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Exploring the role of digital tools in rare disease management: An interview-based study

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Abstract

While digital tools, such as the Internet, smartphones, and social media, are an important part of modern society, little is known about the specific role they play in the healthcare management of individuals and caregivers affected by rare disease. Collectively, rare diseases directly affect up to 10% of the global population, suggesting that a significant number of individuals might benefit from the use of digital tools. The purpose of this qualitative interview-based study was to explore: (a) the ways in which digital tools help the rare disease community; (b) the healthcare gaps not addressed by current digital tools; and (c) recommended digital tool features. Individuals and caregivers affected by rare disease who were comfortable using a smartphone and at least 18 years old were eligible to participate. We recruited from rare disease organizations using purposive sampling in order to achieve a diverse and information rich sample. Interviews took place over Zoom and reflexive thematic analysis was utilized to conceptualize themes. Eight semistructured interviews took place with four individuals and four caregivers. Three themes were conceptualized which elucidated key aspects of how digital tools were utilized in disease management: (1) digital tools should lessen the burden of managing a rare disease condition; (2) digital tools should foster community building and promote trust; and (3) digital tools should provide trusted and personalized information to understand the condition and what the future may hold. These results suggest that digital tools play a central role in the lives of individuals with rare disease and their caregivers. Digital tools that centralize trustworthy information, and that bring the relevant community together to interact and promote trust are needed. Genetic counselors can consider these ideal attributes of digital tools when providing resources to individuals and caretakers of rare disease.

KEYWORDS
caregivers, complex disease, digital tools, disability, parents, rare disease

1 | INTRODUCTION

Digital tools, such as the Internet, smartphones, and social media, play an important role in modern society. The Pew Research Center estimates that 93% of the US adults use the Internet and 85% own a smartphone (Pew Research Center, 2021a, 2021b, 2021c). In addition, the use of social media outlets (such as Facebook and X, formerly known as Twitter) has grown. As of the end of 2021, the number of daily active Facebook users was 1.93 billion worldwide, which is almost 25% of the world's population ("Meta 2021 Q4 Results Conference Call," 2022). For the same time period, the number of daily active X users was 920 million ("X Q1 Earnings Release," 2022). Moreover, for many users, using social media is part of their daily routine. Seven in ten Facebook users and five in ten X users visit these sites at least once a day (Pew Research Center, 2021a, 2021b, 2021c). From these numbers, it is clear that most people actively use digital tools in their everyday lives.

Rare diseases are by nature low prevalence – in the United States, a rare disease is defined as one affecting fewer than 200,000 people. However, low prevalence does not mean low impact. There are approximately 8000 rare diseases that collectively directly affect between 6% and 10% of the global population (Zurynski et al., 2008). According to a 2020 analysis of the Orphanet database, there are over 400 million people worldwide living with rare disease at any given time (Nguengang Wakap et al., 2020).

Both individuals with rare diseases (IRD) and caregivers of individuals with rare diseases (CRD) are affected by the "burden of care." As Pelentsov and colleagues write in a 2015 scoping review, for parents of a child with a rare disease, the burden of care spans many years and involves a lifetime commitment. It often requires a change in work patterns, income, and domestic responsibilities (Pelentsov et al., 2015). In a systematic review focusing on IRD, key themes included the physical limitations and psychological impact of dealing with their condition, how they dealt with social stigma and lack of social support, and lack of knowledge they faced in the healthcare system about their condition (von der Lippe et al., 2017).

There is current literature focused on examining certain aspects of digital tool usage by the rare disease community, though most studies focused only on caregivers of individuals with rare diseases. Studies have examined how parents of children with rare conditions use the Internet to find information about their children's rare conditions (Barton et al., 2019; Bouwman et al., 2010; Deuitch et al., 2021; Litzkendorf et al., 2020; Nicholl et al., 2017; Tozzi et al., 2013). These studies examine the positive aspects, such as convenience and cost-effectiveness, as well as the negative aspects, such as the overwhelming and sometimes dubious nature of the information. In this context, searching for information can help CRD gain a sense of coherence to help them cope with their situation. Becoming knowledgeable about a condition is essential for managing a situation that initially seems overwhelming and distressful.

Other studies highlight the social and emotional aspects of parents using social media, including Internet support groups, to help deal with their children's medical management journeys, and to feel less isolated by finding a community. Key activities include exchanging information, encountering emotional support, sharing experiences, helping others, and deriving amusement (Cacioppo et al., 2016; DeHoff et al., 2016; Deuitch et al., 2021; Tozzi et al., 2013; van Uden-Kraan et al., 2008).

Thus, it is reasonable to assume that individuals in the rare disease community, including both IRD and CRD, often turn to digital tools for their informational, emotional, and logistical needs when managing their care. Logistically, digital tools can be used for care coordination, such as telehealth appointments and remote follow up for individuals under care. In terms of information gathering, digital tools can be important because medical information on rare disease conditions are often scarce. Moreover, feelings of loneliness and isolation are often present in both individuals with rare diseases and their caregivers. Thus, it is important to examine the positive and negative aspects of digital tools in rare disease management, as well as any healthcare gaps not addressed by currently available digital tools.

In this study, we examine the role that digital tools play in rare disease management. Through interviews with both IRD and CRD, we sought to explore the following: (a) ways in which digital tools help the IRD and CRD experience; (b) healthcare gaps not addressed by current digital tools; and (c) recommendations for features in a perfect digital tool. Our hope is that relevant parties, such as rare disease organizations and product developers, could utilize the knowledge gained in this research to provide better digital tools that would improve the IRD and CRD experience; and that genetic counselors can consider relevant digital tool features when providing resources to individuals with rare disease or their caretakers.

What is known about this topic

There is current literature focused on examining certain aspects of digital tool usage by the rare disease community, though most studies focused on caregivers of individuals with rare diseases instead of individuals with rare diseases. To our knowledge, there is no literature focused on holistic digital tool usage by the rare disease community.

What this paper adds to the topic

Our study fills gaps in the literature by including perspectives of both caregivers and individuals with rare diseases and by employing a user research perspective to identify features of "the perfect digital tool" for the rare disease community. Digital tools have a role in addressing informational support, emotional support, and community building.

2 | MATERIALS AND METHODS

We used an interview-based qualitative study design to explore the role that digital tools play in rare disease management.

Qualitative methodology is ideal for this purpose, as it is a way for gaining deeper insight into people's experiences and for seeking to understand the meaning or nature of these experiences (Corbin & Strauss, 2014).

The UCLA Institutional Review Board approved all aspects of this study (Protocol IRB-21-001547).

2.1 | Sample, recruitment, and procedures

We decided to partner with rare disease organizations in the recruitment phase because reaching individuals in the rare disease community can be difficult – due to the fact that there is a low prevalence of conditions, most individuals of the community are geographically dispersed (Griggs et al., 2009; Yu et al., 2020). The four rare disease organizations (Undiagnosed Diseases Network Facebook Group, United Leukodystrophy Foundation, Rare New England, and Global Genes) were chosen because of their work with their respective rare disease communities, and because of their willingness to allow recruitment materials to be distributed to their members.

We chose to use social media to recruit interview participants from these organizations because these organizations utilize social media as a primary vehicle of engagement with members. In addition, IRD and CRD tend to engage in the same social media platforms to connect with others and learn strategies for disease management; thus, utilizing social media can be effective in reaching them (Close et al., 2013; Miller et al., 2021; Schumacher et al., 2014).

We used a purposive sampling approach to recruit participants from the aforementioned four rare disease organizations. Purposive sampling was utilized because we wanted to achieve a diverse and information-rich sample of interviewees. Thus, between October 2021 and March 2022, the research team posted information about the research study with a link to an online eligibility screening survey via the rare disease organizations' social media or the first author's X account. Inclusion criteria included caregivers and individuals affected by rare disease who owned a smartphone. Exclusion criteria included anyone who was under 18 years of age, those not comfortable using Zoom, and those unable to participate in the interview in English.

Individuals who were interested completed the survey, and interview invitations were sent out based on recency criteria (individuals who filled out the screening survey first were considered first), as well as diversity criteria (with a goal of equal numbers within each criterion): rare disease organization, caregiver/individual, ethnicity, diagnosed/undiagnosed, gender, and condition phenotype.

In preparation for the interview, participants were given a digital diary template, in which they were asked to note how they interacted with digital tools during a "typical" week in their lives. Information given in the digital diary was utilized by the interviewer during the interview. A copy of the digital diary template is available as a supplemental file. Interviews were conducted by AC and took place over Zoom in English during the period from October 2021 to March 2022. At the beginning of each interview, the interviewer reviewed the purpose of the study and that it was being conducted by a genetic counseling graduate student as part of program requirements. No relationship was established prior to the interview. Present during the interview were the interviewer and the participant. Participants did not review their transcripts, nor did they provide feedback on the research findings. An e-gift card of $25 was given to each participant who completed the interview.

2.2 | Instrumentation

We constructed a semistructured interview guide based on published principles for user research from a business perspective (Fitzpatrick, 2013), as well as research team experience and expertise in rare disease, digital tools, and interview guide development. Questions focused on three main topics: information gathering, social/emotional support, and logistics.

Interviewees were asked questions about specific features of existing digital tools that they liked and did not like, as well as details about their "perfect" digital tool. A copy of the semistructured interview guide is available as a File S1.

2.3 | Analysis

Interviews were audio-recorded, transcribed, and the transcriptions were then uploaded into Dedoose for data management. Data were analyzed inductively using reflexive thematic analysis, an approach to conceptualize or construct themes across the dataset (Braun & Clarke, 2022). We used reflexive thematic analysis because this approach acknowledges that themes are produced (constructed) at the intersection of the data and research team members' positionalities, which in this research included gender diversity, expertise in business (AC, DB, JS) and product development (AC), rare disease (AC, JS, CP), qualitative methods (NG), as well as genetic counseling backgrounds and interest (AC, JS, CP, SDH, NG). Two research members (AC and SDH; overseen by NG) independently reviewed the transcripts to familiarize themselves with the data and to develop codes, then met to review and discuss discrepancies, achieving 100% coding consensus. Coding consensus is not considered relevant within reflexive thematic analysis (Braun & Clarke, 2022), but the exercise was valuable for facilitating thorough data engagement and for developing codes fine-grained enough to assess research-relevant meaning from the dataset (Braun & Clarke, 2023). This iterative process continued until no new codes were created from the dataset.

The codes were then categorized into categories, which then were analyzed to construct themes (central organizing concepts) and subthemes. The research team met regularly to review and discuss the analysis process.

3 | RESULTS

3.1 | Sample characteristics

A total of 790 individuals completed the screening survey; 671 were excluded from consideration because we deemed them to be fraudulent accounts, 119 received an interview invite, and 14 interviews were conducted. Of these 14 interviews, 6 were deemed fraudulent and removed from analysis; ultimately, 8 interviews were included in the analysis. Interviews lasted on average 40 min (min = 25 min, max = 47 min).

We adopted several approaches to address the challenges we faced regarding fraudulent accounts during the recruitment process (Levi et al., 2022; Pratt-Chapman et al., 2021). First, we flagged all suspicious email addresses (e.g., uncommon account providers or flagrant mismatch between the name and email address). Second, we looked at the start and end times regarding survey submission. If the time to complete the survey was less than a minute, the surveys were deemed likely to have been completed by a bot and dropped from consideration (since the average length of time needed to complete the survey was estimated to be at least 2 min). Third, we looked at the IP address to see if they repeated, or had been spoofed (where the source address has been modified to hide the identity of the sender). Fourth, we looked at the survey responses themselves, and any incomplete or incomprehensible submissions were dropped from consideration.

In the interview phase, we eliminated six interviews from consideration because we identified sufficient inconsistencies during the interview to question the authenticity of the participant and their eligibility to participate in the study. One example was when an individual mentioned they used PubMed journals in their research for rare diseases and (incorrectly) compared finding PubMed articles to be like "Google searches." This led the research team to conclude that the individual did not know what PubMed was, as Google searches are a no-cost method of searching, while PubMed is a literature citation database that links to articles that may be free or require a fee. In combination with other discrepancies, the research team decided to eliminate this interview from consideration.

We ultimately analyzed eight semistructured interviews in English with individuals from the rare disease community, including four individuals with a rare disease and four caregivers of individuals with a rare disease. Table 1 shows summary characteristics of these eight individuals. As the table shows, sample characteristics included: five diagnosed and three undiagnosed individuals, six women and two men, two individuals from each of the four rare disease organizations, four individuals with rare diseases and four caregivers, and seven white and one Hispanic individuals.

3.2 | Overview of themes

Three major themes and six subthemes (two subthemes within each major theme) were produced from our analysis; in addition, four recommendation themes were produced. These themes and recommendations were informed by a user research perspective and other aspects of research member's positionalities. Of note, all themes were produced from collective consideration of IRD and CRD transcripts; no themes unique to either IRD or CRD were constructed from the transcripts.

3.3 | Theme 1: Digital tools should lessen the burden of managing a rare disease condition

IRD and CRD spoke of being overwhelmed by managing a rare disease. Digital tools can help make their experience less burdensome.

3.3.1 | Subtheme 1: IRD and CRD constantly need to be proactive in managing the rare disease condition

One key component of the burden that IRD and CRD feel in managing their rare disease condition is the need to constantly be proactive in all aspects of management. IRD and CRD take it upon themselves to fit disease management tasks into their already busy and exhausting day-to-day lives. Over time, this can lead to suffering in other aspects in their lives, such as with their mental health, personal relationships, or professional success.

For example, one IRD discusses how to balance her work commitments and managing her disease in this way:

TABLE 1 Sample characteristics.

Rare disease organization	Patient/caregiver	Race	Diagnosed/undiagnosed	Gender	Condition phenotype
UDN:[a] 2	Patient: 4	Black: 0	Diagnosed: 5	Male: 2	Cardiology: 1
Rare NE: 2	Caregiver: 4	White: 7	Undiagnosed: 3	Female: 6	Orthopedics: 2
ULF:[b] 2		Hispanic: 1			Neurology: 2
Facebook:[c] 2		Asian: 0			Metabolic: 1
					Endocrinology: 1
					Leukodystrophy: 1

[a]Undiagnosed Disease Network Facebook Group.

[b]United Leukodystrophy Foundation.

[c]Participant declined to specify which organization's Facebook Group.

> So learning when to take a day, because for my work, I only get five sick days a year…So is this migraine going to be okay? Do I hold off? Do I take [the migraine medication]? It's like a game of chance every time.
>
> IRD #97

In addition, IRD and CRD spend a lot of time researching information, such as potential diagnoses or new therapeutics. These research tasks include using Google, asking for advice in support groups, or trying to gather information from experts.

> So what I did was I took those genes and I researched them, and connected with other families, mostly via Facebook. Literally it was just typing these genes into the Facebook search bar and then messaging strangers, and saying like, "I just read that you said that your child had whatever," and connecting with them. And kind of all of that led me to a geneticist who was able to diagnose her based on these variants of unknown significance.
>
> CRD #30

Another example of a key management task that IRD and CRD take on is advocating for themselves or their loved ones. This could be arguing with an insurance company to cover a certain medication or pushing doctors to get an accurate diagnosis.

One IRD comments on how she has to proactively gather information about herself to discuss with her doctor:

> I only go, I think it's once every 6 months to see my neurologist. So I'm trying to remember 6 months of stuff in a half hour meeting. So I know something is going to get missed. I can only do so much and with my migraines over the years, it's taken a toll on my short term memory. I have Post-it notes everywhere because I'm going to forget. So for me, it's writing down notes everywhere helps, but I can't bring a whole pack of sticky notes in here and be like, 'Hey, this and this'.
>
> IRD #97

One CRD spoke about how she had to proactively go to multiple providers with paperwork and advocate to get a test to confirm her daughter's diagnosis:

> For instance, my daughter, she was diagnosed with a condition that [our] Hospital tried to say that she didn't have. They gave her a test that wasn't even the test for that condition. Then I took her to a specialist that does specialize and they gave her the real test, and she did in fact have it…but then I had to bring that test to her other providers at [another] Hospital so then they would believe me because then that was in her medical [records] … And so there's no easy fix to that, other than to keep bringing your documentation with you everywhere you go, which is fine but that shouldn't be the responsibility of the patient.
>
> CRD #30

In response to the burdensome nature of dealing with a rare disease, IRD and CRD want digital tools that enable them to be more passive to save energy for other important things. Examples of features that enable IRD and CRD to be passive include alerts, such as when new research about a condition has been published, or announcements about relevant new events.

One IRD describes this:

> I mean, it would be really awesome if there was a website or app where everything was all tied together. I don't know if that even makes sense but having all of your medications, your support groups, messages, if everything was intertwined, in a perfect world that would be great.
>
> IRD #98

3.3.2 | Subtheme 2: Managing everything in different places is emotionally and logistically taxing

Another way that IRD and CRD find managing rare disease conditions burdensome is that they have to manage everything in different places, which is emotionally and logistically taxing. One key management task is the administrative burden of gathering records from multiple organizations. This takes time (many report taking time off of work), effort (spending mental energy on the phone speaking to hospitals), and money (payment may be required to order the records).

One CRD relates her experience gathering her daughter's records:

> I had another person that… said, 'Hey, send me your daughter's records, and I want the originals.' And I'm like, Ugh, I'm going to have to take a day off from work just to get records. And I actually haven't sent them yet because I have to go out to at least three large institutions. And one of them that I've reached out to in the past was a really bad experience.
>
> CRD #26

Moreover, as a result of the decentralized and siloed nature of electronic health records, especially those from different hospital systems, medical advice from one doctor could conflict with that of another doctor. It would then be up to the IRD or CRD to reconcile conflicting advice. IRD and CRD want digital tools to help centralize information.

As one IRD puts it:

> Logistically it's hard because my PCP doesn't know anything about it. So her tips are so counteractive to what my neurologist says. So I'm like, I don't know which one of you to believe. I'm going to go with the neurologist. But, so, even with doctors, I wish there's a place that I could type one message, it would go to the three of my providers who manage this. And then I'd actually get a smart answer instead of this one telling me, oh, go see a neurologist, my neurologist saying, go see that one. And my PCP being like, oh, have you tried water all day?
>
> IRD #97

The desire for centralization extended to other areas as well, as one CRD notes:

> And there's so many different companies out there that are trying to do something in the rare disease space and some are doing a lot more than others. So if I could have a central area where the trusted sources or the ones that I'm interested in came through or be alerted when something is published.
>
> CRD #26

3.4 | Theme 2: Digital tools should foster community and promote trust

Our second theme organizes around comments that leveraging community for support, advice, and understanding was invaluable to IRD and CRD for managing rare disease conditions. Digital tools were helpful in finding and sustaining these online communities.

3.4.1 | Subtheme 1: Only rare disease community members truly understand the rare disease journey and can give meaningful support

In interviews with IRD and CRD, a common refrain was that having a rare disease can be a very isolating experience. Geographically, people dealing with a rare disease often do not live near one another due to the low prevalence of the specific condition. Having an online community helps IRD and CRD to not feel so isolated.

As one CRD puts it:

> So for me, I think the use of digital tools in our journey has been super helpful and I don't know where we would be without it. It's just given me the ability to connect with families all over the world that I probably would have never spoken to and I would have still thought that we were the only ones. So for us, it's been super helpful.
>
> CRD #30

In addition, family members and friends, though supportive, can never really truly understand what rare disease community members are going through on a day-to-day basis. Thus, having a community who has been through similar experiences helps to mitigate this isolation. These community members truly understand the highs and lows of managing a rare disease, and can offer the emotional support and motivation to keep going.

As one IRD puts it:

> Because one of the biggest things I've learned through dealing with this for so many years is, the friends that I have that never have had to deal with anything like this in their lives before they met me, they do their very best and they're very caring and understanding and I can explain it to the best of my ability, but the only people that truly understand it, are the people that deal with it themselves.
>
> IRD #117

Moreover, both IRD and CRD spoke about how one of the most positive aspects of having been supported by a community is that they are motivated to give back to the same community. For example, they made sure to take the time to "like" a post from a fellow member, give messages of support, or respond to questions. Giving back to the same community that nurtured them helps new members as they come into the group and begin their rare disease journey.

As one IRD puts it:

> Anytime I get a notification from one of those groups, like that someone posted something, I'll always take a minute to look at it. And like, especially if I see that it's someone that's like new to the group and it's like, 'Hey, this is my son or daughter, this is their story.' I make it a point to read through that whole thing and like comment on it, like, 'Hey, I don't know you and you don't know me, but like we're in this together.' There's nothing else to it. Other people don't know we got to stick together.
>
> IRD #117

As one CRD puts it:

> If I see somebody's commented, a parent or somebody has commented on the UDN Facebook page, then I make a real effort to respond to them regardless if I have something to say or just a thumbs up, because I want them to know that we're all here,

> we're all together. So just virtually supporting them that way.
>
> CRD #26

3.4.2 | Subtheme 2: Rare disease community members are able to provide advice from their "lived" perspective

IRD and CRD report that, while doctors as the experts are important dispensers of key information and advice, it is often the case where even experts have a lack of knowledge about the condition. Or, the IRD and CRD may be seeing multiple specialists, all of whom have conflicting medical advice, leaving the IRD and CRD trying to make decisions without guidance.

As one IRD puts it:

> It's not only not where to go, but who to trust. I'm at this point going to random strangers on the Internet for my information. Is it good? I don't know, there's no science behind it. There's just someone's experience. Which in a way is better because a doctor with a test tube, looking around can't actually feel what I'm feeling.
>
> IRD #97

Thus, rare disease community members find it helpful to leverage online communities to find specific and timely advice from fellow members who have a "lived" perspective. This ranges from advice on diet, dealing with supervisors at work, American of Disability Act (ADA) rights, finding housing if evicted, or finding nurse or caregiver support. Having the advice be specific to their unique situation as well as timely due to the active nature of the group is what makes the advice so helpful. Essentially, having these community members' advice turns the unknown into the known.

As one CRD puts it:

> I don't know what I should have done when I found out because this group it's been really, really helpful. At first, I was crying and I was like, 'I don't know what's going to happen with my son'. And they told me and they showed me support. They gave me advice and they told me, you have to feed him every 2 h. If something's going on, just take him right to the ER and bring the letter with you and advocate for him.
>
> CRD #100

Almost all CRD and IRD we interviewed used Facebook Groups as one of their online communities. Most agreed that Facebook Groups were generally a "great" tool for the purpose of fostering a strong community. However, they also indicated that trust in any online community was a growing concern of theirs. Trust in each other was the glue that held the community together. Thus, they desired better features to foster and maintain trust in the online community.

While the IRD and CRD that were interviewed generally had positive experiences in their online interactions, there have also been negative aspects, such as group members who engage in "shady" multi-level marketing to other participants or spread misinformation.

As one CRD notes:

> This might sound weird but sometimes I do worry about other families connecting with people who … And maybe families that might not have the same safety concerns that somebody else… You know what I mean? So I very rarely will connect with somebody that I don't know through another mom in this community.
>
> CRD #30

As one IRD puts it:

> Because on Facebook a couple years ago, there was a life hack that if you have a migraine and you take two Excedrin and drink a can of Bang energy drink, it'll cure it. Yeah. That almost landed me in the hospital. So I've been very cautious since then.
>
> IRD #97

Thus, features that promote and maintain trust like a verification process, and rules around what can be said or actions that could be taken were recommended.

3.5 | Theme 3: Digital tools should provide trusted and personalized information about the condition and what the future may hold

Our third theme organizes around information needs of IRD and CRD. Because rare diseases are by definition rare, it is often the case where IRD and CRD searching for information about a condition find it difficult to ascertain whether it is from a trusted source. Moreover, because of this dearth of information, there also can be a lack of understanding about the disease progression.

3.5.1 | Subtheme 1: There is a lack of trusted information that IRD and CRD can find about their rare disease condition

IRD and CRD report that searching for trusted information about the rare disease condition is challenging. There can be an overwhelming amount of information that they have to wade through; however, CRD and IRD are challenged by not knowing which sources are reputable.

As one CRD notes:

> So if I could create a tool or even some filtered search function that had trusted resources, scientific resources. I'd definitely stay away from anything that's not published by an institution or a nonprofit that I don't know.
>
> CRD #26

Moreover, even if IRD and CRD were able to rely on the more science-focused, trusted sites like GeneReviews, they are often unable to comprehend materials without a dictionary on hand. The level of science terminology is too advanced for most non-science people without enlisting outside help or dedicating time to educate themselves.

As one CRD puts it:

> Because, literally, I'm not kidding. There was one paper ...[where] I sat and I literally Googled all 23 words.
>
> CRD #118

3.5.2 | Subtheme 2: There is a lack of knowledge that IRD and CRD have about the disease progression or what the future holds

In addition, the rarity of the disease can also result in a lack of knowledge that IRD and CRD have about what is happening with regard to the person with the rare disease, and what the disease progression could look like. Doctors often do not know due to the rarity of the disease, and different specialists could give conflicting advice. Patients then have to act as their own care manager.

As one CRD puts it:

> Like, how do we plan to move forward in life? Like, what is our plan? Do I just ... You need to know this stuff and I knew nothing.
>
> CRD #30

As one IRD states:

> Like I said, I can check my muscle mass with that device. And I submit that stuff to the dietician, and they say, 'Great. Thank you.' But they don't know how to use the information I'm giving them, because they never had a patient like me before.
>
> IRD #115

Care management for rare diseases often encompasses non-medical areas, such as housing and employment. CRD and IRD discussed their experiences with homelessness and job loss, highlighting the need for digital tools to address uncertainty in these areas.

As one CRD puts it:

> There was not FMLA at the time, so I ended up getting evicted from our home. Ever since then we've been homeless, doubled up with family members. So, that's been, what? Going on four years. And it's just a hole that we've never been able to ... And it's just ... So, I feel like as far as that goes there's another layer of things that rare disease people deal with that there is nothing. There is just nothing.
>
> CRD #30

Or, as another IRD puts it:

> The first being something to help people with one of these rare diseases to get a job. I know it wouldn't be useful to everyone, but I feel that it could help enough. In my case, I don't have many options at all for jobs due to my condition, transportation, and the likes. I am healthy enough to not be able to get disability, but not quite so healthy to do most jobs. Most jobs require you to stand nearly the entire time.
>
> IRD #117

Another IRD notes:

> I'm very lucky, my work is very supportive, but I don't know what to ask for. I qualify under the Americans with Disabilities Act, but I don't know what, like, recommendations I could do to make my life easier at work.
>
> IRD #97

4 | DISCUSSION

We conducted an interview-based study with eight caregivers and individuals affected by a diversity of rare diseases to identify digital tool needs of IRD and CRD. To our knowledge, this is the first study focused on holistic digital tool usage by the rare disease community, and to examine it from a user research perspective to understand what types of features they need in digital tools.

Through our analysis, we constructed three major themes related to the role of digital tools in rare disease management: (1) digital tools should lessen the burden of managing a rare disease condition; (2) digital tools should foster community and promote trust; and (3) digital tools should provide trusted and personalized information about the condition and what the future may hold. Our study fills gaps in the literature by interviewing both CRD and IRD about the use of digital tools in rare disease management, and covering a wide

range of subject matter related to digital tools – logistics, emotional support, and information gathering.

The first theme is "Digital tools should lessen the burden of managing a rare disease condition." Consistent with previous studies of parents of children with rare conditions (Barton et al., 2019; Bouwman et al., 2010; Deuitch et al., 2021; Litzkendorf et al., 2020; Nicholl et al., 2017; Tozzi et al., 2013), we found that both CRD and IRD use the Internet to find information about their rare conditions. Imperfect care coordination, which includes IRD and CRD having to coordinate their care in the face of absent or conflicting medical advice, led our IRD and CRD participants to experience additional burdens because they had to act as the informal case worker for their family. Impacts of such burdens can be physical (fatigue), financial (loss of earnings), and psychosocial (disruption to school/work and emotional burden) (Simpson et al., 2021). Only a few studies specifically addressed care coordination, even though understanding how IRD and CRD try to coordinate their nonmedical care is often cited as being equally important in disease management (Simpson et al., 2021; Walton et al., 2022). Participants described a need for digital tools to assist with information gathering and centralization.

The second theme is "Digital tools should foster community and promote trust." We found that both CRD and IRD seek social and emotional support using social media, including Internet support groups, to help navigate their rare disease management journeys, and to feel less isolated by finding a community. For both CRD and IRD, joining online support groups had an empowering effect. Key activities include exchanging information, encountering emotional support, finding recognition, sharing experiences, helping others, and deriving amusement, consistent with van Uden-Kraan et al. (2008). Being able to hear about other families' lived experiences through social media platforms were both valuable and emotionally challenging. Caregivers and individuals were able to find advice on day-to-day management, healthcare providers, and therapy interventions. Connecting online with other rare disease community members allows them opportunities to share more relevant and personalized advice than from Internet searches or a healthcare provider.

We also found that there are drawbacks to using digital tools in the context of health. Facebook is widely used as a support group platform, but privacy was cited as being increasingly top of mind by users. In addition, there is concern over the quality of the information discussed as well as being exposed to the negative side of the disease, consistent with the findings of other studies (Titgemeyer & Schaaf, 2020, 2022). Participants expressed a need for digital tools to support community building, prevent misinformation, and protect privacy.

While there is published literature supporting the first two themes, the third theme, "Digital tools should provide trusted and personalized information to understand the condition and what the future may hold," may be a new, exciting finding and worthy of further research.

Based on our analysis, there was a core set of needs regarding digital tools that all CRD and IRD seemed to require, regardless of their condition or phenotype. These provide a roadmap for recommendations for how digital tools could better support the rare disease community. Specifically:

1. Enable CRD and IRD to be more passive rather than actively expending time and energy. Examples of features that enable IRD and CRD to be passive include alerts, such as when new research about a condition has been published, or a relevant new event that has been organized. On the user interface side, digital tools can make existing information easier to find by organizing it in a better, more streamlined fashion. One way to do this is to allow CRD and IRD to customize the information to make it relevant to their needs. For example, making filtering easier, or utilizing technologies like chatbots that would respond to the preferences or needs of the IRD or CRD. This would help them manage the overwhelming amount of information, and decide what is relevant and not.
2. Centralize key information to minimize the logistics of going to multiple places for information and resources. Examples of features that help centralize information would be websites that collected information in a central hub that acted as a one-stop shop. This might include the ability to send messages to multiple doctors at once so that everyone is aligned with the care the patient is receiving; or it could include links to frequently accessed support groups.
3. Enhance the trust and privacy of online communities. Digital tools can help to provide more relevant, trusted, and actionable information. They can do this by improving the content being offered and making the user interface more accessible and effective. Content creators can make sure they have Search Engine Optimization to make their websites more easily discoverable, or resource pages with links to trusted sites.
4. Personalize and customize information to make it more relevant, and trustworthy, for example, chatbots. Especially helpful would be to ensure that the information is vetted from a trusted source, and written in easy-to-understand language (perhaps using the Flesch–Kincaid readability test). Moreover, having links to information regarding housing, ADA disability rights, and more would be appreciated by IRD and CRD.

None of these findings are contrary to what generally is considered good practice in building products; rather, they are in line as to what is recommended regarding good user interface and relevant content (Crumlish & Malone, 2009). These findings help to contextualize general principles in terms of rare disease consumers by shining a brighter light on their specific needs.

Geoffrey Moore, in his book "Crossing the Chasm" (2014), describes the challenge of producing a consumer product that successfully transitions through the main stages of user acceptance: first the innovators start using the product, then the early adopters, then the early majority, then the late majority and finally the laggards. Once it has reached the laggards, the product has hit the mass market (Moore, 2014). However, many products fail to "cross the chasm" between early adopters and early majority.

Because the rare disease community members may by necessity be very willing to try new digital tools, they can be viewed as "early adopters" in Moore's paradigm with regard to how a product is launched and adopted by the mass market. Thus, if product designers can utilize feedback from these early adopters, it may help digital tools to "cross the chasm" and reach a wider healthcare audience.

4.1 | Limitations

There are several limitations to this research. First, we recruited participants through social media rather than directly through venues such as clinics. A limitation to online recruitment is potential for collecting fraudulent data (Dewitt et al., 2018; Glazer et al., 2021; Hausmann et al., 2022; Pozzar et al., 2020; Wisk et al., 2019). We had challenges with fraudulent participants (e.g., participants are not who they say they are) as well as bots throughout the recruitment process, similar to what Levi et al. (2022) and Pratt-Chapman et al. (2021) reported in their research. While it is not known why such participants want to participate fraudulently, we speculate that it could be the desire to be heard or the anticipation of receiving the $25 gift card. In fact, research has shown that studies with a participation incentive have 6× the fraudulent behavior than studies that did not include participant payments (Bowen et al., 2008). We addressed this concern by employing strategies to ensure eligibility of interviewees.

Another study limitation is the reduced sample size of eight interviewees with analyzable data, though 14 interviews were conducted and our target sample size was 15. A potential contributing cause for the small sample size could be the request for participants to fill out a digital diary prior to the interview. However, the data from these eight interviews produced an adequate depth of understanding of digital tool needs for IRD and CRD. A third limitation is that, while we strived for diversity on multiple factors in our interview sample, we did not achieve diversity in terms of race and gender with a sample predominately white and female. Our study participants' demographics are similar to a 2021 study in the rare disease community (86% reported as female and 63% reported as white [RARE-X, 2021]), suggesting that the results may be transferable to that community. Finally, as per the inclusion and exclusion criteria, this study only evaluated participants who were reasonably comfortable utilizing digital tools and did not consider the perspectives of those who are unable to navigate the online world.

Due to the qualitative nature of this study, as well as the limited number of interviews, this research is likely not transferable to the wider rare disease community. However, rare disease can affect all demographic groups, and efforts must be made to reach and serve underrepresented individuals with rare disease and their caregivers.

4.2 | Future research

Future directions to build upon this research could include partnering with existing patient communities, rare disease organizations, and startups in the health space. These organizations have made it easier for those in the rare disease community to engage with each other, share resources, and advocate for therapeutics. Partnering with organizations like these could be a fruitful next step to delving deeper into digital tool research.

Areas of future research should address potential variability in availability of resources by condition, such as whether some rare conditions have fewer digital tool resources than others. In addition, future research could address any differences in digital tool usage perspectives between individuals and caregivers with varying levels of digital literacy. Finally, although the research team found no differences in themes for CBD and IRD in the scope of this article, additional research with larger sample sizes focusing on potential differences in CBD and IRD needs with regard to digital tools could prove useful.

5 | CONCLUSION

We hope that future product designers can utilize this research to build more useful digital tool features. Not only will this be useful for the rare disease community, but viewing the rare disease community through a user research lens can help improve digital tools for a wider health consumer audience.

Genetic counselors can benefit from this research in terms of understanding the digital needs of those navigating the rare disease experience. Such understanding can help genetic counselors offer more targeted referrals and resources to patients. Moreover, genetic counselors can be key contributors to the creation of digital tools for rare disease patients, such as by being part of focus groups or in product development roles at companies.

AUTHOR CONTRIBUTIONS

Andrea Chang conceptualized the study. All authors contributed to the study design. Andrea Chang recruited the participants, developed the interview guide, and conducted the interviews with participants. Andrea Chang and Sarah D. Huang developed the codebook, performed the coding, and conducted the data analysis with input from Christina G. S. Palmer, Nanibaa' A. Garrison, Johanna L. Schmidt, and Daniel J. Benjamin. Andrea Chang and Sarah D. Huang wrote the first draft. All authors critically revised the manuscript for intellectual content. Andrea Chang confirms that she had full access to all the data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis. All of the authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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CONFLICT OF INTEREST STATEMENT

Authors Andrea Chang, Sarah D. Huang, Daniel J. Benjamin, Johanna L. Schmidt, Christina G. S. Palmer, and Nanibaa' A. Garrison declare that there are no conflicts of interest to report. This research was conducted prior to Andrea Chang's current employment at Quest Diagnostics.

DATA AVAILABILITY STATEMENT

Data presented in this manuscript are not available as per IRB protocol.

ETHICS STATEMENT

Human studies and informed consent: Informed consent was obtained from all participants included in the study. The UCLA Institutional Review Board approved all aspects of this study (Protocol IRB-21-001547). All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000.

Animal studies: No nonhuman animal studies were carried out by the authors for this article.

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REFERENCES

Barton, K. S., Wingerson, A., Barzilay, J. R., & Tabor, H. K. (2019). "Before Facebook and before social media…we did not know anybody else that had this": Parent perspectives on Internet and social media use during the pediatric clinical genetic testing process. *Journal of Community Genetics*, *10*(3), 375–383. https://doi.org/10.1007/s12687-018-0400-6

Bouwman, M. G., Teunissen, Q. G. A., Wijburg, F. A., & Linthorst, G. E. (2010). "Doctor Google" ending the diagnostic odyssey in lysosomal storage disorders: Parents using Internet search engines as an efficient diagnostic strategy in rare diseases. *Archives of Disease in Childhood*, *95*(8), 642–644. https://doi.org/10.1136/adc.2009.171827

Bowen, A. M., Daniel, C. M., Williams, M. L., & Baird, G. L. (2008). Identifying multiple submissions in Internet research: Preserving data integrity. *AIDS and Behavior*, *12*(6), 964–973. https://doi.org/10.1007/s10461-007-9352-2

Braun, V., & Clarke, V. (2022). Conceptual and design thinking for thematic analysis. *Qualitative Psychology*, *9*(1), 3–26.

Braun, V., & Clarke, V. (2023). Is thematic analysis used well in health psychology? A critical review of published research, with recommendations for quality practice and reporting. *Health Psychology Review*, *17*(4), 695–718.

Cacioppo, C. N., Conway, L. J., Mehta, D., Krantz, I. D., & Noon, S. E. (2016). Attitudes about the use of Internet support groups and the impact among parents of children with Cornelia de Lange syndrome. *American Journal of Medical Genetics. Part C, Seminars in Medical Genetics*, *172*(2), 229–236. https://doi.org/10.1002/ajmg.c.31504

Close, S., Smaldone, A., Fennoy, I., Reame, N., & Grey, M. (2013). Using information technology and social networking for recruitment of research participants: Experience from an exploratory study of pediatric Klinefelter syndrome. *Journal of Medical Internet Research*, *15*(3), e48. https://doi.org/10.2196/jmir.2286

Corbin, J., & Strauss, A. (2014). *Basics of qualitative research*. SAGE Publications Inc. https://us.sagepub.com/en-us/nam/basics-of-qualitative-research/book235578

Crumlish, C., & Malone, E. (2009). *Designing social interfaces: Principles, patterns, and practices for improving the user experience*. O'Reilly Media, Inc.

DeHoff, B. A., Staten, L. K., Rodgers, R. C., & Denne, S. C. (2016). The role of online social support in supporting and educating parents of young children with special health care needs in the United States: A scoping review. *Journal of Medical Internet Research*, *18*(12), e333. https://doi.org/10.2196/jmir.6722

Deuitch, N. T., Beckman, E., Halley, M. C., Young, J. L., Reuter, C. M., Kohler, J., Bernstein, J. A., Wheeler, M. T., Ormond, K. E., & Tabor, H. K. (2021). "Doctors can read about it, they can know about it, but they've never lived with it": How parents use social media throughout the diagnostic odyssey. *Journal of Genetic Counseling*, *30*(6), 1707–1718. https://doi.org/10.1002/jgc4.1438

Dewitt, J., Capistrant, B., Kohli, N., Rosser, B. R. S., Mitteldorf, D., Merengwa, E., & West, W. (2018). Addressing participant validity in a small Internet health survey (the Restore study): Protocol and recommendations for survey response validation. *JMIR Research Protocols*, *7*(4), e96. https://doi.org/10.2196/resprot.7655

Fitzpatrick, R. (2013). *The mom test: How to talk to customers & learn if your business is a good idea when everyone is lying to you*. Robfitz Ltd.

Glazer, J. V., MacDonnell, K., Frederick, C., Ingersoll, K., & Ritterband, L. M. (2021). Liar! Liar! Identifying eligibility fraud by applicants in digital health research. *Internet Interventions*, *25*, 100401. https://doi.org/10.1016/j.invent.2021.100401

Griggs, R. C., Batshaw, M., Dunkle, M., Gopal-Srivastava, R., Kaye, E., Krischer, J., Nguyen, T., Paulus, K., & Merkel, P. A. (2009). Clinical research for rare disease: Opportunities, challenges, and solutions. *Molecular Genetics and Metabolism*, *96*(1), 20–26. https://doi.org/10.1016/j.ymgme.2008.10.003

Hausmann, J. S., Vizcaino-Riveros, J., Marin, A. C., Minegishi, M., Cox, R., Chang, M.-L., Schanberg, L. E., Natter, M., & Weitzman, E. R. (2022). Feasibility and efficacy of online strategies to recruit parents of children with rheumatic diseases for research. *ACR Open Rheumatology*, *4*, 410–416. https://doi.org/10.1002/acr2.11360

Levi, R., Ridberg, R., Akers, M., & Seligman, H. (2022). Survey fraud and the integrity of web-based survey research. *American Journal of Health Promotion: AJHP*, *36*(1), 18–20. https://doi.org/10.1177/08901171211037531

Litzkendorf, S., Frank, M., Babac, A., Rosenfeldt, D., Schauer, F., Hartz, T., & Graf von der Schulenburg, J.-M. (2020). Use and importance of different information sources among patients with rare diseases and their relatives over time: A qualitative study. *BMC Public Health*, *20*(1), 860. https://doi.org/10.1186/s12889-020-08926-9

Meta Platforms Inc. (2022). *Fourth Quarter 2021 Results Conference Call Transcript*. https://s21.q4cdn.com/399680738/files/doc_financials/2021/q4/Meta-Q4-2021-Earnings-Call-Transcript.pdf

Miller, E. G., Woodward, A. L., Flinchum, G., Young, J. L., Tabor, H. K., & Halley, M. C. (2021). Opportunities and pitfalls of social media research in rare genetic diseases: A systematic review. *Genetics in Medicine: Official Journal of the American College of Medical Genetics*, *23*(12), 2250–2259. https://doi.org/10.1038/s41436-021-01273-z

Moore, G. A. (2014). *Crossing the chasm, 3rd edition: Marketing and selling disruptive products to mainstream customers*. HarperCollins.

Nguengang Wakap, S., Lambert, D. M., Olry, A., Rodwell, C., Gueydan, C., Lanneau, V., Murphy, D., Le Cam, Y., & Rath, A. (2020). Estimating cumulative point prevalence of rare diseases: Analysis of the Orphanet database. *European Journal of Human Genetics: EJHG*, *28*(2), 165–173. https://doi.org/10.1038/s41431-019-0508-0

Nicholl, H., Tracey, C., Begley, T., King, C., & Lynch, A. M. (2017). Internet use by parents of children with rare conditions: Findings from a study on parents' web information needs. *Journal of Medical Internet Research*, *19*(2), e51. https://doi.org/10.2196/jmir.5834

Pelentsov, L. J., Laws, T. A., & Esterman, A. J. (2015). The supportive care needs of parents caring for a child with a rare disease: A scoping review. *Disability and Health Journal*, *8*(4), 475–491. https://doi.org/10.1016/j.dhjo.2015.03.009

Pew Research Center. (2021a). *Internet/broadband fact sheet*. Pew Research Center. https://www.pewresearch.org/internet/fact-sheet/internet-broadband/

Pew Research Center. (2021b). *Mobile fact sheet*. Pew Research Center. https://www.pewresearch.org/internet/fact-sheet/mobile/

Pew Research Center. (2021c). *Social media fact sheet*. Pew Research Center. https://www.pewresearch.org/internet/fact-sheet/social-media/

Pozzar, R., Hammer, M. J., Underhill-Blazey, M., Wright, A. A., Tulsky, J. A., Hong, F., Gundersen, D. A., & Berry, D. L. (2020). Threats of bots and other bad actors to data quality following research participant recruitment through social media: Cross-sectional questionnaire. *Journal of Medical Internet Research*, *22*(10), e23021. https://doi.org/10.2196/23021

Pratt-Chapman, M., Moses, J., & Arem, H. (2021). Strategies for the identification and prevention of survey fraud: Data analysis of a web-based survey. *JMIR Cancer*, *7*(3), e30730. https://doi.org/10.2196/30730

RARE-X. (2021). *Strategic directions and recommendations: Diversity, equity, and inclusion scoping review* (p. 102). RARE-X. https://rare-x.org/wp-content/uploads/2022/02/dei-020722.pdf

Schumacher, K. R., Stringer, K. A., Donohue, J. E., Yu, S., Shaver, A., Caruthers, R. L., Zikmund-Fisher, B. J., Fifer, C., Goldberg, C., & Russell, M. W. (2014). Social media methods for studying rare diseases. *Pediatrics*, *133*(5), e1345–e1353. https://doi.org/10.1542/peds.2013-2966

Simpson, A., Bloom, L., Fulop, N. J., Hudson, E., Leeson-Beevers, K., Morris, S., Ramsay, A. I. G., Sutcliffe, A. G., Walton, H., & Hunter, A. (2021). How are patients with rare diseases and their carers in the UK impacted by the way care is coordinated? An exploratory qualitative interview study. *Orphanet Journal of Rare Diseases*, *16*(1), 76. https://doi.org/10.1186/s13023-020-01664-6

Titgemeyer, S. C., & Schaaf, C. P. (2020). Facebook support groups for rare pediatric diseases: Quantitative analysis. *JMIR Pediatrics and Parenting*, *3*(2), e21694. https://doi.org/10.2196/21694

Titgemeyer, S. C., & Schaaf, C. P. (2022). Facebook support groups for pediatric rare diseases: Cross-sectional study to investigate opportunities, limitations, and privacy concerns. *JMIR Pediatrics and Parenting*, *5*(1), e31411. https://doi.org/10.2196/31411

Tozzi, A. E., Mingarelli, R., Agricola, E., Gonfiantini, M., Pandolfi, E., Carloni, E., Gesualdo, F., & Dallapiccola, B. (2013). The Internet user profile of Italian families of patients with rare diseases: A web survey. *Orphanet Journal of Rare Diseases*, *8*, 76. https://doi.org/10.1186/1750-1172-8-76

Twitter, Inc. (2022). *First quarter 2022 earnings release: Operational and financial highlights*. Twitter announces first quarter 2022 results, San Francisco. https://investor.twitterinc.com/files/doc_financials/2022/q1/Final-Q1%E2%80%9922-earnings-release.pdf

van Uden-Kraan, C. F., Drossaert, C. H. C., Taal, E., Shaw, B. R., Seydel, E. R., & van de Laar, M. A. F. J. (2008). Empowering processes and outcomes of participation in online support groups for patients with breast cancer, arthritis, or fibromyalgia. *Qualitative Health Research*, *18*(3), 405–417. https://doi.org/10.1177/1049732307313429

von der Lippe, C., Diesen, P. S., & Feragen, K. B. (2017). Living with a rare disorder: A systematic review of the qualitative literature. *Molecular Genetics & Genomic Medicine*, *5*(6), 758–773. https://doi.org/10.1002/mgg3.315

Walton, H., Simpson, A., Ramsay, A. I. G., Hunter, A., Jones, J., Ng, P. L., Leeson-Beevers, K., Bloom, L., Kai, J., Kokocinska, M., Sutcliffe, A. G., Morris, S., & Fulop, N. J. (2022). Development of models of care coordination for rare conditions: A qualitative study. *Orphanet Journal of Rare Diseases*, *17*(1), 49. https://doi.org/10.1186/s13023-022-02190-3

Wisk, L. E., Nelson, E. B., Magane, K. M., & Weitzman, E. R. (2019). Clinical trial recruitment and retention of college students with type 1 diabetes via social media: An implementation case study. *Journal of Diabetes Science and Technology*, *13*(3), 445–456. https://doi.org/10.1177/1932296819839503

Yu, J., Paranagama, D., & Parasuraman, S. (2020). Recruitment strategies and geographic representativeness for patient survey studies in rare diseases: Experience from the living with myeloproliferative neoplasms patient survey. *PLoS One*, *15*(12), e0243562. https://doi.org/10.1371/journal.pone.0243562

Zurynski, Y., Frith, K., Leonard, H., & Elliott, E. (2008). Rare childhood diseases: How should we respond? *Archives of Disease in Childhood*, *93*(12), 1071–1074. https://doi.org/10.1136/adc.2007.134940

SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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