing flexibility where needed (e.g. cartilage). The problems seen in patients with EDS can be due to either the poor strength of collagen. It may alternatively be due to the absence of sufficient amounts of structurally normal collagen. The primary complications seen in EDS involve the skin, muscles, skeleton, and blood vessels. Patients with EDS often have skin that can be describes as 'velvety', 'loose'. This skin characteristic predisposes patients to problems with wound healing. Patients will often note that they develop 'paper-thin' scars. Patients also have excessively flexible, loose joints. These 'hypermobile' joints can be easily and frequently dislocated. Finally, fragile blood vessels leave patients experiencing easy bruising, even an increased tendency to serious episodes of bleeding."[4]

[2]https://rarediseases.org/rare-diseases/chiari-malformations/
[3]https://rarediseases.info.nih.gov/diseases/9486/congenital-anosmia
[4]https://rarediseases.org/rare-diseases/ehlers-danlos-syndrome/

Familial Transthyretin Amyloidosis Familial transthyretin amyloidosis (FTA) is a progressive condition characterized by abnormal deposits of a protein called amyloid in the body's organs and tissues. Signs and symptoms can vary significantly depending on the location of deposits. Most people have nervous system symptoms that may affect the peripheral, autonomic or central nervous system (brain and spinal cord). Amyloid deposits in the nerves of the peripheral nervous system cause peripheral neuropathy (loss of sensation). Deposits affecting the autonomic nervous system cause a disruption to involuntary body functions, such as blood pressure, heart rate, and digestion. Other areas of the body affected may include the heart, kidneys, eyes, and gastrointestinal tract. FTA is caused by mutations in the TTR gene. Inheritance is autosomal dominant, but not all people with a TTR gene mutation will develop FTA. There is no treatment available that reverses damage caused by amyloid deposits, but there are treatments that may prevent or delay progression. Treatment depends on the tissues affected, and may include liver transplantation (which removes the source of the deposits), heart and/or kidney transplantation, vitrectomy, and/or various medications. Life expectancy depends on many factors and may range from several years to decades after symptoms begin.[5]

Hereditary Angioedema "Hereditary angioedema is a rare inherited disorder characterized by recurrent episodes of the accumulation of fluids outside of the blood vessels, blocking the normal flow of blood or lymphatic fluid and causing rapid swelling of tissues in the hands, feet, limbs, face, intestinal tract, or airway. Usually, this swelling is not accompanied by itching, as it might be with an allergic reaction. Swelling of the gastrointestinal tract leads to cramping. Swelling of the airway may lead to obstruction, a potentially very serious complication. These symptoms develop as the result of deficiency or improper functioning of certain proteins that help to maintain the normal flow of fluids through very small blood vessels (capillaries). In some cases, fluid may accumulate in other internal organs. The severity of the disease varies greatly among affected individuals. The most common form of the disorder is hereditary angioedema type I, which is the result of abnormally low levels of certain complex proteins in the blood (C1 esterase inhibitors), known as complements. They help to regulate various body functions (e.g., flow of body fluids in and out of cells). Hereditary angioedema type II,

[5]https://rarediseases.info.nih.gov/diseases/656/familial-transthyretin-amyloidosis

a more uncommon form of the disorder, occurs as the result of the production of abnormal complement proteins."[6]

Hereditary Spastic Paraplegia "The hereditary spastic paraplegias (HSP) are a large group of inherited neurologic disorders that share the primary symptom of difficulty walking due to muscle weakness and muscle tightness (spasticity) in the legs. There are more than 80 different genetic types of HSP. There may be significant variation in the severity of leg weakness (varying from none to marked), the degree of spasticity (varying from minimal to severe), and the occurrence of other neurologic symptoms between different genetic types of HSP; as well differences in the nature and severity of symptoms between individuals who have exactly the same genetic type of HSP."[7]

Inclusion Body Myositis "Sporadic inclusion body myositis (sIBM) is an acquired progressive muscle disorder that becomes apparent during adulthood. The symptoms and progression of sIBM vary from one person to another. In most cases, sIBM is characterized by progressive weakness and degeneration (atrophy) of the muscles especially those of the arms and the legs. sIBM can progress to cause severe disability. The exact cause of the disorder is unknown and the muscle tissue of affected individuals shows both inflammatory and degenerative changes. sIBM is a complex disorder and, most likely, multiple factors including genetic, immunological and environmental ones in combination all play a role in its development."[8]

Kallmann Syndrome "Kallmann syndrome (KS) is a rare genetic disorder in humans that is defined by a delay/absence of signs of puberty along with an absent/impaired sense of smell. A closely related disorder, normosmic idiopathic hypogonadotropic hypogonadism (nIHH), refers to patients with pubertal failure but with a normal sense of smell. Both KS and nIHH are due to an isolated deficiency of a key reproductive hormone called gonadotropin-releasing hormone (GnRH). KS and nIHH occurs in both sexes but males are more commonly diagnosed with this condition. Patients with KS/nIHH typically present at adolescence due to the delay in the onset of physical changes associated with puberty. KS patients are often aware of their lack of

[6] https://rarediseases.org/rare-diseases/hereditary-angioedema/
[7] https://rarediseases.org/rare-diseases/hereditary-spastic-paraplegia/
[8] https://rarediseases.org/rare-diseases/sporadic-inclusion-body-myositis/

sense of smell but most may not have sought medical advice for this symptom. While these reproductive symptoms predominate in their presentation, non-reproductive features that may be present in KS/nIHH subjects include: facial abnormalities (eg. cleft lip/palate), absence of one kidney, shortened digits, deafness, eye movement abnormality etc. [...] For therapy, initially, hormone replacement therapy (testosterone in males; estrogen and progesterone in females) is used to induce secondary sexual characteristics. [...] While both KS and nIHH are usually life-long in their nature, about 10-15% of patients may experience a recovery of their hormonal system, the reasons for which currently remain unclear. "[9]

Morgellons "Morgellons is a controversial and poorly understood condition in which unusual thread-like fibers appear under the skin. The patient may feel like something is crawling, biting, or stinging all over. Some medical experts say Morgellons is a physical illness. Others suggest it is a type of psychosis called 'delusional parasitosis,' in which a person thinks parasites have infected their skin. Your doctor may call it an 'unexplained dermopathy,' which means a skin condition that occurs without a known reason. Other medical professionals have dubbed the condition 'fiber disease.'"[10]

Multifocal Motor Neuropathy "Multifocal motor neuropathy (MMN) is a rare neuropathy characterized by progressive, asymmetric muscle weakness and atrophy (wasting). Signs and symptoms include weakness in the hands and lower arms; cramping; involuntary contractions or twitching; and atrophy of affected muscles. MMN is thought to be due to an abnormal immune response, but the underlying cause is not clear. Most people treated with intravenous immune globulin (IVIG) have rapid improvement in weakness, but maintenance IVIG is usually required for sustained improvement."[11]

Ocular Cicatricial Pemphagoid "Ocular cicatricial pemphigoid (OCP) is a form of mucous membrane pemphigoid (a group of rare, chronic autoimmune disorders) that affects the eyes. In the early stages, people with OCP generally experience chronic or relapsing conjunctivitis that is often characterized by tearing, irritation, burning, and/or mucus drainage. If left

[9] https://rarediseases.org/rare-diseases/kallmann-syndrome/
[10] https://www.webmd.com/skin-problems-and-treatments/morgellons-disease-what-is-it#1
[11] https://rarediseases.info.nih.gov/diseases/11011/multifocal-motor-neuropathy

untreated, OCP can progress to severe conjunctiva scarring and vision loss. Involvement of other mucosal sites and the skin may also occur in OCP. The exact underlying cause is currently unknown. The treatment of OCP aims to slow disease progression and prevent complications. This usually involves long-term use of medications called immunomodulators which help regulate or normalize the immune system."[12]

Osteonecrosis "Osteonecrosis, also known as avascular necrosis, is a disease resulting from the temporary or permanent loss of the blood supply to the bones. Without blood, the bone tissue dies and causes the bone to collapse. If the process involves the bones near a joint, it often leads to collapse of the joint surface. This disease also is known, aseptic necrosis, and ischemic bone necrosis. Although it can happen in any bone, osteonecrosis most commonly affects the ends (epiphysis) of long bones such as the femur, the bone extending from the knee joint to the hip joint. Other common sites include the upper arm bone, knees, shoulders, and ankles. The disease may affect just one bone, more than one bone at the same time, or more than one bone at different times. Orthopaedic doctors most often diagnose the disease. The amount of disability that results from osteonecrosis depends on what part of the bone is affected, how large an area is involved, and how effectively the bone rebuilds itself. The process of bone rebuilding takes place after an injury as well as during normal growth. Normally, bone continuously breaks down and rebuilds–old bone is reabsorbed and replaced with new bone. The process keeps the skeleton strong and helps it to maintain a balance of minerals. In the course of osteonecrosis, however, the healing process is usually ineffective and the bone tissues break down faster than the body can repair them. If left untreated, the disease progresses, the bone collapses, and the joint surface breaks down, leading to pain and arthritis."[13]

Systemic Capillary Leak Syndrome "Systemic capillary leak syndrome (SCLS) is a rare acquired disorder characterized by acute and severe recurrent attacks associated with a rapid fall in blood pressure. Attacks often last several days and require emergency care. They are sometimes fatal. SCLS occurs most often in adults and the disease is very rare in children. SCLS is not hereditary. Symptoms of systemic capillary leak syndrome classically involve a

[12] https://rarediseases.info.nih.gov/diseases/8759/ocular-cicatricial-pemphigoid
[13] https://rarediseases.org/rare-diseases/osteonecrosis/

brief warning, which can include nasal congestion and cough that can be mistaken for a viral upper respiratory infection. Patients develop malaise, nausea, lightheadedness, a faint feeling, abdominal pain headache and swelling of the extremities. There is a characteristic absence of fever, chills, rash, or signs of infection. Overt prostration and fainting may occur."[14]

Wilson Disease "Wilson disease is a rare genetic disorder characterized by excess copper stored in various body tissues, particularly the liver, brain, and corneas of the eyes. The disease is progressive and, if left untreated, it may cause liver (hepatic) disease, central nervous system dysfunction, and death. Early diagnosis and treatment may prevent serious long-term disability and life threatening complications. Treatment is aimed at reducing the amount of copper that has accumulated in the body and maintaining normal copper levels thereafter. Wilson disease [begins with] liver dysfunction where damage begins by six years of age, but usually presents clinically in teenage years or early twenties. Common signs of associated liver disease include a yellow discoloration (jaundice) of the skin, mucous membranes and the membranes (sclera) that line the eye, swelling (edema) of the legs and abdomen (ascites) due to abnormal retention of fluid, presence of abnormal blood vessels in the esophagus that may bleed (esophageal varices), a tendency for bruising and prolonged bleeding, and excessive tiredness (fatigue). Some individuals with Wilson disease may have only abnormalities of liver function test and may show no other symptoms until many years later."[15]

[14] https://rarediseases.org/rare-diseases/systemic-capillary-leak-syndrome/
[15] https://rarediseases.org/rare-diseases/wilson-disease/

Appendix G

List of Abbreviations

AFM	L'Association Français Contre Les Myopathies
ALIBER	Ibero-American Rare Disease Alliance
ARC	Asynchronous Remote Communities
AUC-PR	Area Under Precision-Recall Curve
AUC-ROC	Area Under the Receiver Operating Characteristic Curve
CBPR	Community-Based Participatory Research
CORD	Canadian Organization for Rare Disorders
CPC	Cost per Click
CPR	Cost per Respondent
CTR	Click through Rate
DT	Decision Trees
EURORDIS	European Organization for Rare Diseases
FGB	Functional Gradient Boosting
HCI	Human Computer Interaction
I-ORD	Indian Organization for Rare Diseases

IRB Institutional Review Board

IU Indiana University

LR Logistic Regression

LSVM Linear Support Vector Machine

NB Naïve Bayes

NHS National Health Service

NIH National Institutes of Health

NN Nearest Neighbors

NORD National Organization of Rare Diseases

ODA Orphan Drug Act

RCT Randomized Control Trial

RDI Rare Diseases International

RVA Rare Voices Australia

SMOTE Synthetic Minority Oversampling Technique

Soft-FGB Soft-Margin Functional Gradient Boosting

UK United Kingdom

US United States

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EDUCATION

June 2018 — **PhD in Informatics (Concentration in Health Informatics)**
Indiana University (Bloomington, IN, USA)
Minor: Human Computer Interaction Design
Advisors: Kay Connelly & Katie Siek

2015 — **MS in Human Computer Interaction Design**
Indiana University (Bloomington, IN, USA)
Advisors: Kay Connelly & Katie Siek

2013 — **Bachelor of Commerce**
University of Calgary (Calgary, AB, CAN)
Minor: Computer Science
Advisor: Tony Tang

RESEARCH EXPERIENCE

2017–2018 — UX Researcher (Contingent Worker)
Pro Unlimited on behalf of **Facebook** (Remote)
Manager: Anand Tharanathan

2017 — UX Research Intern
Facebook (Menlo Park, CA, USA)
Mentor: Julia Mayer

2016 — Research Intern
Microsoft Research (Redmond, WA, USA)
Mentors: Ed Cutrell & Meredith Ringel Morris

2015 — HCI Research Intern
PARC, A Xerox Company (Webster, NY, USA)
Mentor: Karen Braun

2013–2015 — Research Assistant
PHIT Lab, Indiana University (Bloomington, IN, USA)
Advisor: Kay Connelly

2012–2013 — Research Assistant
Interactions Lab, University of Calgary (Calgary, AB, CAN)
Advisor: Tony Tang

TEACHING EXPERIENCE

2016–2017	Teaching Assistant, Department of Computer Science DePauw University (Greencastle, IN, USA) **Bridge to Informatics** (Winter 2016, Winter 2017)
2014–2017	Teaching Assistant, School of Informatics & Computing Indiana University (Bloomington, IN, USA) **Advanced Prototyping** (Fall 2017) **Pervasive & Mobile Design** (Fall 2016) **Introduction to Health Informatics** (Fall 2014, Fall 2015) **Mobile Application Development** (Spring 2014)
2012	Teaching Assistant, Haskayne School of Business University of Calgary (Calgary, AB, CAN) **Intro. to Management Information Systems** (Winter 2012)

SERVICE

Mentoring & Advising

2017–2018	Gustave Ekobe	Undergraduate Research Opportunities in Computing
2016–2017	Vanessa Pereira	Emerging Scholars Research for Undergraduate Women
2016–2017	Jasmine Mcclain	Emerging Scholars Research for Undergraduate Women
2015–2016	Grace Bastin	Emerging Scholars Research for Undergraduate Women
2015	Katie Spoon	Emerging Scholars Research for Undergraduate Women
2015	Krish Puri	Undergraduate Research Opportunities in Computing
2015	Gleb Alexeev	Undergraduate Research Opportunities in Computing
2014–2015	Alec Byrd	Senior Thesis
2014	Taha Merghani	HBCU Summer Scholar's Institute
2014	Danika Geisler	Summer Research Experiences for Undergraduates

Conference Program Committees

2019	ACM CHI Health Subcommittee (Associate Chair)
2017	ACM DIS Specific Application Areas Subcommittee (Associate Chair)
2016, 2018	ACM CHI Late Breaking Work (Associate Chair)

Conference Organizing Committees

2018, 2019	CHI Student Volunteer Co-Chair
2017	Indiana Women in Computing (InWIC) Sessions Committee Member
2016	Indiana Women in Computing (InWIC) Social Media Chair

Conference Student Volunteer

2013–2017	ACM Conference on Human Factors in Computing Systems (CHI)
2013	GRAND Network Centres of Excellence Meeting (GRAND)

Reviewing

ACII, AMIA, CHI, CSCW, DIS, EAI Transactions, iConference, Graphics Interface, GRAND, IEEE Access, IU GPSG Awards, JMIR, MobileHCI, Pervasive Health

Community Service

2016–2018 **Graduate & Professional Student Government (GPSG)**
Indiana University (Bloomington, IN, USA)

- Benefits Officer (2017)
- Student Academic Appointments Review Board (2016–2018)
- Assembly Representative for Informatics (2016)
- Benefits Committee Member (2016)

2014–2017 **Graduate Informatics Students Association (GISA)**
Indiana University (Bloomington, IN, USA)

- Institutional Voice Chair (2015–2017)
- Informatics Grad. Program Committee Student Rep. (2015–2017)
- Prospective Student Visit Weekend Organizing Committee (2015)
- Graduate Student Health & Wellness Committee (2014–2015)
- Student Ambassador (2014–2017)

STEM Education Programs

2017	Program Partner for Tech-xpert classes at WonderLab Museum (Bloomington, IN, USA)
2015–2017	Research Mentor for the Center of Excellence for Women in Technology (Bloomington, IN, USA)
2014–2018	Research Mentor for IU's Undergraduate Research Opportunities in Computing program (Bloomington, IN, USA)
2014	Research Mentor for the HBCU Summer Scholar's Institute (Bloomington, IN, USA)
2013	Assistant Instructor for the CEMC Workshop in Computer Science for Young Women (Calgary, AB, CAN)
2010–2012	VP Communications for the University of Calgary's Women in Science & Engineering student group (Calgary, AB, CAN)
2009-2011	Mentor with the Cybermentor organization (Calgary, AB, CAN)
2009-2010	Assistant Instructor for the Explore IT workshop (Calgary, AB, CAN)

AWARDS & RECOGNITIONS

Research Funding

2017	Rare Disease Foundation: Investigating a Recommender System Approach to Tangible Support for People with Rare Diseases. $2,450. (PI: **Haley MacLeod, with Kay Connelly and Katie Siek**).

Best Paper Awards & Nominations

2017	ACM CHI 2017 Best Paper Nomination
2015	EAI Pervasive Health 2015 Best Paper Nomination
2015	ACM CHI 2015 Best Paper Nomination
2014	WISH 2014 Best Paper Award
2014	ACM CHI 2014 Best Paper Nomination

Travel Awards

2017	NSF Smart & Connected Health Outreach Event Travel Award
2016	GPSG Travel Award
2016	IU School of Informatics & Computing InWIC 2016 Travel Grant
2016	NSF Student Travel Award for TAPIA 2016
2016	NSF Student Travel Award for IEEE CHASE 2016
2016	IU Provost's Travel Award for Women in Science
2016	ACM Travel Award for CHI 2016
2014	IU School of Informatics & Computing Grace Hopper 2014 Travel Grant
2014	Student Travel Award for AMIA 2014

Other Awards

2017	IU School of Informatics & Computing Cross-Track Collaboration Award
2015–2016	IU School of Informatics & Computing Associate Instructor of the Year
2008	University of Calgary Admission Scholarship
2008	University of Calgary Dean's Merit Admission Award
2005–2008	Alexander Rutherford Scholarship

PUBLICATIONS

Journal Papers

[J1] Haley MacLeod, Ben Jelen, Annu Prabhakar, Lora Oehlberg, Katie Siek, and Kay Connelly. 2017. A Guide to Using Asynchronous Remote Communities (ARC) for Researching Distributed Populations. *EAI Endorsed Transactions on Pervasive Health and Technology* 17, 11 (7 2017). DOI: http://dx.doi.org/10.4108/eai.18-7-2017.152898

Conference Papers

[C11] Juan Maestre, Haley MacLeod, Ciabhan Connelly, Julia Dunbar, Jordan Beck, Katie Siek, Patrick Shih. 2018. Defining Through Expansion: Conducting Asynchronous Remote Communities (ARC) Research with Stigmatized Groups. In Proceedings of the 2018 CHI Conference on Human Factors in Computing Systems (CHI '18). ACM, New York, NY, USA,

[C10] Fateme Rajabiyadi, Charles Perin, Jo Vermeulen, Haley MacLeod, Diane Gromala, and Sheelagh Carpendale. 2017. Differences That Matter: In-Clinic Communication Challenges. In Proceedings of the 11th EAI International Conference on Pervasive Computing Technologies for Healthcare (PervasiveHealth '17). (Acceptance: 25%)

[C9] Haley MacLeod, Cynthia L. Bennett, Meredith Ringel Morris, and Edward Cutrell. 2017. Understanding Blind People's Experiences with Computer-Generated Captions of Social Media Images. In Proceedings of the 2017 CHI Conference on Human Factors in Computing Systems (CHI '17). ACM, New York, NY, USA, 5988–5999. DOI: http://dx.doi.org/10.1145/3025453.3025814. (Acceptance: 25%)

[C8] Haley MacLeod, Grace Bastin*, Leslie S. Liu, Katie Siek, and Kay Connelly. 2017. "Be Grateful You Don't Have a Real Disease": Understanding Rare Disease Relationships. In Proceedings of the 2017 CHI Conference on Human Factors in Computing Systems (CHI '17). ACM, New York, NY, USA, 1660–1673. DOI: http://dx.doi.org/10.1145/3025453.3025796. (Acceptance: 25%)

[C7] Annu Sible Prabhakar, Lucia Guerra-Reyes, Vanessa M. Kleinschmidt, Ben Jelen, Haley MacLeod, Kay Connelly, and Katie A. Siek. 2017. Investigating the Suitability of the Asynchronous, Remote, Community-based Method for Pregnant and New Mothers. In Proceedings of the 2017 CHI Conference on Human Factors in Computing Systems (CHI '17). ACM, New York, NY, USA, 4924–4934. DOI: http://dx.doi.org/10.1145/3025453.3025546. (Acceptance: 25%) – **Honourable Mention Award** (top 5%).

[C6] Haley MacLeod, Shuo Yang, Kim Oakes, Kay Connelly, and Sriraam Natarajan. 2016. Identifying Rare Diseases from Behavioural Data: A Machine Learning Approach. In Connected Health: Applications, Systems and Engineering Technologies (CHASE), 2016 IEEE First International Conference on. IEEE, 130–139. DOI: http://dx.doi.org/10.1109/CHASE.2016.7. (Acceptance: 23%).

[C5] Haley MacLeod, Ben Jelen, Annu Prabhakar, Lora Oehlberg, Katie Siek, and Kay Connelly. 2016. Asynchronous Remote Communities (ARC) for Researching Distributed Populations. In Proceedings of the 10th EAI International Conference on Pervasive Computing Technologies for

*Co-author is a trainee

Healthcare (PervasiveHealth '16). ICST (Institute for Computer Sciences, Social-Informatics and Telecommunications Engineering), ICST, Brussels, Belgium, Belgium, 1–8. (Acceptance 35%).

[C4] Kim Oakes, Katie A. Siek, and Haley MacLeod. 2015. MuscleMemory: Identifying the Scope of Wearable Technology in High Intensity Exercise Communities. In Proceedings of the 9th International Conference on Pervasive Computing Technologies for Healthcare (PervasiveHealth '15). ICST (Institute for Computer Sciences, Social-Informatics and Telecommunications Engineering), ICST, Brussels, Belgium, Belgium, 193–20. (Acceptance: 30%) – **Best Paper Nomination.**

[C3] Haley MacLeod, Kim Oakes, Danika Geisler*, Kay Connelly, and Katie Siek. 2015. Rare World: Towards Technology for Rare Diseases. In Proceedings of the 33rd Annual ACM Conference on Human Factors in Computing Systems (CHI '15). ACM, New York, NY, USA, 1145–1154. DOI: http://dx.doi.org/10.1145/2702123.2702494. (Acceptance: 23%) – **Honourable Mention Award** (top 5%).

[C2] Maayan Cohen, Kody R. Dillman, Haley MacLeod, Seth Hunter, and Anthony Tang. 2014. OneSpace: Shared Visual Scenes for Active Freeplay. In Proceedings of the SIGCHI Conference on Human Factors in Computing Systems (CHI '14). ACM, New York, NY, USA, 2177–2180. DOI: http://dx.doi.org/10.1145/2556288.2557117. (Acceptance: 22.8%) - **Honourable Mention Award** (top 5%).

[C1] Haley MacLeod, Anthony Tang, and Sheelagh Carpendale. 2013. Personal Informatics in Chronic Illness Management. In Proceedings of Graphics Interface 2013 (GI '13). Canadian Information Processing Society, Toronto, Ont., Canada, Canada, 149–156. (Acceptance 38%).

Extended Abstracts

[EA2] Haley MacLeod, Ben Jelen, Annu Prabhakar, Lora Oehlberg, Katie A. Siek, and Kay Connelly. 2016. Lessons Learned from Conducting Group-Based Research on Facebook. In Proceedings of the 2016 CHI Conference Extended Abstracts on Human Factors in Computing Systems (CHI EA '16). ACM, New York, NY, USA, 804–815. DOI: http://dx.doi.org/10.1145/2851581.285161

[EA1] Haley MacLeod and Anthony Tang. 2013. Towards Personal Informatics Tools for Chronic Illness Management. In Proceedings of GRAND Network Centres of Excellence Meeting. (Acceptance 53%).

Workshop Papers

[W7] Haley MacLeod, Jacob Abbott, Sameer Patil. Small Data Privacy Protection: An Exploration of the Utility of Anonymized Data of People with Rare Diseases. In Proceedings of the 2017 Workshop on Interactive Systems in Healthcare (WISH '17).

[W6] Julia C. Dunbar, Ciabhan L. Connelly, Juan F. Maestre, Haley MacLeod, Katie Siek, Patrick C. Shih. 2017. Considerations for Using the Asynchronous Remote Communities (ARC) Method in Health Informatics Research. In Proceedings of the 2017 Workshop on Interactive Systems in Healthcare (WISH '17).

*Co-author is a trainee

[W5] Haley MacLeod, Maia Jacobs, Katie Siek, Kay Connelly, and Elizabeth D. Mynatt. 2016. Ethical Considerations in Pervasive Health Research. In Pervasive Health 2016 Workshop on the Future of Pervasive Health.

[W4] Haley MacLeod, Kim Oakes, Kay Connelly, and Katie Siek. 2015. Methods for Studying Rare Disease Populations. In CHI 2015 Workshop on Advancing Health and Wellness Technology Research in Home and Community Settings.

[W3] Haley MacLeod, Denique Ferguson, Scott Trepper, and Steve Layton. 2014. Better Bites: Healthy Food Within A Budget. In Proceedings of the 2014 Workshop on Interactive Systems in Healthcare (WISH '14) – **Best Paper Award.**

[W2] Haley MacLeod and Anthony Tang. 2013. Shared Displays to Facilitate Discussions Surrounding Patient Data. In Proceedings of the CHI 2013 Workshop on Patient-Client Communication.

[W1] Haley MacLeod and Anthony Tang. 2012. Understanding Personal Informatics Needs of Individuals with Chronic Health Conditions. In Proceedings of the 2012 Workshop on Interactive Systems in Healthcare (WISH '12).

Doctoral Consortia

[DC3] Haley MacLeod. 2016. Design and Development of a Sociotechnical Intervention for People with Rare Diseases. In Proceedings of the 2016 ACM Richard Tapia Celebration of Diversity in Computing (TAPIA '16).

[DC2] Haley MacLeod. 2016. Rare World: Designing for Dispersed Populations with Rare Diseases. In Proceedings of the 2016 CHI Conference Extended Abstracts on Human Factors in Computing Systems (CHI EA '16). ACM, New York, NY, USA, 229–233. DOI: http://dx.doi.org/10.1145/2851581.2859026.

[DC1] Haley MacLeod. 2014. Rare World: Designing for Dispersed Populations with Rare Diseases. In Proceedings of the AMIA 2014 Doctoral Consortium on Sociotechnical Issues in Biomedical Informatics (AMIA '14).

Posters

[P12] Haley MacLeod, Jacob Abbott, Sameer Patil. Small Data Privacy Protection: An Exploration of the Utility of Anonymized Data of People with Rare Diseases. Poster presented at WISH 2017.

[P11] Haley MacLeod. 2017. Rare World: Designing for Dispersed Populations with Rare Diseases. Poster presented at NSF Smart & Connected Health Meeting.

[P10] Haley MacLeod, Shuo Yang, Kim Oakes, Kay Connelly, and Sriraam Natarajan. 2016. Identifying Rare Diseases from Behavioural Data: A Machine Learning Approach. Poster presented at InWIC 2016.

[P9] Haley MacLeod. 2016. Rare World: Designing for Dispersed Populations with Rare Diseases. Poster presented at CHI 2016.

[P8] Grace Bastin, Haley MacLeod, Leslie S. Liu, Katie Siek, and Kay Connelly. 2015. "At Least You Don't Have a Real Disease": Understanding Rare Disease Relationships. Poster presented at CEWIT Women's Research Poster Competition 2016.

[P7] Haley MacLeod. 2015. Rare World: Designing for Dispersed Populations with Rare Diseases. Poster presented at CEWIT Women's Research Poster Competition 2016.

[P6] Haley MacLeod. 2016. Rare Diseases. Poster presented at GISA Un-poster Session 2016.

[P5] Haley MacLeod. 2015. Technology for Rare Diseases. Poster presented at GISA Un-poster Session 2015 – **Most Human-Oriented Award**.

[P4] Haley MacLeod. 2014. Design and Development of a Sociotechnical Intervention for People with Rare Diseases. Poster presented at WISH 2014.

[P3] Haley MacLeod and Kay Connelly. 2014. Developing Mobile Self-Monitoring Support for Healthy Me. Poster presented at InWIC 2014.

[P2] Haley MacLeod, and Anthony Tang. 2012. Understanding Personal Informatics Needs of Individuals with Chronic Health Conditions. Poster presented at WISH 2012.

[P1] Haley MacLeod, and Anthony Tang. 2012. Understanding Personal Informatics Needs of Individuals with Chronic Health Conditions. Poster presented at the W21C 2012 International Innovation Forum.

Invited Talks

[IT3] Asynchronous Remote Communities (ARC) for Researching Distributed Populations. 2017. Invited talk for W21C Journal Club in Calgary, AB, Canada.

[IT2] Whose Life Is It Anyways? Empowering Personal Decision Making in Health. 2015. Keynote speaker for the ANT+ Alliance Symposium in Banff, AB, Canada.

[IT1] Rare World: Towards Technology for Rare Diseases. 2015. Invited talk for the Interactions Lab at the University of Calgary in Calgary, AB, Canada.

Conference Presentations

[CP8] Understanding Blind People's Experiences with Computer-Generated Captions of Social Media Images. 2017. Presented at CHI 2017.

[CP7] "Be Grateful You Don't Have a Real Disease": Understanding Rare Disease Relationships. 2017. Presented at CHI 2017.

[CP6] Identifying Rare Diseases from Behavioural Data: A Machine Learning Approach. 2016. Presented at CHASE 2016.

[CP5] Asynchronous Remote Communities (ARC) for Researching Distributed Populations. 2016. Presented at Pervasive Health 2016.

[CP4] Lessons Learned from Conducting Group-Based Research on Facebook. 2016. Presented at CHI 2016.

[CP3] Rare World: Towards Technology for Rare Diseases. 2015. Presented at CHI 2015.

[CP2] OneSpace: Shared Visual Scenes for Active Freeplay. 2014. Presented at CHI 2014.

[CP1] Personal Informatics in Chronic Illness Management. 2013. Presented at Graphics Interface 2013.

Panels

[P3] Grace Bastin, Katherine Metcalf, Joyce Main, and Haley MacLeod. Exemplary Research in Computing. Panel presented at INWiC 2017.

[P2] Ray Maker, Jon Bruner, Paul Dorotich, and Haley MacLeod 2015. The Quantified Self: Capturing, Connecting and Interpreting. Panel presented at the ANT+ Alliance Symposium 2015.

[P1] Jennifer Bernhard, Tiffany Trusty, Haley MacLeod, and Priyanka Brunese. 2014. Graduate School Panel Session. Panel presented at InWIC 2014.

Demonstrations

[D1] Kody Dillman, Haley MacLeod, David Ledo, Bon Adriel Aseniero, Sebastian Boring and Anthony Tang. 2013. OneSpace: Seeing Oneself in a Shared Mirror for Play and Learning. Demonstration given at GRAND 2013.

Workshop Presentations

[WP11] Rare World: Investigating Social Support in Rare Disease and Common Chronic Illness Communities. 2017. Presented at the NSF Smart & Connected Health Meeting.

[WP10] Rare World: Designing for Dispersed Populations with Rare Diseases. 2016. Presented at the TAPIA 2016 Doctoral Consortium.

[WP9] Ethical Considerations for Pervasive Health Research. 2016. Presented at the Pervasive Health 2016 Workshop on the Future of Pervasive Health.

[WP8] Rare World: Designing for Dispersed Populations with Rare Diseases. 2016. Presented at the CHI 2016 Doctoral Consortium.

[WP7] Methods for Studying Rare Disease Populations. 2015. Presented at the CHI 2015 Workshop on Advancing Health & Wellness Technology Research in Home and Community Settings.

[WP6] Design and Development of a Sociotechnical Intervention for People with Rare Diseases. 2014. Presented at the AMIA 2014 Doctoral Consortium on Sociotechnical Issues in Biomedical Informatics.

[WP5] Better Bites: Healthy Food within a Budget. 2014. Presented at the Workshop on Interactive Systems in Healthcare (WISH) 2014.

[WP4] Developing Mobile Self-Monitoring Support for Healthy Me. 2014. Presented at Consumer Health Informatics in the Midwest (CHIM) 2014.

[WP3] Towards Personal Informatics Tools for Chronic Illness Management. 2013. Presented at GRAND 2013.

[WP2] Shared Displays to Facilitate Discussions Surrounding Patient Data. 2013. Presented at CHI 2013 Workshop on Patient-Clinician Communication.

[WP1] Understanding Personal Informatics Needs of Individuals with Chronic Health Conditions. 2012. Presented at the Workshop on Interactive Systems in Healthcare 2012.