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ORIGINAL RESEARCH

Alone in a Crowd? Parents of Children with Rare Diseases' Experiences of Navigating the Healthcare System

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Abstract

A disorder is considered a rare disease if it affects 1 in 2000, hence, while independently unique, collectively, these conditions are quite common. Many rare diseases are diagnosed during childhood, and therefore parents become primary caregivers in addition to their parental role. Despite the prevalence of rare diseases among children, there has been little research focused on parents' experiences of navigating the healthcare system, a gap we begin to address in this study. Guided by an interpretive description methodology, participants were recruited through online listservs and posting flyers at a pediatric hospital in Western Canada. Sixteen parents (15 mothers and 1 father) participated in in-depth, semi-structured interviews between April 2013 and March 2014. Data were analyzed inductively, generating the main study themes. Findings illuminated the challenges parents' experienced on their child's diagnostic journey—from seeking, to receiving, to adjusting to the rare disease diagnosis. Following diagnosis, gaps, and barriers to services resulted in parents pursuing services that could support their child's unique care needs, which often resulted in out-of-pocket payments and changes to employment. Parents found peer support, both online and in person, to be an effective resource. This study illustrates the common challenges experienced by parents of children with rare diseases as they navigate the healthcare system. Parents' role as "expert caregiver" was rarely acknowledged by healthcare providers, pointing to the need to foster more egalitarian relationships. As well, parents were burdened with the additional role of care coordinator, a role that could be filled formally by a healthcare provider. Lastly, peer support was a key resource in terms of information and emotional support for parents who often begin their journey feeling isolated and alone. Policies and programs are needed that validate the invisible care work of parents and ensure adequate formal supports are in place to mitigate potential sources of inequity for these families. Furthermore, genetic counselors can play a key role in ensuring parents' informational needs are addressed at the time of diagnosis and in connecting families who share common experiences regardless of the rare disease diagnosis.

Keywords Rare disease · Family caregiving · Healthcare system · Navigation · Qualitative · Patient experience

Introduction

Rare disease (RD) is a term used to describe a very heterogeneous group of disorders, which can affect any body system (Schieppati et al. 2008). A diagnosis is considered rare if it affects 1 in 2000 people (Beaulieu et al. 2014). In Canada, for example, 1 in 12 people have 1 of the 7000 diagnoses classified rare genetic diseases and even more go without formal diagnosis (Beaulieu et al. 2014). A large percentage of RDs (roughly 80%) affect children (Dodge et al. 2011) and almost half of all RDs have their onset in childhood (Zurynski et al. 2008), which in turn has a significant impact on the well-being of families.

While individually these diseases are rare, they collectively result in a significant number of childhood illnesses, deaths, and associated healthcare costs. Rare diseases result in a wide variety of healthcare needs, stemming from the involvement of multi-organ systems and cognitive and/or developmental issues. The symptoms that children with RDs experience may require the support of a variety of services, multiple healthcare professionals, equipment support, and orphan drug therapies (Zurynski et al. 2008). While important research is gaining traction on discovering the genetic aetiology of RDs

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(Beaulieu et al. 2014) and orphan drug policy research is growing (Herder and Krahn 2016; Menon et al. 2015), there is a gap in research about the experiences of families caring for a child with a RD.

Rare diseases significantly impact the economic, psychosocial, and physical well-being of individuals and their family members (Zurynski et al. 2008). Rare diseases have unique factors, which necessarily impact family health. Most broadly, individuals with RDs and their family members often have limited evidence-based information to guide decisions about disease management and symptom relief (Anderson et al. 2013; Forsythe et al. 2014). Further, the inherent uncertainty that comes with having a RD, including delays in diagnosis and a lack of knowledge about current and future care needs (Grut and Kvam 2013; Huyard 2009), impact access to services and management of the RD (Dellve et al. 2006). Research on the experience of having a RD indicates that care and service needs are often not determined simply by the severity of the health condition, but rather are a combination of poor quality of care and barriers to access (Farmer et al. 2004; Huyard 2009).

Research conducted with families of children with RDs has found a high level of parental stress associated with the intensive care needs of this population (Dellve et al. 2006). Parents, particularly those of children not-yet-diagnosed, also experience depression and anxiety (McConkie-Rosell et al. 2018). The experience of caring for a child with a RD often parallels research conducted with parents of children with disabilities. These studies suggest that family adaptation to a child's disability is complex and a range of factors and resources, both internally and externally, impact family health and well-being (Dellve et al. 2006; Neely-Barnes and Dia 2008). As well, parents of children who are not yet diagnosed emphasize their feelings of uncertainty and an inability to make plans, which affects overall family health (Spillmann et al. 2017). In a scoping review of the unmet needs of parents of children with RDs, the authors identified social, informational, and emotional needs as the most pressing. They also suggested the need for more research in this area to better address the unique care needs of these families (Pelentsov et al. 2015). The purpose of this study, therefore, was to explore parents' experiences of navigating the healthcare system for their child with a RD.

Methods

The methodology guiding this study was interpretive description. This approach was useful in that it allowed us, as researchers, to go beyond description to explore meaning and explanations of the lived experience of parents of children with RDs. Interpretive description does not require using a theoretical framework per se, but rather is a pragmatic methodology focused on generating findings that are useful for clinical practice, programming, and policy development (Thorne et al. 2004). The focus on practical applications makes this methodology suitable for research that can inform genetic counseling practice.

Setting and Participant Recruitment

A convenience sample of participants was recruited from Western and Central Canada. From the outset, we chose to have a broad geographical recruitment strategy given the unique study population, but to remain in Canada as families would experience some similarities in healthcare delivery across the country. To recruit participants, information about the study was shared on a closed listserv for families of children with RDs in Canada and flyers were posted in a pediatric hospital in Vancouver, British Columbia. Participants were also recruited through word of mouth from individuals who participated in the study. Inclusion criteria for participation were (i) live with infant/child/youth diagnosed with a RD (defined as lifetime prevalence of 1 in 2000) or was not yet diagnosed due to the rarity of their condition and (ii) ability to converse in English. Through this recruitment approach, 16 parents (15 mothers, 1 father) participated in the study.

Data Collection

Data were collected using semi-structured interviews between April 2013 and March 2014. Semi-structured interviews allow for the generation of rich, in-depth data, while using an interview guide to direct the content (Ryan et al. 2009). In this study, participants were offered the option of being interviewed in-person or by phone. Only three parents chose to be interviewed in person and this included a mother/father dyad; all other participants either chose to be interviewed by phone or were geographically distant from the researchers.

The interview involved open-ended questions and prompts, which encouraged narrative accounts of the topic. The main interview questions were as follows: "Describe the process of your child becoming diagnosed with a rare disease," "Can you tell me about your experience of obtaining healthcare services and supports for your child?," and "Are there additional supports that have benefitted your family in terms of navigating the healthcare system in addition to those obtained through the healthcare system?" Each interview question had a series of probes to use if needed. A benefit of using probes is that they allow for the clarification of interesting and pertinent issues raised by participants and gives the interviewer the ability to explore and clarify inconsistencies within and between respondents' answers (Barriball and While 1994). Demographic information was also collected during the interview.

The interviews ranged between 22 and 118 min and were an average of 59 min in length. Data collection continued until informational redundancy was achieved (Sandelowski 1995).

All interviews were recorded and transcribed verbatim. Participants were assigned a code to provide anonymity and confidentiality. In the Findings section, participants are identified by their code (e.g., P1) and are given a brief descriptor the first time they are quoted (e.g., P1, 48-year-old mother to a 14-year-old son).

Data Analysis

The data were analyzed using a constant comparative and iterative approach (McAllister 2001; Thorne et al. 2004). The first and second authors conducted the interviews, and all of the authors individually read and hand-coded the data. Codes were then compared collectively to develop and evolve the codebook. In this manner, emerging themes from concurrent analysis informed subsequent data collection, which in turn helped to improve the validity of analysis. Interviews were coded for emerging themes in keeping with the research topic at hand. These themes were verified during subsequent interviews through questions and probes introduced by the interviewer. Rigor was therefore embedded within the data collection and analytical processes.

Findings

Sixteen parents of children with RDs participated in the study. Fifteen were mothers and 1 was a father. Their ages at the time of the interview ranged from 29 to 48, with an average age of 40 years. Participants were located in British Columbia (N = 11), Ontario (N = 1), Manitoba (N = 3), and Quebec (N = 1). The participants reported having 17 children with RD diagnoses, (4 female, 13 male). Nine children were under 5 years, five were between 5 and 10 years, and three were between 10 and 20 years. Given that reporting the child's RD diagnosis could potentially identify participants, we have not included specific information about their diagnoses (see Tables 1 and 2 for further demographic information).

The findings illustrate that parents shared common experiences of navigating the healthcare system despite the uniqueness of their children's diagnoses. Three main themes emerged including the following: "the diagnostic journey," "seeking and accessing services," and "peer support." Each theme was comprised of several sub-themes which illustrated families' experiences.

The Diagnostic Journey

All of the participants shared their narrative of the diagnostic journey. This theme was comprised of the linear events from "seeking a diagnosis" through the experience of receiving and adjusting to a diagnosis. One family in the study was still on this journey, as their child was not yet diagnosed, due to the rarity of their condition.

Table 1 Participant demographics

	Total sample (N = 16)	Percentage
Age (years) (missing 2)		
Mean	40	
Range	29–48	
Gender		
Female	15	94
Male	1	6
Education level		
High school diploma	1	6
College diploma	6	38
Bachelor degree	6	38
Master degree	2	12
Other	1	6
Employment status		
Full-time	7	44
Part-time	3	19
Not employed	2	12
On paid leave (e.g., parental leave	3	19
Other	1	6
Annual income (single)		
< $30,000	3	19
$31,000–$40,000	2	12
$41,000–$50,000	0	0
$51,000–$60,000	0	0
$61,000–$70,000	1	6
$71,000–$80,000	0	0
$81,000–$90,000	1	6
$91,000–$100,000	2	12
> $101,000	4	25
Unknown/did not answer	3	19
Language primarily spoken at home (missing 2)		
English	13	93
Arabic	1	7
Number of people living in household (missing 1)		
Mean	4	
Range	3–7	
Relationship child(ren) with RD		
Mother	15	94
Father	1	6

Seeking a Diagnosis

Parents were seekers on this journey. In the initial part of their journey, they were seeking a diagnosis, which often emerged to be a complicated experience. Participants spoke about numerous doctors' appointments and persistence in their pursuit of a definitive diagnosis for their child.

As some participants sought diagnoses, they felt blamed for the symptoms by healthcare providers. P3 (47-year-old

Table 2 Demographic information about child with Rare Disease

	Total sample ($N = 17$)	Percentage
Age (years)		
Mean	6	
Range	6 weeks—16	
Gender		
Male	13	72
Female	4	28
Age of child at diagnosis in years (missing data on 1 child)		
Range	At birth—5	
Mean	1.6	
In utero	4	24
Not yet diagnosed	1	6

mother to a 16-year-old son) said "The GI team at the hospital insisted that what was wrong with [child] was functional and our fault, in fact, because we weren't being strict enough with this little person who was two in terms of helping him clear out his GI tract." And went on to say "It was terrible and it was frustrating and at one point I had a nurse accuse me of Munchausen Syndrome By Proxy, and that was awful." Despite being blamed both explicitly and implicitly for their children's symptoms, parents were persistent in their pursuit of a diagnosis. Other participants reported that their children were blamed for their symptoms:

> Everybody at the hospital kept trying to say, "We know what's wrong with him and why his bowels aren't working. He's choosing to not go to the bathroom." You know, "It's all in his head" is what I was told by a certain doctor. And he was little when we first approached the doctor saying, "He has this problem and he's sick all the time." I mean, a two-year-old doesn't really choose to make himself sick over going to the bathroom (P7, 42-year-old mother to a 5-year-old son and 7-year-old daughter)

Despite the blaming of themselves and their children, parents continued to pursue a definitive diagnosis. P7 described the process: "alot of years and a lot of arguing and a lot of just begging and pleading for somebody to please take me seriously." For some parents, the journey to diagnosis was not done, "So yeah, so we were involved in some kind of a study after I beat my endocrinologist to death because she wasn't [spending too much time with our stuff]. And we got involved in some kind of a study. He hasn't showed anything so they haven't found anything so far" (P15, 42-year-old mother to a 2½-year-old son). For many, this was a long, arduous, and drawn-out process that involved appointments with many different specialists as well as visits to the Emergency Department.

Receiving a Diagnosis (or Not)

With the exception of one participant, parents in the study had finally received a diagnosis for their child. However, even once a diagnosis was determined, there remained a lot of unknowns about how to proceed with treatment given the rarity of the conditions. P8 (47-year-old mother to a 12-year-old son) described the experience:

> [Son] wasn't meeting his milestones, so the pediatrician sent us to the geneticist, and that's when they diagnosed him and basically gave us the diagnosis and said, "Here. Go out in the world. You'll have services. People will help you through the daycare, and then they'll help you through the school, and that's it." And they pretty much washed our hands of us. From genetics. Our pediatrician, too, was, "Oh, as long as he's progressing and improving, that's all we can ask for.

Other parents had similar experiences at the time of diagnosis. P1 shared her experience

> Asking, "what does this mean, we know he has the [gene]. What's going to happen? What does this mean?" And basically the advice we got is, "We don't know. He's writing the book." So it's not like there's somebody else ahead of him that we can say, "Okay, we got to watch for this. We got to look for this. We can do this." It's, "We got to wait and see what happens with him and then he's writing the book for others."

Parents of children who were still undiagnosed had similar experiences at a certain point on the diagnostic journey:

> But [doctors] basically said, "Sorry, he's going to have developmental delays." "Well, what does that mean?" "Don't know. Go home. Take care of him." So the first three months of his life, he's taking feeds, he's gaining weight, we had to adjust his seizure meds, up and down, up and down, this one, that one. So I pretty much spent … I had never, ever witnessed a seizure in my life and having this baby seize … every two days, we were at the hospital. Finally, they declared him epileptic. I'm like, "Okay, at least he's got one name for what he has." (P12, mother, age not provided, to a 7-year-old son)

Hence, receiving a label or diagnosis was not the same as receiving help. Many participants felt that they were left with the responsibility of figuring out their next steps. As P2 (41-year-old mother to a 2-year-old daughter) shared, "I feel like it shouldn't be a family that's receiving a diagnosis, a devastating diagnosis, to then have to figure out what services they need to get." Another participant also described the sense of

isolation immediately following the delivery of the diagnosis: "In the hospital they didn't offer to have anyone come talk to us after about how we were feeling. And that was kind of disappointing. You're just kind of … after everyone finally leaves at the end of the day, you're just kind of left there and at that point you're emotionally exhausted and you're just kind of left." (P11, 29-year-old mother to a 6-week-old son).

This sense of "aloneness" following diagnosis was echoed by other participants. For example, P4 (30-year-old mother to a 1¾-year-old daughter) said "Even if they had given us some sort of connection to a group or to certain people, either when we first got the diagnosis or for right after she was born, I think that connection would've been huge." Similarly, P9 (36-year-old mother to a 5½-year-old son) explained, "Definitely, the first year of [child]'s life, I was trying to gain knowledge about what his disease was all about and how to manage it but I just didn't know that there were other resources available to me that he would qualify for." The general lack of guidance or follow-up may also have unintended consequences for delaying treatment or accessing valuable community-based supports for these families.

After the Diagnosis

Following a diagnosis, there was a period of adjustment. This period involved emotional adjustment to the implications of the RD diagnosis for their child and for their family. Parents also began engaging in a variety of activities focused on seeking knowledge about the diagnosis, and becoming care coordinators.

The initial period following diagnosis involved an emotional adjustment. Eleven of the parents described feelings of isolation and lack of belongingness. A participant described her feelings when taking part in typical groups for new mothers, "I have felt at times that I want to belong with … just be a regular mom with a regular kid and craved that experience. And then when I'm there, there's times where I just realize that we are different or non-typical experience, and it's so obvious and apparent, that it is hard to participate" (P13, mother, age not provided, to a son, age not provided). Another participant described the isolation even in a large geographic area: "There's no one else that also has this disease. Even in [Province], there's very few. I'd be so excited, like, to meet someone [laughs], how sad." (P14, 40-year-old mother to a 5-year-old son and a 6-year-old daughter).

The post-diagnosis period was also characterized by seeking knowledge. Many parents wanted more information from healthcare providers about their child's RD, including biomedical information and also availability of service supports in the community. Most frequently, participants wanted tangible information about *relevant* support services available for their child and family and, *how* they should access them. P4 said, "most healthcare providers were learning with us, I guess. We didn't get a lot of information. We have a lot of information now, but a lot of that has been us finding that out on our own." The lack of professional guidance in seeking knowledge meant that parents had to evaluate the quality of information they were finding, which often came from the internet. Many participants expressed frustration regarding the lack of information resources provided by healthcare providers and were worried about the quality of information they found. As P11 noted: "So you're kind of left to find this information on your own and you don't know if it's accurate. So you never know if you're actually reading the correct information. It would be nice if they say, okay, your child has this rare disease, here's a website. So not only does my child have this rare life limiting disease, but I don't know what to watch for." Parents tried to be critical of the information they gathered and looked for what they considered to be more reputable sources from scientific journals, RD literature, and connected with healthcare providers with specialized knowledge in the area.

During the post-diagnosis periods, parents also adopted the role of care coordinator, and 14 of the participants described this role in detail during their interview. Given the complex health needs of their children, most families had interactions with many specialists and health services. A common experience was the lack of formal care coordination and communication between healthcare providers. A participant described her experience:

> And then all of a sudden [genetics specialist] dropped me like a hot potato. She says, "You have to deal with your pediatrician." And the pediatrician, like I said, he's retiring. Every time I ask him for a referral or something, "I can't do it. They have to do it. The specialist has to do it. They have to do this. It's not mine." Always passing the buck to somebody else. (P8)

So parents found themselves being pushed back and forth between providers, without anyone taking responsibility for their child's overall care. Another participant succinctly described the situation, "you have a whole team, but nobody's playing together [laughs]." (P14) Ultimately, participants recognized that they would need to take on the role of care coordinator, "You have to be a case manager, because nobody does it. Everybody just wanders around, [go see a good doctor, go see a bad doctor], they don't talk to each other." (P15)

The overall lack of coordination could contribute to delays in accessing services, as well the delivery of services that were ineffective. Participants were very aware that their child's pediatrician was limited in their capacity to coordinate services; however, they felt that they needed help. As P13 said,

> Somebody says, "Okay, you have that going on and this and, you should think about phoning this person and

> doing …" And I said that to my pediatrician and she says, "Well, that's my job." "Well, then you could maybe just bump it up a little bit?" … And I think she's wonderful, she's a really great paediatrician. We're very lucky to have her. But she doesn't have the time to be that person. There needs to be another person whose job is just the big picture.

All of the participants noted that they were the ones who coordinated their child's healthcare providers and ensured that they were communicating with one another and relaying important information. However, participants found this role burdensome and overwhelming and wanted assistance.

Seeking and Accessing Services

Another theme involved participants' experiences with seeking and accessing services in the healthcare system. These services included referrals to medical specialists, therapy from allied healthcare providers (e.g., physio, occupational, and speech therapists) in the community, as well as programs that facilitate inclusion in educational and other social settings. Due to the lack of formal care coordination, as described previously, eight participants were not even aware of the existence of these community-based programs. As said by P9 "[social worker] was talking about this child development program. Well, that's the first time I had ever heard about it. Nobody has ever told me anything about it before and all the resources that they have available."

In order to access some services, a child must have a diagnosis. When asked about this issue, P1 said "We're hoping … actually, [a diagnosis] would be good because then [child] would get more funding for speech or physical therapy or OT or whatever, so that would be good." She went on to explain that her child was not able to access additional staffing supports at school or respite funding because of the lack of formal medical diagnosis.

Even when their child had a RD diagnosis, all of the participants spoke at length about the barriers they faced in accessing services. They noted that sometimes the qualification processes they had to follow to access services were challenging to understand and could create contentious relationships with healthcare providers. As P2 described, "sometimes you have to fight the doctors as well. That's kind of tricky. You shouldn't have to fight with doctors to get services for your child." Further, the rarity of the condition meant that many providers were unfamiliar with the condition and therefore did not necessarily understand what services were required, further hindering access. In particular, specialist services were one area that many participants experienced care access barriers. Some participants experienced challenges in receiving referrals to specialists that they felt would help their child. As one participant reflected, "It's been huge hurdles just sort of getting to see the specialists that they need to be seeing, but now we're seeing them." (P7)

Beyond the primary RD diagnosis, participants noted that secondary diagnoses (e.g., attention deficit hyperactivity disorder, autism spectrum disorder) were sometimes more helpful in accessing services because there were dedicated resources for these diagnoses. As noted by P8, "There are different programs and whatnot, but unless you have the autism diagnosis, you can't get into any of that stuff. So that would have helped much with early intervention years ago." This silo approach to services also meant that participants had to advocate for their child to be assessed for a particular secondary diagnosis in order to access much needed services.

A major barrier in the process of accessing supports was qualifying for the associated funding. In order to qualify, participants often required documentation from healthcare providers, typically physicians. This situation meant that certain healthcare providers acted as gatekeepers to valuable funding for services, "So once you get proper diagnosis, you have to get this form and get your doctor to fill it out. Some doctors don't fill them out. So I had to have a discussion with my child's paediatrician that he had to fill it out." (P2) The physician also completed forms to access additional staffing supports at school. P7 reflected on her conversation with her child's pediatrician "The pediatrician yesterday had said she doesn't think he's disabled enough to qualify for this [school] funding. She had said in order for the school to be able to access funding to apply for extra services for him, he would have to meet this [criteria]. She said he won't. But once he starts school, they will have problems and looking for more help." In this situation, the parent had a different perspective on the child's abilities than the pediatrician; however, it was ultimately the pediatrician's decision whether or not to apply for funding. This situation was very disempowering and frustrating for participants.

Accessing services also had implications for a family's financial resources. Many families paid out-of-pocket for medications and therapies. P16 (35-year-old mother to a 8-year-old son) described the financial strain on families: "you have to be at the point where you've got the 'For Sale' sign outside, and you're not working, and it's falling apart, and your debt's really high. And that's what's happened to us, where it's going to take us years to get out of that hole, and you know, we're going to do whatever we need to for [child.]" Another participant (P14) reported spending $1000 per month on a medication that was not covered by government programs. Her child was taken off the medication when their symptoms subsided in order to give the family a break from paying for it. However, she anticipated needing it again in the future. In addition to costs for therapies and medications, there were incidental costs. P13 reported spending over $2000 the previous year to pay for parking at the hospital where the specialists' offices were located.

In addition to out-of-pocket costs, accessing services also led to employment changes for some participants. P3 reported missing months of work and leaving the workforce due to her child's hospitalizations. P12 shared her experience of losing her job:

> One day, my boss pulled me aside and said, "I'm sorry that you have a child with disabilities, but that's not my problem. And tough that you had this happen to you; it's not fair, but you can't really work in the store anymore. And you're depressing the staff. They're constantly thinking about you and clients are constantly asking about you."

P13 summed up the time involved in accessing services, care coordination, and managing her child's appointments, comparing it to paid employment: "just organizing and phoning back appointments that can be 20 hours for me a week … that is my part-time job."

Peer Support

Informal peer support from other parents of children with RDs emerged as a key resource as participants navigated the healthcare system. Fourteen of the participants described receiving valuable support through informal networks, and this support mitigated some of the shortcomings of the formal healthcare system. Even though most of the children had unique diagnoses, the participants found that their experiences of navigation and coordination of healthcare services were similar.

A valuable aspect of having access to other parents' expertise was their availability. As P1 described "problems don't happen just two hours once a month. You're dealing with something that problems are coming up on a daily basis of some sort or another and you don't want to have to wait three weeks to get an answer." Many of the parents were connected through social media platforms and were able to communicate regularly in order to exchange information and support. Social media was also considered a better way to connect because their children's intense care needs often meant parents were not able to attend groups in person. A variety of online platforms were used including email lists, Facebook groups, blogs, and forums. As noted by P7, "I talk to [other parents] on the phone. We spend a lot of time on the computer talking to each other. So, that's a huge part of my support system." They also used the online groups as a springboard for in-person get-togethers. P10 (48-year-old mother to a 4½-year-old son) described how her group worked:

> The mum's group actually formed a new, separate mum's group and it's a private facebook group and we meet once a month for a meeting and twice a month at a pub. We're all mothers … it's all run by us and we support each other. We got tired of agencies all telling us how we should do it or we didn't meet criteria. We were, like, screw you. So the biggest support has come through other mothers with kids with special needs.

Participants also found other parents to be excellent sources of information. P2 said "you don't know what's out there without asking the other moms. So I just asked other moms what they were doing for their kids, and that helped a lot." Using the internet to connect meant that some participants could "meet" other parents whose children had a similar or the same RD even if they were geographically distance. P4 described this experience, "we got a lot of information from other parents on there. And they're from all over the world."

Many participants were members of RD-related organizations, patient associations, and/or parent advocacy groups. Participants often found out about these groups through a peer's referral or through their own searching, not by referral by a healthcare provider. For these parents, meeting others in person had a powerful impact. P5 (44-year-old mother to a 4-year-old daughter) shared "just meeting the other families was huge for us, you know, just seeing the other parents going, 'Oh, they're still sane. They're still okay.'" All participants spoke of the benefits of meeting parents of children with similar conditions or who had the same condition. Many of them also found that peer-to-peer interaction provided them with a sense that they were not alone.

Discussion

This study sheds light on the experiences of parents of children with RDs as they navigate key activities in the healthcare system. In particular, parents' experienced challenges in seeking and receiving a diagnosis for their child, and then subsequently accessing services. Their information needs were not adequately addressed by general practitioners or specialists, including geneticists. However, the inadequate information sharing from genetic healthcare providers is likely in part due to the lack of knowledge about newly recognized RDs. The lack of knowledge on the part of key providers further emphasizes the importance of peer support in the journeys of families of children with RDs, in particular the common experiences of families regardless of the child's actual diagnosis. These finding have implications for genetic counselors' practice, as they are often a key contact for parents before and at the time of diagnosis.

Despite the heterogeneity of the RDs among the participants, they reflect common experiences and needs, including larger systemic issues and moral imperatives regarding quality of care and access. Participants' narratives revealed challenging experiences, specifically related to the provision (or lack thereof) of information at time of diagnoses, barriers in

accessing certain services, and poor care coordination. Our findings parallel the small but growing body of research about the complex challenges of navigating services, accessing appropriate and effective care (Budych et al. 2012; Grut and Kvam 2013; Huyard 2009) and the resultant challenging psychosocial, social, and financial impacts that RDs can have on families (Anderson et al. 2013; Dellve et al. 2006; Spillmann et al. 2017). Given that genetic counselors are in contact with many families, they are in a key position to assist parents with their informational needs before and at the time of diagnosis. Counselors can assist parents to identify high-quality sources of information about their child's diagnosis online, where most parents go to search for assistance in understanding their child's diagnosis. As well, counselors can ensure that parents have information about key resources and formal supports available to families of children with RDs.

Given the lack of knowledge, treatments, and research on RDs, many participants in this study had to actively seek out information on their child's disorder, which is not unusual for those in the RD community (Kesselheim et al. 2015). In a systematic review of qualitative research about living with a RD, the notion of "expert patient" was identified (von der Lippe et al. 2017). In our study, parents became "expert caregivers." Despite the toll on their own mental health, parents tend to remain active in managing their child's health (McConkie-Rosell et al. 2018). As with the participants in this study, previous research has indicated that healthcare providers often reject or struggle with accepting their patients' expertise about their own diagnosis (Smith et al. 2015). This can contribute to fractured care and negative outcomes for individuals with RDs. Further, participants in this study took on the role of care coordinator in order to improve care continuity and fill gaps in healthcare provider communication deficits. Again, this is congruent with previous research with individuals with RDs (von der Lippe et al. 2017). Given that parents of children who are not-yet-diagnosed are similar to parents of children with chronic illnesses (McConkie-Rosell et al. 2018), in that they may experience depression and anxiety, genetic counselors are in a key position to conduct screening and help direct parents to resources to support their mental health.

Given these gaps in care, particularly around knowledge and care coordination, our findings echo the broader call for improvements in patient and family engagement in care, including clinical decision-making (Barry and Edgman-Levitan 2012). Family-centered care is a philosophical approach that involves supporting and consulting with parents as experts in care for their child, as well as developing parent's knowledge, skills, and self-efficacy to provide and manage care (Dellve et al. 2006; MacKean et al. 2005). In the context of RD, fostering a more egalitarian relationship between healthcare providers and parents is particularly salient, given that parents often have as much, if not more, knowledge of their child's diagnosis than generalist providers. Recognizing and validating the role of "expert patient," or in this case, "expert caregiver," must be central to care provision for those with RDs. Genetic counselors are well positioned to advocate for and facilitate relations between parents and healthcare providers involved in their child's treatment.

Out-of-pocket payments and changes to employment were common among participants in this study. While research in cancer care has also identified out-of-pocket payments as an unanticipated consequence of illness for Canadian patients (Housser et al. 2013; Longo and Bereza 2011), unlike cancer treatments which are often time-limited, the participants in our study anticipated these costs for the duration of their children's lives. There was also a gendered aspect of this finding, as the majority of participants were women ($N = 15$) and many reported either decreasing their employment to part-time or leaving the workforce entirely in order to care for their children during times when they were hospitalized or to attend numerous appointments. These findings warrant further investigation regarding the intersection of gender and caregiving, as the financial impact of RD on families, and the potentially disproportionate impact on women, has not been well researched.

For many participants, issues regarding the deficits in access and service support resulted in increasing caregiver advocacy, as participants became actively involved in promoting their own child's care and the interests of other parents of children with RDs. A number of studies have highlighted how family and patient engagement in advocacy and research transform the distribution of power between experts and lay people, and consequently contributes to the development of new modes of care delivery (Rabeharisoa et al. 2014). This study builds on increasing call for meaningful partnerships and also recognition of the role of individuals with RDs and their families with regard to policy development, clinical decision-making, and research related to advancing RD care.

Similar to research with families of children with disabilities (Farmer et al. 2004; Woodgate et al. 2012), the findings highlight the value and importance of peer support and collective advocacy. In Canada, for example, the Rare Disease Foundation, which was founded by parents, provides opportunities for parents to form social connections, attend informational meetings, develop educational resources, support RD research, and do RD advocacy. Genetic counselors are often involved in such organizations as volunteers and board members. Other researchers have found that attending conferences with other parents can increase knowledge and self-efficacy, particularly after attending several times (Bogart and Hemmesch 2016). In terms of the mode of support, previous research with mothers of children with RDs found that connecting with peers online provided access to informational and emotional support (Glenn 2015). For participants in our study, connecting with peers via social media platforms was identified as a key resource. It is therefore imperative that

organizations where children with RDs attend treatments have mechanisms to inform parents about these peer support resources even when healthcare providers themselves are not involved with facilitating or participating in the groups.

Engaging patients and families is also an important and growing focus in research. When individuals with RDs and/or their family members partner with researchers, there is potential to identify more relevant clinical and policy questions with an emphasis on patient-centered health outcomes (Forsythe et al. 2014). There are recent examples of engagement of patients and their families in RD research (Kesselheim et al. 2015); however, there is an apparent gap in the implementation of meaningful collaborative and bi-directional approaches (Forsythe et al. 2014). Researchers who are genetic counselors should consider including parents and patients in their research programs. Given the depth of knowledge that parents have related to their child's RD specifically, and to the experience of navigating the healthcare system more generally, co-development of research priorities and projects with families and patients who experience RD ought to be mandatory for agencies funding research.

Limitations

There were several limitations in this study. Participant recruitment focused on sources where families of children with RDs were engaged—hospital settings and rare disease advocacy and support groups—and this limited participation by those who are less connected to services and peer groups. As well, all the participants lived in Canada and so their experiences reflect the country's healthcare system and services. Further, the participant group was almost entirely comprised of women, which is similar to research with parents of children with disabilities and special needs (Braunstein et al. 2013). The absence of fathers in this body of literature should be addressed in future research.

Implications for Policy and Practice

Although a single RD may be unique in itself, 1 in 2000 individuals will be diagnosed with a RD in their lifetime, which has implications for healthcare providers, particularly genetic counselors, pediatricians, family physicians, nurses, and allied healthcare professionals. Education is needed to ensure clinicians are aware of the common aspects of RD experienced by individuals and their family members, particularly around informational and social support. Given that genetic counselors have great expertise in working with families of children with RDs, they could provide leadership in developing interdisciplinary curriculum in the health sciences in this area. Policy and programming structures need to address the need to have formal care coordination for individuals with RDs so that this role does not add to patient or family caregiver burden. Moreover, formal care coordination could mitigate potential inequities among individuals with RDs, given that the ability to navigate healthcare systems can vary based on the different socio-structural conditions experienced by individuals. The study also highlighted how narrow diagnostic criteria for services can unintentionally exclude those with unique diagnoses. We suggest that these criteria may need to be reconsidered and be based more on functional needs than diagnostic labels. Lastly, it is clear that peer support plays an important role for parents of children with RDs. Most of the participants in the study discovered peer support through informal networks and social media, and it may be helpful to have formal mechanisms for genetic counselors to better connect families at the time of diagnosis, or during the pre-diagnosis stage. Overall, genetic counselors have a key role in advocating for changes to policy and implementing findings from this research into their practice.

Conclusion

In conclusion, although their diagnostic labels are unique, individuals with RDs and their families have many experiences in common. In particular, participants identified issues related to the diagnostic journey, accessing services, and peer support. In order to foster more equitable and relational care for these families, policy and programming changes are needed. Moreover, the broader research mandate about rare diseases must move beyond diagnostics and treatment to address the very real and pressing issues that arise from the patient—and by extension family—experience in navigating healthcare systems.

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S. Mayer made substantial contributions to the acquisition, analysis, and interpretation of the data and assisted in drafting the initial manuscript and revision and gave final approval of the version to be published and agrees to be accountable for all aspects of the work.

I. Sloan Yip made substantial contributions to the analysis and interpretation of the data and assisted in drafting the initial manuscript and revision and gave final approval of the version to be published and agrees to be accountable for all aspects of the work.

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Compliance with Ethical Standards

Conflict of Interest Jennifer Baumbusch, Samara Mayer, and Isabel Sloan-Yip declare that they have no conflict of interest.

Human Studies and Informed Consent All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (The University of British Columbia Behavioural Research Ethics Board) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all participants for being included in the study.

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

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