"I think we know more than our doctors": How Primary Caregivers Manage Care Teams with Limited Disease-related Expertise

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Healthcare providers play a critical role in the management of a chronic illness by providing education about the disease, recommending treatment options, and developing care plans. However, when managing a rare disease, patients and their primary caregivers often work with healthcare systems that lack the infrastructure to diagnosis, treat, or provide education on the disease. Little research has explored care coordination practices between patients, family members, and healthcare providers under these circumstances. With the goal of identifying opportunities for technological support, we conducted qualitative interviews with the primary caregivers of children with a rare neurodegenerative disorder, ataxia-telangiectasia. We report on the responsibilities that the primary caregivers take on in response to care teams' lack of experience with the illness, and the ways in which an online health community supports this care coordination work. We also describe barriers that limited participants' use of the online health community, including the emotional consequences of participation and information overload. Based on these findings, we discuss two promising research agendas for supporting rare disease management: facilitating primary caregivers' care coordination tasks and increasing access to online community knowledge.

CCS Concepts: • **Human-centered computing → Empirical studies in collaborative and social computing**.

Additional Key Words and Phrases: chronic disease management; care coordination; rare diseases

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1 INTRODUCTION

Managing a chronic disease requires access to healthcare professionals and medical expertise to learn about the disease, treatments, symptoms, and progression [18, 26, 52]. Access to this expertise can influence a person's quality of life. Prior research has identified correlations between information access and emotional and functional well-being, health competence, and positive health

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behavior change [3, 4, 20]. However, when managing a rare disease people often face difficulties accessing disease-related expertise and work with care teams who have limited knowledge about their disease or are inexperienced in its treatment [8, 55]. Few studies have explored how patients and primary caregivers cope with these challenges, and the strategies they use to learn about their health situations. A better understanding of current health management strategies is needed to inform the design of support systems that can help people overcome challenges within this context.

Towards this goal, we conducted an interview study with 15 primary caregivers of children with a rare neurodegenerative disorder, ataxia-telangiectasia (A-T). We use A-T as a case study to explore opportunities to better support the management of rare chronic conditions. A-T is an informative disease to study because it exemplifies many challenges faced by families coping with chronic degenerative diseases. First, the disease trajectories result in regular changes in health status which, in turn, necessitate changes in treatment and support strategies. Second, children with A-T follow diverse health trajectories, so while there are opportunities to learn from other families, there is also much uncertainty about each individual patient's future trajectory. Finally, like other rare genetic diseases, A-T is a multi-system disorder. It affects the central and peripheral nervous systems, the immunological system, and has a predisposition for cancer development. Thus, optimal care of individuals with A-T involves care distributed among a complex care team, often including neurologists, immunologists, pulmonologists, oncologists, therapists, and school systems. Therefore, effective strategies for supporting A-T management may be useful for other families experiencing a range of rare chronic diseases.

In this study, we partnered with the Ataxia-Telangiectasia Children's Project (A-TCP), a non-profit organization that raises funds to support the development of life-improving treatments for A-T and develops resources for families coping with A-T. Events organized by the A-TCP provided us with the opportunity to meet with families caring for children with A-T who live across the United States, and who each work with a different healthcare system. We were interested in learning how primary caregivers manage A-T in daily life, the challenges they face related to health management and care coordination, and current strategies they use to address these challenges. Specifically, our research questions included: (1) What are primary caregivers' health management tasks when caring for a child with A-T? (2) What challenges do primary caregivers encounter when working with care teams inexperienced with their child's disease? (3) What are primary caregivers' current strategies for coping with challenges related to A-T management and care coordination?

Utilizing language from prior care coordination research, and for clarity and consistency, we use the term "primary caregiver" to refer to the participants of this study. As defined in prior literature, a primary caregiver is the person responsible for the daily care and health decision making for the child [16, 45]. Every participant we interviewed was the parent or guardian of a child with A-T and a primary caregiver for their child. We use the term "care teams" to describe the large, complex, and multidisciplinary teams of people involved in the child's care. As described in [69, 76], care teams include healthcare workers with different backgrounds and goals. In the context of A-T, these care teams (described in more details in section 4.1.2), often included neurologists, immunologists, therapists, primary care providers, as well as school nurses, teachers, and aids.

Through interviews, we found that primary caregivers regularly took on health management tasks as a direct response to working with care teams who had limited knowledge of the disease itself. While our focus was on understanding health management and care coordination experiences, a cross-sectional theme that emerged within our data was the important role of online community support. Therefore, we first present the tasks that primary caregivers regularly undertook while interacting with their care teams, and then outline the ways in which use of an online health community (OHC) supported this care coordination work. We also report on barriers, such issues

related to emotional readiness and information seeking, which at times prevented access or effective utilization of this important online community support.

This work helps to set an agenda for supporting individuals managing rare diseases and people who face barriers to accessing care teams with disease-related expertise. Our findings contribute to our understanding of how families manage rare diseases and utilize online community support, as well as current barriers that limit access to important community discussions. We reflect on how the roles and responsibilities described by A-T primary caregivers differ from prior analyses of chronic disease management, and outline opportunities to support caregivers' care coordination tasks and to increase access to online community support. Our contributions include:

- We identify care coordination tasks that primary caregivers take on when working with care teams who have limited experience with their child's disease.
- We describe how an online health community provides access to resources and strategies which support primary caregivers' care coordination tasks.
- We describe barriers that discourage, and at times prevent, primary caregivers from participating in and using the online health community.
- We identify opportunities for CSCW and HCI research to support primary caregivers' care coordination tasks and to increase access to shared community experiences and information.

2 RELATED WORK

2.1 Chronic Disease Management

Following a chronic disease diagnosis, patients and primary caregivers must take on a number of tasks as they learn to live with the disease. These tasks include participating in decision-making processes, adhering to treatment, monitoring one's health at home, adapting health-promoting behaviors, and managing symptoms [18, 36, 74, 78]. In addition, managing a chronic disease often involves communication and coordination between patients, family members, and a complex care team including physicians, nurses, therapists, and, in the case of pediatric diseases, school teachers, nurses, and aids.

Care team members are often considered the experts on the disease trajectory, side effects, and possible therapies and accommodations, and are the primary source of information to help patients and primary caregivers learn about the disease [15, 23, 28]. Healthcare providers are expected to serve as advisors, providing information about one's condition, treatment options, and management plan, and recommending optimal care plans or changes to treatment when needed [6, 39, 71]. Existing models, such as the patient-centered care model and shared-decision making model, involve patients and providers bringing together their different expertise and perspectives to collaborate throughout the decision making process [22, 24, 57]. These models have been used to inform the design and development of health tools that help patients connect to their care teams for medical guidance [11, 46]. While these tools have established benefits, the existing research is based on the fundamental assumption that healthcare providers possess adequate expertise about the individual's disease, health status, and treatment options [8]. Far less research has explored what happens when this assumption fails. Our study helps to show that working with a care team with limited experience or knowledge of a patients' disease blurs the division of responsibilities, and places some responsibilities held by clinicians and other care team members in a more traditional setting onto the primary caregivers. We consider how this shift in roles could inform the design of future health tools to support patients and primary caregivers managing a rare disease.

2.2 Online Health Communities

Patients and primary caregivers now commonly go online to seek support. An estimated 51% of adults with a chronic condition use the internet to find health information or support [29]. OHCs in particular provide people with a space to share and receive socio-emotional and informational support. Access to OHC support can have important benefits for community members, increasing feelings of empowerment and perceived empathy [43, 66].

Patients and family members will often join OHCs, and remain engaged with them, to receive emotional support [58, 80]. This type of support is broad, but can include empathy, reassurance, motivation and companionship [66, 70, 79]. Community members will also use OHCs to access informational support, helping them to better understand their health situation [60]. Patients will often use their own experiences to help others develop strategies to address personal health management problems [38]. For chronic diseases, understanding others' illness trajectories can help patients to identify possible futures and cope with the anxiety that can come from feeling uncertain about about one's own prognosis [38]. OHCs also provide a space for collective sensemaking. As these experiences are shared among community members, the community engages in rich discussion and debate and develops a shared knowledge of treatments and effective health management behaviors [59].

OHCs are particularly useful in the case of rare diseases because families typically live far from others with similar health situations [56]. Through OHCs, both patients and family members can receive social support and mentorship to help them learn how to live with the disease in everyday life [21]. We expand on this work, showing how caregivers leverage peer support to acquire tools that help them to work with care teams with limited familiarity with their child's disease.

2.3 Managing a Rare Disease

Approximately 7,000 diagnoses have been classified as a rare disease. Each of these diseases affect fewer than 200,000 people in the United States [62]. However, collectively 30 million people in the U.S. have a rare disease[1]. Treatments are available for less than 10% of individuals with a rare disease [62].

Managing a rare disease can influence a person's mental, physical, and social well-being. Many of the health management tasks and challenges that patients face are similar to those managing more common chronic illnesses. For example, people with rare diseases must learn how to communicate their health situation to their family and friends, cope with the fear and distress that accompany the diagnosis, and manage their physical health in daily life [21, 55].

Families often face additional challenges when managing a rare disease. The low prevalence of a disease can lead to greater geographical distances between patients and experienced providers [47]. Patients and family members can also develop a greater expertise of the disease over time than their healthcare providers, thus changing the medical encounter. In such situations, patients and primary caregivers may take a more active role in seeking health information and making care decisions, occasionally leading to tension in the relationship [8]. In the case of pediatric rare diseases, it is often the parent who must learn to navigate this world. Patients and their families also often receive insufficient educational materials from the health system, leading to greater reliance on online information [32]. Parents of children with a rare disease often use online informational support to cope with a distressing diagnosis [32]. While prior work has shown that working with healthcare providers with limited disease-related expertise affects the patient-provider relationship [8], little work has explored how this change influences patients' or their families' daily lives.

[1] 'FAQs About Rare Diseases', National Institute of Health, 2017, https://rarediseases.info.nih.gov/diseases/pages/31/faqs-about-rare-diseases

2.4 Ataxia-telangiectasia

Ataxia-telangiectasia (A-T), falls under the umbrella of hereditary ataxias. These diseases are characterized by abnormal and uncoordinated gait, movements, and speech [42]. Distinguishing features of A-T include immune deficiency, increased cancer risk, and telangiectasias (widened blood vessels). The disease can be diagnosed through genetic testing. Approximately 1% of the population carries the A-T gene mutation, and A-T is diagnosed in approximately 1/40,000 – 1/100,000 of live births [27].

The onset and progression of A-T symptoms can be highly variable. Symptoms of A-T are often noticeable by the time a child starts to walk, around 12–18 months old, though in some cases the disease does not become apparent until a person's teens [75]. A-T progresses gradually, and the majority of children use a wheelchair by age ten [14]. Many, but not all, children are immunodeficient, and cancer occurs in approximately one-third of people with A-T [14]. Cognitive skills can also be affected in some individuals. While there is no cure for A-T, symptomatic treatments attempt to improve quality of life by reducing bothersome symptoms (e.g., tremors, dystonia, drooling) [68]. A-T is a life-limiting disease, with an average life expectancy of 25 years [19].

Few studies have investigated the lived experience of managing A-T. Fanos and Mackintosh studied how the diagnosis affected parents [25]. They found that parents used coping strategies that are similar to those discussed in other health contexts, such as living in the moment and not focusing on the future. Our work elaborates on the long-term management of this disease and how primary caregivers' roles must adapt throughout the disease development and prognosis.

3 METHODS

We interviewed primary caregivers of children diagnosed with A-T. This study was approved by the Institutional Review Board at Massachusetts General Hospital. Recruitment was supported by the Ataxia-Telangiectasia Children's Project (A-TCP). This organization works closely with A-T families, hosting caregiver weekends, organizing fundraisers, and connecting families with up-to-date research and resources. The organization reached out to parents who would be attending upcoming events, allowing us to speak in person to families who were managing the illness across the country, each working with different healthcare systems.

To recruit participants, A-TCP sent eligible participants an information sheet explaining the study's objectives and procedures. Interested parents contacted A-TCP directly, and were then put in contact with the research team. We interviewed 15 primary caregivers (13 female, 87%) representing 14 families and 17 children with A-T. All participants lived in the United States, were fluent English speakers, and had at least one child who had been diagnosed with A-T.

All interviews were conducted in-person and lasted one hour. We used a semi-structured interview format, with questions focused on health management practices, care coordination challenges, and technology use. We did not intentionally investigate online health community use, but we did ask about general technology use related to A-T management. Our interview guide included the following questions:

- How do you define health management?
- What tasks and responsibilities does A-T management include?
- What have been the biggest challenges related to your child's care?
- When was the last time you felt unsupported?
- What advice would you give to a family who just received an A-T diagnosis?
- Do you use technology to help with A-T management?

All interviews were audio recorded and transcribed. We analyzed the data using an iterative inductive analysis [13]. Two researchers analyzed the data, one with a clinical background and

one with a background in human-computer interaction, to minimize researcher bias in the data analysis. The two researchers first open coded three interviews. They then met to discuss emergent themes, and used this initial set of themes to independently code an additional three interviews. The researchers then met again to discuss missing themes and qualitative coding discrepancies. One researcher then reviewed the full dataset, with all interview transcripts, to validate themes and assign codes to transcript segments. The full research team then met to verify the emergent themes and associated transcript segments.

3.1 Ethical Considerations

In writing this manuscript, we faced a tension between bringing awareness to the disease that was the focus of our study and increasing the risk of participant identification due to the rarity of the disease. Based on conversations with the research team, research participants, and our collaborators, we have decided to share the disease name. We therefore purposefully omit demographics about participants and their children, such as age, location, and years since diagnosis, to reduce the risk of re-identification.

4 FINDINGS

We organize our results into four sections. First, we provide an overview of primary caregivers' daily health management behaviors and the most common members comprising patients' care teams (4.1). We then describe three tasks that primary caregivers took on in response to working with care teams with limited disease-related expertise (4.2). We also report on ways in which the community support through an OHC helped primary caregivers to assume these roles (4.3). Finally, we highlight barriers that limited individuals' participation in the OHC (4.4). Such barriers highlight the need for tools that help families to more easily find and interact with existing community knowledge, as we comment on in the discussion. Figure 1 provides a summary of primary caregivers' care coordination tasks, the ways an OHC supported these tasks, and barriers to using the OHC.

4.1 Overview of A-T Management

4.1.1 Daily Management. The most common symptoms reported by primary caregivers were balance issues, uncoordinated walking, and fatigue. Infections caused by immunodeficiency was another common effect of the disease. Daily health management, as described by participants, typically involved helping with everyday tasks such as dressing and eating, and monitoring their child's energy levels.

4.1.2 Patients' Care Teams. In addition to daily management, primary caregivers also coordinated their child's care, often working with large care teams. Participants' care teams included neurologists, immunologists, pulmonologists, occupational, physical and speech therapists, pediatricians, and school teachers, nurses and aids. Many participants also traveled to an A-T clinic to meet with experienced specialists, but this occurred infrequently, with many participants going once or twice over a multi-year period.

As with other chronic diseases, care coordination with these complex care teams could be challenging [2]. Participants described different coordination strategies, such as using a group email with care team members, or having only one parent go to all appointments to ensure continuity. While many participants primarily worked with care teams who did not have prior experience working with A-T, they were able to develop positive relationships with these teams. Yet, collectively participants were required to take on additional care coordination tasks in response to working with care teams with limited disease-related expertise.

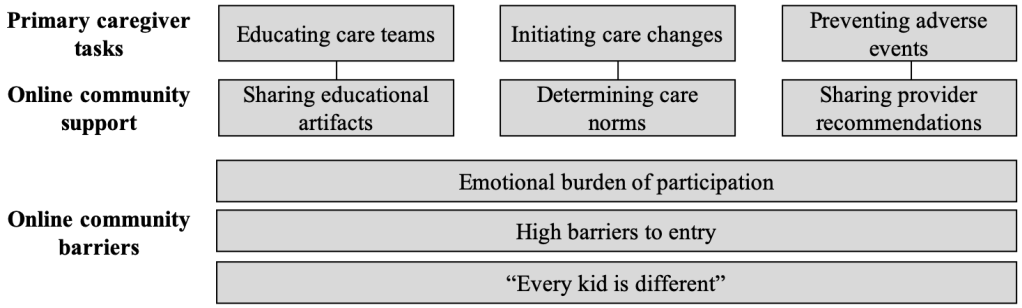

Fig. 1. Summary of findings, including primary caregivers' care coordination tasks, how the online community supports these roles, and barriers to using the online health community.

4.2 Care Coordination Tasks

We found that participants were not only responsible for the daily management tasks highlighted in section 4.1, but also additional roles and responsibilities in response to their care team's limited expertise with A-T. These additional roles included educating care teams, initiating care changes, and preventing adverse events. We describe each of these tasks below, supported by illustrative quotations from our interviews.

4.2.1 Task 1: Educating Care Teams about the Disease and Care Expectations. A common challenges participants faced was finding clinicians who had experience treating or managing A-T. The National Ataxia Foundation lists sixteen specialty clinics in the United States that provide medical care specifically for people affected by the condition[1]. As a result, many participants shared that they worked primarily with care teams who had no prior experience with A-T:

> *"The doctors here don't really understand exactly what A-T is, so they don't know what to tell us, and they pretty much tell us nothing."* – P7

> *"When we were given the diagnosis one of the things that the immunologist told us was, 'you are going to be the primary leading expert on your kid.' Because most doctors you come into contact with are either never going to have heard of the disease or have treated anybody with it."* – P13

When working with healthcare professionals who had no prior knowledge of A-T, primary caregivers took on the role of educating team members. Educating the care team included sharing details about the disease's symptoms, as well as making them aware of ongoing research and potential known and exploratory treatments. Most often, participants used resources that they found online, received from a national A-T clinic, or were provided by the A-TCP. They then found themselves providing these resources to members of their care team.

> *"I have a wonderful pediatrician who asked me to email her all of the breakthroughs. So everything that I find I send her all of that information."* – P1

> *"The A-TCP booklets have been huge. They have the caregivers manual which is very helpful for the doctors and the teachers. It's a big read so they really have to be committed. So I went through it and just made copies, like this is for occupational therapy, this is relevant for physical therapy. Because as a teacher I know when a parent comes in with*

[1]https://ataxia.org/neurologists-and-specialty-clinics/

a mountain of paperwork it can be overwhelming. And you want them to get the most important things quickly. And then they can ask me questions from there." – P15

Even when primary caregivers had a positive relationship with their care teams, they were constantly on the lookout for healthcare professionals who had more experience working with A-T. This was because participants felt the quality of care for their child would be higher if they were able to see a specialist who had experience treating this disease. To access better healthcare, families occasionally made significant life changes, such as moving to a different city. These moves led to developing a completely new care team. Therefore, patients' care teams were changing frequently. Additionally, caregivers frequently educated their school systems about how to care for their child:

"I met with all the teachers before school started to explain ataxia. And I had done that all throughout elementary school with her teachers. Just so they had an idea." – P11

"The school sat down with us, with the district nurse, with everybody. There were 20 people in the room. Physical therapy, Occupational therapy, speech, all the aids, special needs people. Sat down with us and wanted to learn. I gave them the A-TCP books and all the information." – P4

Thus, educating one's care team was not just a one-time task. Rather, educating care team members was an ongoing responsibility. As a result, participants regularly expressed the need for more resources to help them educate their large, and frequently changing, care teams.

4.2.2 Task 2: Initiating Care Changes to Address Disease Progression. In addition to educating care teams, participants were often responsible for initiating changes to their child's care as the disease progressed and their needs changed. This meant that primary caregivers needed to recognize changes in their child's health and when these changes required adjustments in their care routines:

"The big task [as a caregiver] is seeing the change. One day it seems like everything is normal, and then it progresses. Every few months you're adjusting to the decline." – P2

"He was getting physical therapy, occupational therapy, and speech therapy. I felt like a lot of the goals needed to be readjusted. Because they were focusing on fixing, and we needed to make it to management. So if someone's not understanding you, what do you do? So we sat and we revised his IEP first. And every year working with his teachers and his team to make sure that socially he's adapting and his learning is adapting. That's been paramount." – P15

Initiating changes to a child's care was particularly challenging for participants because they had limited information about the disease and its progression. Given the diversity of symptoms that may accompany this disease and the uncertain progression, parents were unsure about whether new developments could be attributed to A-T. Without information to help them understand the disease, participants said they had many unanswered questions about the effects of the disease and the progression of the illness over time:

"I think its hard for moms and dads to know is this A-T or is this just her? I don't know. It's such an early onset. I don't know what is her and what is ataxia." – P9

"Some things, I'm like, is this A-T related? Is it not A-T related?" – P3

Without answers to these questions, caregivers struggled to identify disease progressions, and know if and how their care teams should respond. Participants often described feeling distressed about an unknown future, and having to be ready to respond to any changes despite the uncertainty:

"Educating yourself is really important for parents. To not sit back and wait for a doctor to tell them what they should be doing. You have to be proactive. Really right now we're just treating the immune system and we're doing physical therapy but we haven't had to

> *deal with any other extensive medical care stuff yet. It's kind of looming, you're always afraid of when the other shoe is going to fall."* – P13

Even as primary caregivers served as the experts about the disease when interacting with their child's care teams, this expertise would decline over time as the disease progressed and caregivers had to learn how to continuously adapt. Being proactive was considered critical by many participants, and yet they did not always know what they were preparing for, how to prepare, and how to adapt their child's care. Without more information about the scope and the trajectory of the disease, participants faced the difficulty of trying to be proactive yet remaining uncertain about what changes to prepare for.

4.2.3 Task 3: Preventing or Responding to Adverse Events. One of the most important roles that primary caregivers took on while working with their care teams was preventing clinical errors and adverse events. Almost all of the participants shared examples of adverse events that occurred due to a lack of understanding of how to care for children diagnosed with A-T. These events were due to (1) attributing unrelated symptoms to A-T or failing to recognize atypical manifestations related to A-T (errors of omission) and (2) prescribing treatments that are inappropriate for A-T patients (errors of commission). For example, a participant shared that as their child became increasingly sick, many of the child's health issues were attributed to A-T incorrectly, and therefore not given the attention they deserved:

> *"The liver issues they automatically [attributed to A-T]. Her kidneys were failing and they thought that was A-T. And I'm like not at this age, no it's not... I've talked to our doctors about this. They were just like, we should have known that something wasn't right."* – P3

According to the participant, both the family and doctors were unsure if available treatments could have addressed these particular health issues. However, the doctors did say that, in reflection, they could have recognized the risk of severe health complications earlier.

Errors of commission were also commonly described by participants, in which healthcare professionals recommended treatments or medications that are unsafe for people with A-T. As shown in the example below, primary caregivers had to take an active role to recognize and prevent these errors. However, intervening in these decisions could be a complicated process.

> *"One of the local emergency rooms would have killed [child] if I hadn't been there. He had a sore throat and fever, could barely swallow. We go to the ER, and the doctor wanted to put Lidocaine in his throat. They wanted to just numb his throat. And I said, 'oh no no no, you can't do that.' He goes, 'what do you mean I can't do that?' I said, 'you can't do that, he's got ataxia, he already has swallowing issues. You numb his throat we might lose the ability to swallow. You can't do that.' And he left the room. So I call my best friend who happens to be our primary care doctor, and I said '[Doctor] wants to x, y, and z. He won't listen to me.' So he called the ER, he got the doctor on the phone, and he said, 'you need to stop.' If he gave that to him god only knows what could have happened."* – P14

The above examples demonstrate how participants took an active role in monitoring treatment decisions and advocating for proper care. This often involved questioning and opposing treatment decisions. Many of the participants commented that while opposing a healthcare provider's decision can be difficult, this became a common and necessary part of caregiving for a child with a rare disease, especially when caregivers felt they had more disease-related expertise than their current care team members.

4.3 Online Community Support

We found that when describing health management and care coordination, participants frequently brought up their use of the same OHC: a private Facebook group for A-T parents. All but one of the participants were a current or former member of the group. Below we outline the specific ways in which the OHC supported primary caregivers in taking on the three roles of educating care teams, initiating care changes, and preventing adverse events.

4.3.1 Sharing Educational Artifacts. To support their role as educator, primary caregivers looked to the OHC to find and share resources. Participants described three types of useful artifacts that have been shared through the community: child evaluations, research findings, and individualized education programs (IEPs). Participants used these documents to inform clinicians and schools about their expectations for their child's care.

Child evaluations were one useful resource shared by community members. By having access to the labs or tests that were helpful for other families, participants were able to use them to inform their own care teams about evaluations they would like to consider:

> *"There's this family and she contacted me out of the blue through Facebook. They were doing things that weren't necessary. I printed all of [child's] labs off, and gave them to her. They basically build off of us."* – P4

> *"Someone [on the Facebook group] had a technology evaluation done for their kid. And they actually posted a couple pages of it, and it was very detailed. I actually screenshotted it. And I'm going to take it to my school. That's actually part of what the meeting is going to be about next week, is can I get that evaluation done. And to show them, look this is the expectation, I want something like this. Because a lot of parents are saying they have requested it previously."* – P2

Research studies and results were also shared among community members. Many participants said that research was a source of hope for them, and were very involved in monitoring research studies and results. Participants were then also able to use this research as a backup resource when disagreements arose about appropriate care procedures:

> *"The orthopedist and I had a little tiff over him wanting to do x-rays. He was saying I was referencing old research and you can't believe everything that you hear. There's no reason why he can't x-ray [child]. And I was like, 'well, I am referencing old research because it's some of the only research that exists on A-T, and you're not x-raying [child's] hands.' "* – P15

As in this case, research was a useful tool when primary caregivers were advocating for proper care. Finally, Individualized Education Programs (IEPs) were often shared in the OHC. An IEP is a document that details all of the supplementary services that the school will provide for a child who qualifies for special education, such as one-on-one aids, technology, and therapies. Creating these documents can be challenging if primary caregivers do not know which services may be available or helpful. By sharing these documents with one another, participants provided each other with the tools necessary to educate the school systems about the needed programs and services:

> *"Recently I found out that there's an app where you can take a picture of their assignments. So you can take a picture of the document and it allows them to be on the ipad and they can fill in on this app. It's $5 but it's well worth it. I actually have an IEP meeting with the school on Thursday. And that's something that I am going to bring to them and say let's start using this. You find out about those kinds of things on the Facebook group."* – P2

As these documents are shared within the community, they not only provide a direct resource for parental use, but also reveal how artifacts can differ based on the child's needs. In addition to

IEPs, child evaluations and research findings represented some of the physical documents that were shared within the OHC and became educational artifacts to communicate care expectations to the care team.

4.3.2 Determining Care Norms. Another benefit of the online health community was access to the community's knowledge of what was typical for children with A-T, helping primary caregivers to learn how to adapt care to their child's current health status. When a caregiver began to notice a change in their child's health, the community was able to provide strategies for how to adapt to these changes:

> *"For the leg braces, it would be things that I would notice, like I could tell that [child's] feet were starting to turn in a little bit. And then just out of the blue somebody posted about 'oh look at so-and-so's neat little leg braces.' And it's like, huh, well maybe [child] needs leg braces. 'Honey, when you take [child] to therapy ask her if we need leg braces.' "* – P12

> *"There was just one the other day. It was someone who was just recently diagnosed, and [child] had several swollen lymph nodes. Yeah there's 30 comments in this thread. That mom got a lot of quick responses. Ask for this specific blood test, ask for this, call this A-T clinic."* – P15

As in these examples, participants learned how other families dealt with the disease progression, and used this information to initiate changes in their own child's care. Further, the community has developed a collective understanding of care norms, which they can then share with newer parents. By posting this information, caregivers have not only found recommendations for care strategies, but collectively the community has made important strides in better understanding the scope of the disease and its effects, which is valuable information when managing a rare condition:

> *"I had a concern because she tends to cry a lot in the night complaining that her leg hurts. So I didn't know if that was something specific to her, or something that other A-T kids have in common. And it tends to be that they do get a lot of pain in their body. It makes total sense because I think her muscle are always contracting and moving on their own. It's like she's working out all the time. And that was something I didn't see in Google or the doctors said. So we'll find more interesting data [on Facebook]."* – P5

> *"A lot of parents ask, you know, they get nervous when their child gets to a certain age. Around 7–9 they get fatigued and they stop eating. Everybody automatically thinks that their kid has cancer. You have to say, 'that is normal, you definitely want to go to your doctor, but don't be thinking worst case scenario. These are the symptoms that you need to be sort of looking for, for cancer.' "* – P10

Thus, the community has been developing a collective knowledge about the disease and developing norms for how to care for disease symptoms and progression. The group has developed a knowledge-base around how this genetic disorder most commonly presents, what symptoms and side effects can occur, and how the primary caregiver and the broader care team may respond. These insights helped participants to learn when and how to initiate care changes.

4.3.3 Sharing Provider Recommendations. Almost all participants described adverse events that they experienced or prevented. Despite the frequency of these events, we found few ways in which the community worked together to address these issues or systematically prevent known errors from occurring. Participants did share the contact information for A-T specialists when primary caregivers had a serious health question or issue, though these events were not indicative of an adverse event. Participants would also share local provider recommendations with one another, as described by one participant who shared an interaction she had with another parent:

> *"They were working with the same doctor. I go hold on. And I said the doctor's name, and she said yes. And I said, do not go back. Do not ever go back. And I got her hooked with our doctor. And she said, 'it turned our life around,' it's a whole different experience."* – P4

While parents would share provider recommendations with one another, this only occurred when families were living near one another. Many participants did not have other families living nearby, so we saw few examples of sharing local provider information. Participants did explain that parents would occasionally use the OHC to vent about negative experiences with particular healthcare providers, but others explicitly chose not to talk about these negative events within the community:

> *"I don't share that kind of stuff because I don't want it to be negative. And just because someone doesn't understand A-T doesn't necessarily make them a bad doctor. So I'll usually, like the gentleman who told me I was neurotic, we sent him a letter afterwards explaining that [child] actually had A-T and that was missed, and the CT scan he ordered was actually the opposite of helpful because it damages cells. You want recommendations before you see someone, but I try to keep it more positive."* – P15

Thus, we see a notable gap between the frequency with which caregivers were experiencing adverse clinical events, and the limited ways in which they discussed these events. We found no examples of sharing tools for addressing adverse events within the community, pointing to an important gap for resource development in this community and opportunity for future research.

4.4 Barriers to Online Community Support

As we outline above, participants' OHC played an important role in helping them work with a care team that was unfamiliar with their child's disease. Despite the benefits, we found that several participants infrequently used the Facebook group, or abandoned the OHC altogether. Here we explain three common reasons for this disengagement that emerged through the interviews.

4.4.1 The Emotional Burden of Participation. The most common issue participants identified with the online community was the emotional toll of participation. Parents would often use the group to receive social support, and would therefore share negative experiences and tragedy related to their child's life-limiting disease, which can have severe emotional consequences for other parents viewing this information:

> *"The news is not always good. You get a lot of bad news. You learn about other children and what's happened in their lives, and some of them die. It's crushing, it's painful. I know some parents I met, who were diagnosed around the same time as we were, they couldn't handle that and they actually unfriended some of the people and just kept in touch with the organizers. And I can understand that. It can be emotionally overwhelming."* – P1

In the above example, the participant understood that reading this information was too difficult for some parents, but chose to stay in the group to offer support to others. However, in some cases such information dissuaded parents from participating in the OHC further:

> *"It does get to me a lot that every time a child dies it's put out there. It's really hard because you get to know them. Every time that happens you put yourself in that position because you're like, that could have been my child. Then you start thinking, when will it be time for you to post that? The things that I've seen lately, they're too harsh. So I try to look away, for now."* – P5

Primary caregivers with young children who had not yet experienced much disease progression found these posts especially distressing and too emotionally burdensome. Unfortunately, due to the design of the group, community members could only opt to view all content shared within

the community, or none. Thus, parents must decide between OHC participation and their own emotional well-being.

4.4.2 High Barriers to Entry. Another challenge with using the online community was finding information about a particular topic. Participants commented that the community provided a quick way to receive answers to questions, because "someone's always awake". However, finding previous discussions or information was unsupported within the platform. To overcome this, many caregivers would monitor the group discussions and keep personal notes of information that they thought would be beneficial later. While this strategy was useful for participants, it places the additional burden on primary caregivers to develop strategies for organizing and recalling OHC content. Further, new members had no way to efficiently access prior information:

> *"When I first got on there I was going through old posts. I was on there forever... You can get lost. If you just go to that group and you're just scrolling."* – P2

Thus, primary caregivers had to develop their own methods for saving useful information shared within the OHC. This made the initial entry into the community overwhelming, and highlights the need for interactive features that allow community members to search for, find, and interact with the rich history of content created within the group.

4.4.3 "Every Kid is Different". The final challenge discussed by many participants was finding personally relevant documents and information, which could be difficult due to differences in illness presentations among children and care strategies. Identifying useful educational tools and disease progression information could be challenging as the symptoms of the disease differed between children. For example, while some children had immunodeficiency, others did not. This difference could affect one's approach to finding and recruiting care team members, school services, and daily health management strategies. Based on these differences, many participants talked about how the information they found on the Facebook group was not always relevant to their child:

> *"I'm on a Facebook page for parents. I try not to read too much into it because I know every kid is so different and experiencing very different things, but it's been a good place as a sounding board."* – P13

Beyond the disease variations, primary caregivers' approaches to managing the disease could also differ significantly. Several participants said they were interested in learning more from families who approached the disease and daily life in a similar way as they did. For instance, some families focused on providing their children with many services, such as various therapies, to help them manage the physical effects of the disease. Other primary caregivers chose not use as many therapies so their child would not miss out on school:

> *"I found that I am drawn to other parents who parent and are raising their kids like we did. I'm not drawn to someone who takes their child to therapy all the time. They're missing school."* – P11

Each of these differences can make it difficult for primary caregivers to identify which content would be personally useful. While the online community can be a great source for a variety of information and resources, participants often commented that at times they wanted help identifying resources that came from children with similar disease trajectories or parents with similar parenting styles. The inability to identify such information limited the overall usefulness of the OHC.

5 DISCUSSION

Through this qualitative study of A-T health management behaviors, roles, and resources, we found that primary caregivers' health management challenges were related to working with care teams that possessed limited knowledge of the disease itself, and that caregivers were required to take on additional care coordination tasks in response to these challenges. Use of an online community for A-T families supported this care coordination work, providing a space where caregivers could share information, resources, and strategies related to these care coordination tasks.

Collectively, these findings point to new opportunities for health tools to support individuals managing a rare disease. First, there are opportunities to design health tools that directly support the care coordination tasks outlined in this paper. Second, due to the value primary caregivers placed on the information and resources shared by their peers, a second opportunity is to consider ways for future health tools to increase access to, and interaction with, the knowledge shared by OHC members.

Support for chronic disease management is taking many forms, such as personal health technologies, online communities, and patient portals. Therefore, we do not focus on a particular type of technology, but rather consider guidelines for how future tools could better support the care coordination of rare diseases. We offer these recommendations (summarized in table 1 and elaborated on in the sections below) in order to motivate work that considers the unique support needs of patients and families who thus far have been largely omitted from the health technology design space [55].

5.1 Supporting Care Coordination Tasks

This study extends our understanding of the tasks that individuals take on when managing a rare chronic disease or working with care teams with limited knowledge about one's disease. A large body of literature has explored care coordination and health management of more common chronic illness, such as cancer, diabetes, heart disease, asthma, and many others. While these disease trajectories differ in many ways, such as symptoms and progression rates, the literature also highlights similarities in patients' and caregivers' health management responsibilities [5]. Common chronic disease management tasks include learning about the disease trajectory and planning for the future [9, 49, 64, 67], making treatment decisions and adhering to treatments [7, 50, 72], monitoring and accurately reporting symptoms and side effects [5, 18, 34, 50, 53], coping with the emotional impact of the disease [5, 9, 12, 49, 53, 54, 67], and bridging the communication gap between disconnected institutions [2, 17, 78]. These common responsibilities are relevant to the management of A-T. Primary caregivers were responsible for learning about A-T, monitoring their children's symptoms, and communicating with all members of the child's care team.

However, when managing a rare chronic disease like A-T, many of the primary caregivers were focused on additional care coordination tasks. Two of these roles, educating care teams and initiating care changes, required caregivers to continuously seek out up to date resources about the disease, trajectory, and care norms. These resources became the materials primary caregivers used to educate care team members about the disease and treatment options, and helped primary caregivers to make necessary treatment decisions. Supporting the development of materials to educate care teams about the disease is one way in which technology could help to reduce caregiver burden. In addition, one of the surprising insights from participants was the frequency with which they had to oppose clinical care decisions in order to prevent adverse events. Though we talked to a small group of parents, they often discussed multiple clinical errors which occurred or which they had to oppose directly. Helping primary caregivers to prevent these errors is a necessary step in addressing clinical safety issues [31, 85].

Table 1. Summary of Design Opportunities

Challenge	Design Opportunity
Primary caregivers responsible for care team education	Developing materials for care teams (5.1.1)
Infrequent sharing of adverse events	Explicit sharing of adverse clinical events (5.1.2)
High volume of OHC content makes finding relevant and timely content difficult	Personalized information recommendations (5.2.1)
Tension between OHC participation and members' emotional well-being	Providing control over content topics (5.2.2)

5.1.1 Developing Materials for Care Teams. The relative lack of research on rare diseases limits the information available for families and for care team members. Clinicians see rare disease care as a challenge and experience difficulties in accessing health information about these diseases [85]. Researchers have therefore suggested that technology can provide doctors who do not have disease-related expertise with succinct overviews of their patient's illness [55]. In addition to an overview of the disease, health tools could aggregate resources from specialists' clinics, research publications, and online communities. Up to date collections of these materials could directly support care team education, improve care quality for rare diseases, and reduce patients' and primary caregivers' burden of fulfilling this role.

Developing these materials presents two overarching challenges: synthesizing the complex body of information, and presenting this information in a way that is usable and useful in the time-sensitive medical context. Past work developing meaningful summaries of complex information networks may provide the necessary tools and methods for creating summaries from large collections of information and peer exchanges [1, 83]. This prior work provides mechanisms for synthesizing emergent knowledge within online forums. In parallel, researchers have developed design guidelines for creating information summaries that include patient generated data and are usable in time-sensitive healthcare settings [39, 63]. Bringing together these two areas of research presents one possible approach to integrating online content into materials that are useful to care team members in the context of busy, daily work schedules.

Developing materials that summarize online discussions may also help to provide a process for care teams, patients, and caregivers to collaboratively identify misinformation, a common problem with online health information sharing, while circumventing liability issues that may prohibit healthcare providers from engaging with online communities directly [39]. Importantly, sharing summaries of OHC content with care team members will require researchers and developers to consider the ethical tensions with sharing OHC content with non-community members, addressing issues of transparency, trust, and privacy. Individuals who post personal experiences and content within OHCs have varying perceptions of privacy and can be hesitant to share non-clinical information with healthcare providers [30, 40, 65]. Thus, while future work is needed to develop materials for care team members, researchers ought to consider ways to give community members a voice in deciding how their peer exchanges are used in the development of new materials, without adding undue burden onto the community.

5.1.2 Explicit Sharing of Adverse Clinical Events. Despite the high number of adverse events and clinical errors that participants encountered per our interviews, participants infrequently shared this information with their care teams or within the OHC. While some participants suggested that the OHC was used to share provider contact information or vent about negative interactions with providers, other participants said they purposefully did not share information about negative encounters because they did not want to suggest that the clinician was a bad person.

While parents may not wish to highlight particular clinicians or events, we believe there is an important opportunity to develop systematic ways to identify and report adverse events and normalize the sharing of these experiences. The design space for addressing adverse events is broad. Online community platforms and personal health information tools could afford patients and primary caregivers the ability to answer general, anonymous questions about adverse events or near misses, such as the type of care procedure involved. Likewise, healthcare centers could provide communal spaces to allow for faster sharing of these experiences. Collectively, these technologies could help in identifying patterns of events at the individual, community, and healthcare system levels, which can be addressed at a larger scale. For example, we had multiple caregivers report having had to inform clinicians that x-rays are unsafe to use in children with A-T (who have an increased sensitivity to ionizing radiation), perhaps suggesting that this information needs to be more widely shared with clinicians in their training or by parents early in their child's care process.

Lists of frequently asked questions used by peers can also help individuals learn which questions to ask healthcare providers in order to recognize when their care deviates from expected care [33]. We see this as a useful strategy for helping primary caregivers recognize adverse events before they occur. Future health tools could include features which aggregate questions or evidence that helped patients and primary caregivers when opposing care recommendations.

5.2 Increasing Access to Online Community Knowledge

While we did not set out to study online health community use, participants frequently described how resources shared within their OHC empowered primary caregivers to lead their child's care and educate care team members, primarily through the sharing of educational artifacts, care norms, and provider recommendations. Notably, the types of resources described by participants could at times be found through other sources. For example, rare disease foundations provide contact information for specialists, descriptions of common symptoms, and care procedures. Participants described using these information sources, such as the A-TCP and information from A-T clinics. However, participants frequently discussed resource sharing through the OHC. Through this platform, parents could receive useful resources, view resource variations and learn how they could be adapted to their child's needs, and see how other families operationalized these resources when addressing similar care challenges. The social, adaptive, and timely nature of these communities helped primary caregivers to access resources to address their immediate care challenges.

This work builds on literature examining how online health communities help people manage chronic conditions. Much of this literature has shown the important social and emotional support that individuals receive through these communities [60, 66, 70, 80, 81]. In addition, OHCs provide a space where people can receive information to address self-care challenges and develop a collective knowledge of a disease [38, 59, 84]. Through this emotional and informational support, online health communities are influencing everyday health behaviors such as the development of physical and emotional coping strategies [37, 58, 82]. Our work extends our understanding of how peer exchanges within OHCs influence offline behaviors, demonstrating how primary caregivers used information exchanged within an OHC to support care coordination tasks.

Notably, many OHC platforms are not designed to facilitate a rich exchange of ideas and resources [59]. We found that emotional content, limited ability to view older content, and lack of

connections between post content and poster details can limit OHC participation and use. Similar challenges have been discussed on a variety of platforms, including issues of information overload [44, 65] and information utility [35, 43]. We found that the issue of emotional readiness was less prevalent in OHC literature, but the negative consequences of health information have been considered in studies looking at other information sources [10].

Based on the prominence of these issues, an important research agenda is considering how new technologies, from personal health tools to large community-based platforms, may circumvent existing barriers. We propose two lessons that emerge from this study. First, health tools need to overcome issues related to content volume, which can be overwhelming and make it difficult to learn from previously shared experiences and insights. Second, health tools need to allow individuals to control the types of content they wish to view or to avoid.

5.2.1 Personalized Information Recommendations. Participants discussed how the high volume of content shared online made initial entry into the group overwhelming, and consistently made finding personally relevant information difficult. These barriers highlight the need for tools that allow patients and caregivers to find personally relevant content at the time in which it would be most useful. Participants shared many examples in which timely, personalized information could be beneficial when working with their care teams. For instance, receiving example IEPs prior to meeting with a new school system could help inform both parents and the school about what resources may help the child in their daily education.

Recent studies have looked at using personalized information recommendation systems to connect individuals with content that can help them address their immediate health management challenges [41, 61]. These tools aggregate online health information and utilize user input to connect individuals to small sets of resources that address their immediate health questions or challenges.

Our work suggests that there is a need for personalized information systems for rare disease management and that such systems could be particularly effective if they incorporated both clinical resources and community insights. Our findings also suggest that primary caregivers benefit from viewing diverse sets of artifacts and care strategies, so that they may consider how to adapt recommendations to their specific context. Supporting this adaptive behavior is particularly important in complex pediatric conditions, such as A-T, because the disease symptoms and trajectories can be highly heterogeneous. Personalized information tools ought to present diverse—yet relevant—sets of resources to help caregivers compare and contrast similar types of content and identify resources that are most pertinent to them. Such summaries could have the additional benefit of making community knowledge more accessible to individuals who do not have the means to engage with online communities or are uncomfortable engaging with these communities directly.

5.2.2 Providing Control over Content Topics. The tension between OHC participation and parents' emotional well-being suggests that patients and primary caregivers require greater control over the informational topics with which they wish to engage, particularly in the context of progressive, life-limiting conditions. Issues of distress caused by health information have been documented across multiple forms of support, including online health communities, patient portals, and paper-based informational materials [44, 51, 77]. Designers of health tools ought to be considerate of issues related to emotional readiness and provide individuals with the autonomy to decide which information they wish to view or to avoid.

One way to address these issues is through lightweight features that visualize an information space and then allow people to control which topics they wish to engage and disengage from, such as topics related to loss and grief. Researchers working on facilitating online creativity have developed a viable approach for allowing people to create personally meaningful topics [73]. In this approach, an individual can quickly develop their own clusters of related content as they

look through information to satisfy their own information needs. As more of these clusters are created, they may be used to develop global semantic models, which could be used to recommend common topics. This would put less up front work on the individual to create topics, allowing them to more quickly identify topics they wish to view or avoid. Such an approach could be useful in developing personal health information tools, new community platforms, or even for determining which paper-based materials individuals would like to receive from their healthcare providers.

5.3 Generalizability

This study presents an in-depth case study of primary caregivers' strategies for working with care teams with limited experience treating their child's rare disease. While we focused on one particular rare disease, several studies have cited a lack of clinical expertise as a health management challenge across rare diseases [8, 55]. Therefore, up to 30 million people in the United States [85] are likely working with care teams with limited experience caring for their particular diagnosis. Yet, this challenge has been largely unaddressed in health and design literature.

Working with under-informed care teams is not a unique challenge of rare disease management. Other communities face similar challenges accessing knowledgeable care teams, such as transgender people and individuals with multiple chronic conditions [2, 48]. More work is needed to understand patient and caregivers health management practices in other sociotechnical healthcare systems with limited disease-related expertise, including systems outside of the United States. We anticipate that the responsibilities described in this study, such as educating care teams and addressing adverse clinical events are common in such settings due to the ways in which insufficient expertise of the healthcare providers has been shown to change the medical encounter [8]. The design opportunities suggested in this work may therefore be beneficial for a broader audience.

6 LIMITATIONS

Our study has a number of limitations. First, our study may have a selection bias, as all participants were connected with the A-TCP. More work is needed to understand if the findings we present here apply for other primary caregivers who are not connected to this organization. Future work should also explore the validity of these findings in other communities. In particular, we see an important opportunity to look at how to support language and literacy minority families. We also did not include the perspectives of clinicians and care teams in our data collection. In future studies, we hope to work with care team members to understand their perceptions of caring for a child with a rare disease. This study also focuses on primary caregivers' perceptions of the benefits and barriers of using an OHC. We have yet to study the content shared within the Facebook group.

7 CONCLUSION

Research on chronic disease management has shown that care teams play an important role in educating patients about the disease and treatments, initiating necessary changes in care as diseases progress, and determining the most appropriate treatments for an individual. Our study shows that when care teams have limited disease-related expertise, primary caregivers assume these responsibilities. Resources such as educational artifacts, care norms, and provider information can help caregivers take on these roles. Online health communities provide an important space where caregivers can share these resources, see how resources among community members are adapted to the specific needs of the child, and learn how other families have operationalized these resources to help them manage their care teams. We believe an important research agenda is creating tools to support these care coordination tasks and access to online community knowledge. Such research is a critical step in supporting communities who experience barriers accessing medical expertise.

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