Our “Rare” Children Deserve Better: Mothers of Children with Rare Diseases Speak Out

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Abstract

"Difficult, stressful, frustrating – not the syndrome, not the persons with the disabilities, but our ableist society."

Drawing on my conversations with six mothers of children with rare diseases, I use a critical disability framework and an intersectional lens to explore the limitations of community and healthcare services for children with rare diseases and the implications on parental mental health as they attempt to navigate services for their children. In this paper, I shed light on the hardships and battles experienced by parents of children with rare diseases as they attempt to access community and health care resources and services for their children. These hardships are exacerbated when parents attempt to advocate for acknowledgement and acceptance and inclusion in the world. From initial diagnosis to interactions with health care and broader supports, the effects of a "rare" disease greatly impact the lives of families. The purpose of this research is to raise awareness of rare diseases – specifically, Chromosome 18q Deletion and Pitt-Hopkins Syndrome. As a new parent of a child with rare diseases myself, I present the struggles of families like my own and uncover the deeply-rooted ableist structure of the society we live in while looking at the implications for future social work practice. Our "rare" children deserve better.

*Keywords***:** rare children, disability justice, critical disabilities studies, rare diseases, genetic conditions, Pitt Hopkins Syndrome, Chromosome 18q

Résumé

« Difficile, stressante, frustrante… pas la maladie, ni les personnes qui en sont atteintes, mais bien notre société capacitiste. »

En me référant à mes discussions avec six mères d'enfants atteints de maladies rares, j'utilise une approche critique du handicap et prends une perspective intersectionnelle afin d'explorer les limites des services communautaires et de la santé destinés à ces enfants, ainsi que l'impact de ces limites sur la santé mentale des parents concernés. Dans cet article, j'attire l'attention sur les difficultés et les combats que mènent les parents pour accéder aux ressources et aux services communautaires et de la santé pour leurs enfants atteints de maladies rares. Ces difficultés sont amplifiées lorsque les parents tentent de promouvoir leurs reconnaissances, leurs acceptations et leurs inclusions dans la société. Du premier diagnostic aux interactions avec les services de la santé et les autres soutiens disponibles, les effets d'une maladie « rare » bouleversent profondément la vie des familles concernées. L'objectif de cette étude est de sensibiliser le public aux maladies rares, en particulier la délétion du chromosome 18q et le syndrome de Pitt-Hopkins. Étant moi-même parent d'un enfant atteint d'une maladie rare, j'expose les difficultés que rencontrent les familles comme la mienne et révèle la structure profondément capacitiste ancrée dans notre société, tout en examinant les implications pour les futures pratiques du milieu du travail social. Nos enfants « rares » méritent mieux.

Mots clés : enfants rares, équité pour le handicap, études critiques du handicap, maladies rares, maladies génétiques, syndrome de Pitt-Hopkins, Chromosome 18q

Introduction

Within western society, individuals with disabilities are constantly made to feel like outsiders and as if they do not belong. Our social and political structures are deliberately designed in a way that serve and benefit able-bodied people, even though many individuals do not fall under this socially constructed category and have some kind of impairment, disability, disorder, or disease (Erevelles, 1996). When it comes to rare diseases, different countries have different understandings as to what constitutes a rare disease as this number is usually based on the total population of the country (Dellve et al., 2006). According to the Rare Diseases Foundation (n.d.), "within Canada, a rare disease is defined as a condition that affects fewer than one in 2,000 people (…) and although rare, collectively there are over 7,000 diagnosed diseases affecting more than 3 million Canadians" (as cited in Currie & Szabo, 2019a, p. 96).

In 2017, a working group came together and created 19 recommendations to address the systematic barriers and difficulties faced by Ontarians with rare diseases, as outlined in a rare diseases report produced by the Canadian Organization for Rare Disorders (CORD) (Critical Care Services Ontario, 2017). The objective of the Rare Diseases Working Group was to highlight the strategic goals outlined by CORD's general report for Canadians living with diagnosed and undiagnosed rare diseases and implement a strategy that could benefit future healthcare delivery of Ontarians in similar circumstances (Critical Care Services Ontario, 2017). Despite the Rare Diseases Working Group's report, the recommendations were not addressed by the province (Ontario NDP, 2023). Now, with NDP's support and calls to action, the First Reading of Bill 129, the Rare Disease Strategy Act, 2023, was recently carried to amend the Health Protection and Promotion Act. Once Bill 129 receives Royal Assent and comes into motion, Ontario can start implementing the recommendations set out in the Rare Diseases Work Group Report (Legislative Assembly of Ontario, 2023; Ontario NDP, 2023).

The nature of rare diseases is that they typically manifest due to some form of alteration in one's genetic makeup and can result in chronic and debilitating conditions (Ontario NDP, 2023). While there is not always a cure, early intervention is considered beneficial to one's development (Pelentsov et al., 2016). As a mother of a child with a rare disease, I have come to realize that while there are rehabilitation and "treatment" centres that provide various therapies, unless a formal diagnosis has been made, doctors often suggest waiting to assess if a child will learn to do the things they presumably are "not capable of." This delays referrals to healthcare and community services and results in a delay in potential "treatment" as well. Parents like me are left to worry about their children, self-navigate services, and implore doctors to take their children's conditions seriously. Since parents are often the first to notice their children's conditions, it is necessary to examine their trying experiences and struggles during the process of acquiring support in this ableist society (Brown et al., 2019).

This paper examines the way in which barriers and limitations of healthcare and community supports for rare diseases place profound stresses on families, particularly on mothers as they navigate services for their children. I explore this question using a critical

disability framework (Brown et al., 2019; Erevelles, 1996) and an intersectional lens (Crenshaw, 2012) because barriers to accessing supports for children with rare diseases and the implications of these barriers on parental mental health are a social justice issue. I use a narrative analysis to understand the experiences of mothers of children with rare diseases as they attempt to navigate these barriers, highlight their struggles, and advocate for inclusivity, equality, and social justice (Squire et al., 2014).

Context

Upon examining the scholarly literature on 'the limitations of community and healthcare services for children with rare diseases' and 'parental mental health,' it became evident that the available research was mostly clinical and situated across North America, the United Kingdom, and Europe (Currie & Szabo, 2019a; Dellve et al., 2006; Pelentsov et al., 2016; Titgemeyer & Schaaf, 2020; van Dijk et al., 2019). The scholarly research on the experiences of "disabled children" discussed the unmet needs of parents as well as parental well-being as a result of caregiver "strain," negative societal attitudes toward disability, the value of parental input in services, and a request for social work involvement (Middleton, 1998; Sheenar-Golan, 2016; Woodcock & Tregaskis, 2008). Baumbusch et al.'s (2019) qualitative study noted that the parents of children with varying rare diseases and diagnoses had similar experiences navigating the healthcare system. While there is limited literature identifying supportive care needs of parents of children with rare diseases and examining parental experiences (Aldiss et al., 2021; Baumbusch et al., 2019; Currie & Szabo, 2019a; Hytiris et al., 2021; Pelentsov et al., 2016), there is even less scholarly literature on how social workers should work with children with rare diseases and the mental health impacts on parents (Currie & Szabo, 2019a; Sloper, 1999).

Barriers to Accessing Services

For parents of children with rare diseases, the common barriers to accessing services include: a delay in receiving a diagnosis (Baumbusch et al., 2019; Garrino et al., 2015; Güeita-Rodriguez et al., 2020; Hytiris et al., 2021; Kolemen et al., 2021; Pelentsov et al., 2015); parents being forced to play the role of an expert on their child's condition (Baumbusch et al., 2019; Currie & Szabo, 2019a; Currie & Szabo, 2019b; Güeita-Rodriguez et al., 2020; Nygard & Clancy, 2018; Parish & Cloud, 2006; Pelentsov et al., 2015; Pelentsov et al., 2016); financial implications (Baumbusch et al., 2019; Güeita-Rodriguez et al., 2020; Parish & Cloud, 2006; Pelentsov et al., 2015); and being made to feel alone (Aldiss et al., 2021; Baumbusch et al., 2019; Currie &Szabo, 2019; Garrino et al., 2015; Güeita-Rodriguez et al., 2020; Kolemen et al., 2021; Pelentsov et al., 2016; Titgemeyer &Schaaf, 2020). These common barriers demonstrate a need for further exploration and policy recommendations.

Due to the rare nature of rare diseases, parents often face difficulty in getting a diagnosis for their children (Baumbusch et al., 2019; Sloper, 1999). Even when a rare diagnosis is received, healthcare providers are not always well-informed. This often results in parents being left to research information themselves, not knowing whether the information they found is

credible or not, and they end up having to navigate applications for funding and services on their own (Baumbusch et al., 2019). Baumbusch et al. (2019) also mention how despite learning the diagnosis of their children, parents do not always receive supportive services right away and potentially encounter out-of-pocket expenses as well. Currie and Szabo (2019a) add that proper care of their children might be overlooked as parents are left to take on all the responsibilities of their child's care, navigate any gaps, advocate for better services, manage appointments, and submit applications (Currie & Szabo, 2019b; Vanegas & Abdelrahim, 2016). This lack of supports and barriers to accessing services can result in parents feeling alone in their struggles (Currie and Szabo, 2019a; Güeita-Rodriguez et al., 2020; Middleton, 1998).

According to Garrino et al. (2015), scientific researchers are apathetic to rare diseases since they are 'uncommon.' The frustrations of parents over delays in diagnosis and not receiving enough information or support from healthcare professionals are echoed throughout the current scholarly literature (Güeita-Rodriguez et al., 2020; Kolemen et al., 2021; Pelentsov et al., 2016; Vanegas & Abdelrahim, 2016). However, the literature fails to examine how social structures and systems pathologize and individualize the experiences of individuals with rare diseases and their families (Brown et al., 2019).

Impacts on Parental Mental Health and Unmet Parental Needs

Upon examining the literature for experiences of and impacts on parents of children with rare diseases, the concepts of stress, anxiety, and depression are reiterated (Aldiss et al., 2021; Baumbusch et al., 2019; Currie and Szabo, 2019a; Dellve et al., 2006; Hartley et al., 2012; Kolemen et al., 2021; Nygard and Clancy, 2018; Pelentsov et al., 2015; Pelentsov et al., 2016). According to Dellve et al.'s (2006) prospective intervention study on the well-being of parents of children with rare diseases in Sweden, mothers often face more stress than fathers, and single mothers, especially mothers that have more than one child with a disability, face even more stress compared to mothers with partners (see also Hartley et al., 2012; Kolemen et al., 2021). Dellve et al.'s (2006) research indicates that even though fathers also experience parental stress, their stress often stems from feelings of incompetency tied to not being able to do anything about their children's irreversible conditions and having to serve as support systems for their partners (see also Güeita-Rodriguez et al., 2020; Pelentsov et al., 2015).

Concurring with Dellve et al.'s (2006) findings that mothers experience higher stress levels and emotional distress, Pelentsov et al. (2015) suggest that this is perhaps because mothers are often 'naturally' the primary caregivers. In fact, Pelentsov et al.'s (2015) study found that mothers of children with rare diseases are 'naturally' more inclined to leave their careers and 'choose' to tend to their children, thereby resulting in added financial strain on fathers. The literature reduces mothers to patriarchal standards of being a 'nurturer' while claiming that being regularly exposed to the reality of their children's situation(s) can have a detrimental effect on the mental health of both parents (see also Currie and Szabo, 2019a; Sloper, 1999; Vanegas & Abdelrahim, 2016). Despite their high stress levels, it has been noted within the literature that

parents tend to suppress their worries to focus on their children (Aldiss et al., 2021) and find that their own needs are often unmet and unsupported (Russell et al., 2003).

Scholars note that the need for informational, social, and emotional connection with other families with rare diseases is so important for some people that they would even consider relocating across the globe just to seek *some* support (Baumbusch et al., 2019; Pelentsov et al., 2015; Pelentsov et al., 2016). Scholars note that parents need to understand how to navigate different supports in the community, have their voices and stories heard and their hardships and experiences understood by healthcare professionals (Currie & Szabo, 2019a; Güeita-Rodriguez et al., 2020; Middleton, 1998; Morris, 2014; Pelentsov et al., 2015; Pelentsov et al., 2016; Sloper, 1999). As Sloper (1999) and Middleton (1998) suggest, while supporting parents may not have a direct impact on their children, it can lessen the stress on parents and, in turn, help them to better support their children (see also Nygard and Clancy, 2018).

Practice Response in Supporting Parents: Suggestions from the Literature

Various opinions emerge from scholars regarding the type of supports that currently are—and should be—available for families. According to Selber et al. (2006), social workers need to understand the needs of families and spread awareness about genetic conditions to expand support needs beyond family advocacy. According to Hytiris et al. (2021), rare conditions need to be publicized and brought to the attention of healthcare professionals so that the importance of including patients with rare diseases and their parents in clinical research is recognized and the level of support that they receive is enhanced. Esquivel-Sada and Nguyen (2018) mention that rare disease diagnoses can lead to discrimination from public institutions which a social model of disability can uncover (Oliver, 1999). Some believe "caregiver strain" and parental mental health need to be examined (Dellve et al., 2006; Pelentsov et al., 2016). However, the way supportive programs are run and the assumptions they produce can often feed into family and gender stereotypes. According to Dellve et al.'s (2006) study, family intervention programs do not always have the same positive impact for mothers as they do for fathers. Dellve et al. (2006) state that the "societal economic arrangements that provide greater opportunities for men, in combination with the need for one parent to be available to deal with the child's problems, (…) [result] in the unequal distribution of stress and poor health between mothers and fathers" (p. 399). Mothers of children with rare diseases might experience more stress because of heteronormative gender roles and expectations, which often ignore diverse and queer family arrangements. Studies need to explore diverse gender roles and the importance of women's labour both in and outside of the home.

Pelentsov et al. (2016) also emphasize that mothers of children with chronic illnesses can have a harder time coping and adjusting to their children's health. Scholars add that healthcare professionals should understand how parents view their children's diagnoses and conditions so that they can identify the best ways to support them (Middleton, 1998; Pelentsov et al., 2015; Pelentsov et al., 2016; Shenaar-Golan, 2016). Although the literature stresses the importance of supporting parents of children with rare diseases, rarely do they do so from a broader social and

political standpoint. Scholars note that social workers can get caught up in administrative processes and bureaucratic practices (Güeita-Rodriguez et al., 2020; Middleton, 1998; Parish & Cloud, 2006) which can affect supports, thereby resulting in parents paying for services out-of-pocket instead of staying on long wait-listed services. Güeita-Rodriguez et al. (2020) suggest that better programs need to be implemented and barriers need to be eliminated so that families can receive the support they need, especially lower income families. Unfortunately, there seems to be a lot of emphasis on getting parents to do more – to participate in healthcare research, provide their input on services, and determine which services are needed for their children, thereby excusing the larger systemic problems (Currie and Szabo, 2019b). While it is true that parents of children with rare diseases often know a lot, it should not supplant the need for solid research just because their child's condition is "rare." While parents should have the option to be a part of research (Baumbusch et al., 2019), our systems, policy makers, and society as a whole need to do better.

Moving Forward Based on the Literature

The clinical literature on rare diseases acknowledges that there are not enough studies or comprehensive literature that discuss the unmet needs and/or supportive care needs of caregivers and parents of children with rare diseases, so parents turn to social media for connection (Aldiss et al., 2021; Currie & Szabo, 2019a; Currie & Szabo, 2019b; Esquivel-Sada & Nguyen, 2018; Pelentsov et al., 2015; Titgemeyer & Schaaf, 2020). Many smaller studies are not even applicable on a wider global scale (Pelentsov et al., 2015). I argue that the overall impact on parents as they attempt to navigate social and political barriers needs to be examined from an intersectional lens (Crenshaw, 2012) and critical disability framework (Brown et al., 2019; Erevelles et al., 1996; Oliver, 1999). Examining the structural and systemic barriers that support ableism and contribute to oppressing, silencing, and individualizing rare children and families is integral if we want to work towards an equitable and inclusive society (Brown et al., 2019; Erevelles, 1996). The goal of my study is to collect everyday experiences and stories of parents with children who have rare diseases. I will draw insights through a theoretical lens that critiques the medical model, de-naturalizes the enormous gender disparities in caregiving while attending to class, race, and other categories that contribute to inequities.

Theoretical Framework

For this study, I draw on critical disability studies and feminist intersectionality (Brown et al., 2019; Erevelles, 1996; Oliver, 1999). Throughout the history of social work, disability was regarded from a 'medical model perspective' and seen as an 'individual issue' (Hiranandani, 2005; Oliver, 1999). Many scholars in Critical Disability Studies, however, view disability from a 'social model perspective' arguing that "it is not the physical and/or mental impairment that disables an individual, but the 'handicapping' effects of a society whose interests are geared toward an 'able-bodied' population" (as cited in Erevelles, 1996, p. 519). Society is physically, socially, and politically constructed in a way that serves able-bodied people and it is through this deliberate construction that some individuals become "disabled" (Davis, 1995; Erevelles, 1996).

Society and societal practices are intentionally designed to place people with any kind of disability at a disadvantage in all aspects of life, as "the world we live in was never built for disabled people" (Brown et al., 2019, p. 182).

A critical disability framework seeks to highlight that "disabled" people do not simply exist but there is a "disabler" that tactfully frames individuals who do not fit the 'norm' (Davis, 1995). This framework also seeks to publicize obstacles and barriers that people with disabilities face on account of ableism and calls out the attitudinal barriers and social and political structures that contribute to their oppression (Brown et al., 2019). Collectively, many critical disability scholars believe that disability is not a 'distinct' or 'discrete' identity because people with disabilities also face oppression due to their race, culture, class, gender, sexuality, or other forms of social identities (Brown et al., 2019; Erevelles, 1996; Hiranandani, 2005). Thus, a critical disability framework strives to advocate for equality and social justice because all forms of oppressions, including capitalism, colonialism, and white supremacy will continue to exist if there is no 'disability justice' (Brown et al., 2019; Hiranandani, 2005).

I also draw on feminist theory, specifically using an intersectional lens. A feminist framework considers how people with disabilities face multiple social oppressions instead of being reduced to one identity (Hiranandani, 2005). Notable scholar Kimberlé Crenshaw brought forth the idea of intersecting social identities which argues that everyone is made up of complex social identities that are deeply intertwined and ultimately shaped by larger social, political, and economic structures that serve to benefit or marginalize them (Crenshaw, 2012). Crenshaw's (2016) theory suggests that individuals who are often oppressed or disadvantaged because of one social identity such as 'ability,' are often oppressed due to other social identities as well, such as class and gender. An intersectional lens provides insight into the kinds of social and political barriers individuals face and the implication of these barriers on their lived realities.

The lived reality of children with disabilities or genetic disorders often includes having to rely on the support of family since society is constantly rendering their disability as a personal "issue/concern," (Hiranandani, 2005; Wendell, 1989). In fact, "the entire physical and social organization of life assumes that we are either strong and healthy and able to do what the average able-bodied person can do, or that we are completely disabled, unable to participate in life" (Wendell, 1989, p. 111). Since parents – especially mothers – of children with rare diseases are often the ones to navigate barriers to community and healthcare resources, it is important to understand the different kinds of struggles they encounter because of society's 'disabling' effects (Hiranandani, 2005).

Methodology

This study uses a narrative methodology, also known as narrative research or narrative inquiry (Squire et al., 2014). The term "narrative" itself suggests stories (written or oral) and individual accounts of an event or an experience. Narratives are encompassed in various fields of study, mainly social sciences, as an interpretive approach (McAlpine, 2016; Squire et al., 2014).

According to Squire et al. (2014), narrative materials manifest in two ways: "sometimes, they already exist (…) [and] sometimes, that narrative materials come into existence as part of the research" (p. 7). In either case, narrative material and its content provides an opportunity to shed light on the physical and/or social realities of individuals and their families, including the struggles they may have encountered and the potential ways they may have engaged in resistance (Squire et al., 2014). Narrative research provides researchers an opportunity to pay attention to the stories of marginalized groups and examine the ways in which power relations ultimately shape their understanding of the world and their own individual experiences (Squire et al., 2014).

Squire et al. (2014) emphasize, "it is perhaps only through the voice of the narrator that we can gain insight into such personal dimensions of experience" (p. 75). Since narrative methodology provides a platform for stories and voices to be heard, it can help expose problematic macro narratives in society and advocate for social change (Squire et al. 2014). For my study, I engaged in conversational interviews with six participants as this provided a flexible way to explore concepts, clarify any questions and create a safe and more comfortable space to have conversations (Lavrakas, 2008). Participants recruited for my study are parents, specifically mothers of children with rare diseases. The participants of this study were recruited from Facebook support groups, specifically from the "Chromosome 18q Deletion" and "PARENTS of Pitt Hopkins" support groups, which I have been a part of since August 2021 as a parent of a child with rare diseases myself. Following the work of other empirical researchers (Badwall 2016), I included myself as one of the participants as well.

The interviews ranged from 75-105 minutes in length. It was evident that these mothers were never given a chance to share their stories, experiences, thoughts, and/or feelings before. Participants indicated that whenever they did try to speak up or voice their frustration, they were chronically dismissed or shut down – something that resonated with me as a parent of a child with rare diseases. As a researcher, I recognized that I was asking a lot of the participants. However, their involvement was important and legitimized the struggles and barriers that mothers of children with rare diseases face, something that is not currently recognized by the scientific/scholarly communities. Below, I present profiles of each parents' story as well as research findings that highlight the structural and systemic problems finding a diagnosis, including navigating an ableist healthcare system and the unequal distribution of care labour for mothers of children with rare diseases. I also highlight the strengths that mothers exhibit in spite of intersectional oppression and marginalization, and I emphasize their suggestions and recommendations that can be applied to future legislations pertaining to rare diseases.

Introduction to the Participants and their Families

Sarah

Sarah's six-year-old son was diagnosed with ADHD, Pitt-Hopkins-like Syndrome 2, and most likely Mowat-Wilson syndrome as well, at the age of three-and-a-half. According to Sarah, just being able to see a geneticist took two years. They learned that roughly only 13 people in the entire world have been documented with having both of the rare genetic conditions that her son

has. Sarah and her husband have another son who has his own health issues. As a result, Sarah and her husband are unable to leave their children alone for any amount of time and someone always needs to be available to watch them. At school, Sarah's son receives occupational therapy and physiotherapy; however, for behaviour therapy her son has been on the waitlist for three years. While Sarah and her husband live with family, they find that no one seems to understand their struggles. Sarah experiences anxiety often and used to take medication but she has not been able to find the time to visit the doctor to discuss her own health concerns.

Miranda

Miranda has two daughters, a 32-year-old, and a 28-year-old. Miranda's youngest has Chromosome 18q- Syndrome, which is a "rare chromosomal disorder…" (NORD's Rare Disease Database, 2021, General Discussion section, para. 1). The "initial reaction around [her daughter's] diagnosis (…) was really tough;" her daughter was "delayed in every aspect" –she did not walk until she was almost four years old and she did not start talking until the age of five. Miranda was able to get her daughter into early intervention where they focused on "eating, socializing, making music, speech, and sign language." Miranda attended all of her daughter's appointments on her own and attended Chromosome 18 conferences in an attempt to learn more and to participate in studies. Miranda ended up raising her daughter on her own and had to ask her husband to leave since he never truly accepted their daughter's condition and "added a great deal of stress." Her older daughter eventually "walked away from the family, saying *'I hate everything that's happened, and I'm not getting over this.*'" Miranda currently lives alone and works with students with ADD and autism, and she is "teaching the world how to accept them."

Mona

Mona has a five-year-old daughter who has a diagnosis of autism and Pitt-Hopkins syndrome, a "rare, genetic, neurological disorder" (NORD's Rare Disease Database, 2021, General Discussion section, para. 1). Mona's daughter was almost one year old when the family learned about her diagnosis. According to Mona, her entire day revolves around her daughter and not even a second is spent without her. Mona finds that she and her husband are unable to discuss their child's condition so they avoid the topic. Mona was a professor in her country of origin. However, after coming to Canada, her education was not recognized in the same way, and without any family here to support her daughter, Mona has not been able to resume/pursue her career. Mona experiences quite a bit of social isolation and finds that no one understands what she is going through, except for her mother back home. Mona has encountered many challenges within society when it comes to advocating for inclusion and accommodations for herself and her daughter. Mona greatly fears what her daughter's future will look like due to the lack of support.

Freddie

Freddie, her husband, and their four sons live in a part of Ontario that often gets "forgotten" by the province. Freddie's youngest has Pitt-Hopkins syndrome, with a mutation of R578H on the 18th Chromosome. While Freddie's son is 12 years old, he is "developmentally

about 14 months old. Pretty much what you'd expect from a 'normal' 14-month-old (…) Sippy cups, diapers, crib, stroller, car seat, highchair, only massive versions of it!" Freddie's son was diagnosed when he was 31 months old. Freddie faced many obstacles trying to find a diagnosis and supports for her son, including doing her own research, writing requests, and asking health professionals to sign off on them, appealing rejected applications, and navigating any available community and governmental supports on her own. As the mother, Freddie finds her son's care "has landed (…) almost exclusively" on her, and she has not been able to work until recently. Freddie believes that Pitt-Hopkins "looks pretty bad on paper;" however, she has had the opportunity of meeting "43 other Mamas and 29 Pitt Kids," and it is not so bad in "real life." Freddie has found that meeting other families in similar situations has been a "blessing."

Danish

I am a racialized, Muslim woman, and mother of two daughters, a 3-year-old and an 18-month-old. When my youngest was five months old, we learned that she had a condition called Unicoronal Craniosynostosis, which causes a flattening on one side of the forehead (Great Ormond Street Hospital for Children, 2015, Unicoronal Craniosynostosis section, para. 1). At eight months, my daughter had a chromosomal microarray analysis done which indicated that she also has Pitt-Hopkins syndrome. My daughter was two months old when I knew there was something going on, but six months went by before I finally received "answers." Then, my world changed forever. As the mother, I am primarily responsible for my daughter's care: navigating resources in the community, keeping track of her multiple appointments, connecting with doctors and therapists, all while attempting to wrap my head around this unheard-of diagnosis. While my daughter's father is involved and we have *some* family support, no one will ever truly understand what I go through.

Sabrina

Sabrina's middle child is currently four and a half years old and was diagnosed with Pitt-Hopkins syndrome when he was 18 months old. According to Sabrina, her son's diagnosis came as a "blow" to her and her husband, impacting them both, but her most significantly. Prior to coming to Canada, Sabrina was a doctor. Since Canada did not recognize her credentials, she had to work extra hard to prove herself. Sabrina was preparing for her final licensure exams to pursue her career as a doctor. But upon receiving the diagnosis of her son, Sabrina was "shattered." She socially isolated herself for some time, and she and her husband faced "bouts of depression." Sabrina had to pull herself out of depression to be there for her three kids. During the day her son attends school, and the evenings are "primarily based on his therapies." Sabrina and her family currently live alone here in Canada; they do not have any relatives or friends to support them. According to Sabrina, community therapists have served as a "tremendous" support for her son. While Sabrina believes that 'me-time' is "mandatory" for one's mental health, she has had to set her own time aside in order to care for her son. Sabrina's deep faith in God and her mantra, 'I am chosen for this role,' is what keeps her going.

Dismissal of Concerns: Gaslighting within the Healthcare System

Through conversational interviews with the participants of this study, and after organizing the data accumulated, it became apparent that the first and most difficult encounters that parents of children with rare diseases end up facing is having their concerns dismissed and invalidated by healthcare professionals. In Freddie's experience, when she told the doctor that her son "*wouldn't coo, turn to look at me when I called him, hold toys or try to roll over (…) [the doctor] simply said 'He's a boy'… yes, but I've already had 3 healthy boys and this one is very different"* (Freddie, personal communication, February 9, 2022). Not only did this incident result in Freddie's child being overlooked, but it also resulted in both mother and child being reduced to gender stereotypes, as an 'overly concerned mother' and a 'slow maturing boy.' Despite feeling invalidated and alone, Freddie went as far as consulting with various doctors across the world and bringing information back to her local geneticists, only to have them undermine her research and hypothesis with, "*no, it's too rare*" (personal communication, February 9, 2022). Years of fighting and advocating passed by before a geneticist finally agreed to entertain the idea and test Freddie's son for even the 'rarest' of genetic conditions (Freddie, personal communication, February 9, 2022). As Freddie expressed, having a disabled child is "*nothing compared to fighting the people who are supposed to be helping us"* (personal communication, February 9, 2022).

In my case, while I also recognized early on that my daughter was not meeting her milestones, any time I brought my concerns to the pediatrician, I was simply reminded, *"'sometimes it just takes time'"* (Danish, personal communication, February 2, 2022). In fact, the only reason I was able to get a diagnosis is because I badgered our doctor for months before he suggested that my daughter's DNA could be tested at a microscopic level. Had I not pressed the doctor, I would still be waiting. While the literature highlights delays in diagnosis as a barrier to accessing support (see Baumbusch et al., 2019; Sloper, 1999), my research indicates that advocacy and resistance to societal norms and systems is imperative for receiving answers.

The participants in this study did not find healthcare professionals, or their interactions with them, to be comprehensively supportive. Immediately after receiving her one-year-old daughter's diagnosis, the geneticist advised Miranda, "*'You best get her some intervention, or you might want to think about putting her in a home because* she *is going to be complicated'"* (Miranda, personal communication, January 23, 2022). Instead of reassuring and supporting this scared mother, this ableist healthcare practitioner merely stigmatized and reduced Miranda's *baby* to her disability. Additionally, the participants noted that once a diagnosis is identified, families are off-loaded to other practitioners or specialists as if there is no longer a societal duty to support them. In Sarah's experience, while she believes some practitioners may "*mean well*," they hardly know anything substantial. In fact, she even caught them *"Googling"* her son's condition in front of her (personal communication, January 23, 2022). Unfortunately, this lack of knowledge and support from healthcare professionals has been experienced by every participant in the study.

The shared narratives of the participants demonstrate how healthcare practitioners often provide limited support and turn away from pursuing a diagnosis because of the lack of research and interest in rare diseases.

While these diseases are rare and impact less than one in 2,000 people (see Currie & Szabo, 2019a), "rarity" should not excuse healthcare and government systems from learning and doing more. Instead of being accountable for their roles and responsibilities, societal structures and systems marginalize families and leave them to figure things out on their own (Danish, personal communication, February 2, 2022). It is no wonder that Miranda spent so much time attending rare disease conferences in the States to try and absorb information, make connections with other parents, and learn new ways to support her child in an ableist world, all while investing her time and her family's time as well as money, energy, and resources (Miranda, personal communication, January 23, 2022). Miranda soon learned, as the clinical literature suggests, that rare diseases are not a matter of concern for the scientific community (see Garrino et al., 2015); though 'rare' does not mean nonexistent. Sabrina also shared that, *"Once they tell you the diagnosis, (…) they don't follow up (…) they don't jump in with you in that support system"* (personal communication, January 29, 2022). The participants' narratives of struggling with the healthcare system provide a small glimpse into one of many barriers families encounter when trying to support their 'rare' children.

A critical disability framework helps me question the dismissal of parental concerns and reveals a pattern of doctors delaying the discovery and diagnosis of a child's genetic condition. While it is understandable that professionals may not want to be too quick to diagnose or conduct too many tests on an infant, these dismissals of parents, and especially mothers', concerns about their children are nothing short of medical gaslighting (see Sebring, 2021). It is no coincidence that Sarah, Freddie, Miranda, Sabrina, Mona, and I (all women and some of us women of colour) experienced medical gaslighting at some point in our children's diagnosis journey. Our concerns were downplayed and we were written off as impatient and overly sensitive mothers. It was easier to dismiss us than take us seriously because the latter would increase and further complicate their work. This ableist ideology aligns with the historically dominant 'medical model' where disability is perceived to be within the individual and is limiting and problematic (Hiranandani, 2005). As exemplified through Freddie's and my own personal narrative, doctors tend to avoid investigating potential conditions until they have run out of excuses to not put in 'extra work'. Not only does medical gaslighting potentially delay a diagnosis, but it also delays the opportunity to find and connect with supportive communities and sometimes timely supports.

The gendered dismissal of concerns of mothers is present in the literature. As McKay and Hensey (1990) suggest, sometimes doctors do not take mothers' concerns seriously. If they simply considered that perhaps a mother's concern might hold some weight, families could get answers sooner. Yet, more often than not, "mothers' voices often go unheard" (Swallow & Jacoby, 2001, p. 762), as "factors such as classism, racism, and sexism play a role in the altering, delaying, and absence of a diagnosis" (Pena, Stapleton, & Schaffer, 2016, p. 90). Mothers and

women in general tend to be reduced to the gendered labour of caring for children and the home (Traustadottir, 1991), so when they dare to stand up for their children, they are quickly shut down, gaslighted, and invalidated. While the healthcare system and social policies should be working to normalize disability and offer support to families, they leave them feeling more stressed, scared, frustrated, isolated and alone in their struggles. The current systems only serve to disadvantage disabled children, especially children with rare genetic conditions or diseases.

The ableist nature of healthcare coincides with the ableist nature of the systems that run our society, a nature so deeply engrained that it indoctrinates us to only value and long for an "able body" – for ourselves and for our children (Erevelles et al., 1996). The needs of individuals that are "not recognised as typical are seen as limitations and tragedies rather than as possibilities" (Brown et al., p. 191). Society has inbuilt this ideology that disabled bodies are 'different' and 'undesirable' (Brown et al., 2019). Parents and caregivers of children with rare diseases end up looking for acceptance, support, and guidance within the medical establishment, not recognizing that every aspect of this system is intentionally designed against their children. Instead of recognizing that disability is a natural part of human history and an integral part of certain individuals (see Clare, 2017), society further 'disables' our children with rare diseases and renders them insignificant. Thus, critical social workers and policymakers need to challenge society's preconceived notions of disability and highlight the struggles that parents of children with rare diseases experience in order to advocate for social change.

Impact on Mothers

The lack of support provided to families with children with rare diseases results in a great deal of stress. And since navigating supports lands on the primary caregiver, this study reveals it is most often the mother who suffers the greatest distress attempting to navigate barriers and limitations within the social, political and healthcare support systems. Indeed, the mothers in this study stepped into and normalized their own role as a given responsibility. While more studies are needed on diverse families where adult caregivers and parents identify as queer and disabled, the cisgender heteronormative families in this study assumed stereotypical roles.

Sabrina explained that as a mom she had no choice but to take herself out of her own grief and depression. She explained that she had to keep telling herself, "*I am the Chosen One*" because she knew she had to "*stand up again tomorrow for my family and be in a happy mood to be able to raise my children, to nurture my family in the best possible way* (personal communication, January 29, 2022). 'Grief' was experienced by most of the mothers in this study and includes grieving over a life for their child that 'could have been,' grieving over being 'rare,' and grieving over missing out on various aspects of life. While all the participants, including myself, love their children immensely and wish for their inclusion and acceptance, we too are sometimes entrapped by ableist views and discourses that reduce our children's lived realities to what it means to be disabled in this society (Lalvani & Polvere, 2013).

While as mothers we might be struggling with our own health and mental health issues at times, we are still expected to show up, to console and take care of our families, regardless of whether or not we are okay. We are still expected to assume our 'typical' maternal responsibilities, navigate societal barriers, and help our children 'fit in' however we can. While some of us may be privileged enough to have family around or receive 'help,' that help is often conditional or superficial (Freddie, personal communication, February 9, 2022) and the fact is that no one is able to assume our role for any given amount of time (Mona, personal communication, January 29, 2022). Miranda put it best when she explained, *"people do not offer support (...) and if they do, it's a breadcrumb"* (personal communication, January 23, 2022).

Like Freddie, Miranda, Mona, Sabrina, and Sarah, I too am the one who is most impacted by my child's condition. I am the one who connects with all the doctors, keeps track of and takes my daughter to her appointments, seeks out resources in the community, fills out funding applications, and tries to connect with other parents with similar lives (Danish, personal communication, February 2, 2022), all while being in graduate school full-time, raising my 3-year-old and 18-month-old and dealing with my own clinical anxiety and depression. While my daughter's father is also impacted by her condition and takes care of her when he can, I am almost always solely responsible for her care.

In Mona's case, her partner refuses to even talk about the struggles they face in their lives, resulting in her having to navigate resources and supports for her daughter on her own. Mona is forced to do all of this while facing the challenges and barriers as a newcomer and an immigrant to Canada and not having any family or support system – thereby adding to her stress (personal communication, January 29, 2022).

Despite working in and outside of the home, we women/mothers are shaped by society's heteronormative gendered-norms, patriarchal ideologies, and discourses that place "unrealistic expectations" on us to be the caretakers of our children, and especially our "disabled" children (Currie & Szabo, 2019, p. 1256; Dellve et al., 2006; Khanlou et al., 2017). As mothers, we *want* to be able to protect our children from the ableist world; however, we also recognize that if we do not adhere to society's expectations of providing "proper" care and "securing services" for our children, we will not be deemed a "good" mother, and will be scrutinized "for not doing enough (…) and potentially (…) exacerbating" (see Currie & Szabo, 2019, p. 1257) our children's condition(s). Thus, not only does society expect us to be the experts on our children, we are also expected to adhere to socially constructed ideals on what it means to be a "good mother" (Currie & Szabo, 2019; Traustadottir, 1991).

As mothers of children with rare diseases we try our best to be strong, however, we also have moments where we become *"weak,"* experience *"mental frustration"* and even *"guilt"* over all the things we might not be able to do, like "*not being able to give proper time and attention"* to our other children (Mona, personal communication, January 29, 2022). Additionally, due to primary caregiver responsibilities, Sabrina, Freddie, Mona and I shared the experience of not being able to 'work' for years or had to find outside work that was casual and

flexible. In fact, Sabrina ended up leaving her dream of becoming a doctor, and Mona, a professor in her country of origin, gave up her dream of establishing a career here in Canada – all for the sake of their children. Even though we are *"on duty all the time"* (Freddie, personal communication, February 9, 2022), society discredits, belittles, and devalues "much of the care work that mothers do" (Khanlou et al., 2017, p. 629). Due to this, mothers end up internalizing that 'care work' is not work, when in fact our work both in and outside of the home is crucially important. Thus, a gendered and racialized labour market disadvantages women and especially women of colour, where only labour outside of the home is valued in a capitalist economy.

Even though the mothers in this study spend a significant amount of their time and energy supporting, taking care of and advocating for their children, in Miranda's experience, she and her cousin who has a son with Asperger's syndrome, were contemptuously regarded by society as, "*(...) '*that *mother. Oh,* that *mother. That one that won't shut up. Oh, that one who thinks she knows everything.' (...) And it is so deplorable"* (Miranda, personal communication, January 23, 2022). Miranda's experience showcases how women who advocate for their disadvantaged kids are perceived as 'too emotional and inordinate.' Similarly, Sabina notes how whenever she has called people out for complaining to her about their 'neurotypical' children's 'neurotypical' behaviours, they voice and assume, *"'she's in depression, she's going through a hard phase. Oh my gosh, she needs support!'"* (Sabrina, personal communication, January 29, 2022). Instead of standing up for and with mothers of children with rare diseases, society subjects them to "…this social gaze (…) [and] therefore, [mothers of children with disabilities are] judged by not only others around them including professionals and other mothers, but they also internalize this gaze and judge themselves as a standard of good mothering" (Khanlou et al., 2017, p. 614). Society normalizes 'relying on parents' – especially mothers – to support their disabled and 'rare' children on their own; however, this only serves to "excuse the larger social and political contexts that need to be addressed beyond parents' abilities (…) and serve[s] to create and sustain unrealistic expectation for parents" (Currie & Szabo, 2019b, p. 1256). Despite being the backbone of the home even when they themselves might not be okay, mothers are reduced to "gendered ideologies [that are] functioned to hide women's work by deeming it women's nature" (Kandaswamy, 2018, p. 19). As this study shows, most of these mothers are forced to succumb to society's 'gendered division of labour' (see Traustadottir, 1991) and forced to deal with the gaps that structural social inequities create for their children (Currie & Szabo, 2019a). As such, mothers are expected to assume the roles of caregiver, nurturer, at-home nurse, cook, cleaner, and manager, in addition to being an expert on their child's condition.

Feminist theorists' note that throughout history there has been an unequal distribution of care labour, as caring has most often been viewed as "women's responsibility" (Traustadottir, 1991, p. 216). This division is part of the social reproduction of labour that a capitalist economy and workforce rely on (Kandaswamy, 2018). The unequal division of labor is stretched for mothers of children with rare diseases as they are expected to take on multiple 'professional' roles, doing a whole lot more (Currie & Szabo, 2019; Traustadottir, 1991). While many women work both in and outside of the home in Western society, "as soon as there is an increased

demand for traditional women's work within the home – such as caring for a child with a disability – the boundaries shift" and they are expected to drop everything and 'resume' their roles as caregivers at home (Traustadottir, 1991, p. 225). Not only do these unrealistic expectations result in mothers being overworked and overtired, but it also results in their struggles being exacerbated due to a significant lack of societal support (Lalvani & Polvere, 2013). Therefore, no one can truly understand the magnitude of what we mothers of children with rare diseases deal with on a regular basis on account of the barriers and limitations created by social and political systems. No one understands what it is like to be alone in that struggle.

Critically examining the hardships and struggles of the participants in this study and hearing their accounts of not feeling heard, understood, or truly supported by others in society, it is as if the experience of parenting a child with a rare disease is structurally and intentionally designed to be isolating and stressful. The personal narratives of the participants shed light on how social, political, and attitudinal barriers towards people with disabilities results in children with rare diseases and their families being further marginalized, outcast, isolated, and made to regularly face social exclusion and inequities. As the 'Social Model of Disability' claims, people are not 'naturally disabled;' instead, they are intentionally rendered 'disabled' and 'handicapped' due to society's socially constructed outlook on disability (Erevelles, 1996). Rare diseases exist and rare diseases pose health complications, but it is not the disease itself or the sizeable deletion or mutation in a chromosome or an 'impairment' or 'functional limitation' that serves as the significant barrier for people with rare diseases. The ableist society in which we live is what makes it exceptionally difficult (Brown et al., 2019; Erevelles, 1996) for families with children with rare diseases to simply exist.

Future Recommendations

The insightful conversations with the participants of this study revealed that one of their strongest desires is for the world to include and accept their children for who they are. While this paper generally sheds light on the multiple barriers and limitations mothers and families face when they attempt to navigate services and resources for their children with rare genetic conditions, the lack of inclusion 'on society's part' (Sabrina, personal communication, January 29, 2022) continues to weigh on our children. As such, there are several recommendations that the participants have suggested they would like to see happen for their children. Perhaps their recommendations can be considered when Bill 129, the Rare Disease Strategy Act, 2023 passes legislation (Legislative Assembly of Ontario, 2023). The recommendations include: designing a portal or website where parents of children with rare diseases and/or developmental and intellectual disabilities can access all information on the different healthcare, community, and government-funded programs, services, and resources that are available to support children and families, as well as 'next steps' outlining how to access them (Sabrina, personal communication, January 29, 2022); providing a basic income program for families of children with rare diseases so they have additional funds to access supportive care needs that might not otherwise be available; not placing restrictions on what government-funded support payments can be used for;

not making families spend hours filling out lengthy applications to prove their child's disability (Miranda, personal communication, January 23, 2022); not limiting early support programs to only those children with 'specific' diagnoses (Sarah, personal communication, January 23, 2022); funding families in the same province equally without them having to appeal for equal treatment (Freddie, personal communication, February 9, 2022); and accepting and promoting the inclusion of children with rare diseases, without negative attitudes (Danish, personal communication, February 2, 2022).

As I critically reflect on my study and the struggles that the mothers in my study have endured and voiced, I have a few recommendations for social and health policy changes. These recommendations include: implementing community-led support groups for parents of children with rare diseases; having health professionals involved in children's care ask parents how they are really doing and truly taking the time to listen to them and validate them, and direct them to resources rather than leaving them to fend for themselves; offering government funds for caregivers and parents to use towards their own mental health should they need it; recognizing the importance of an investment in parental mental health so parents can be well and continue to show up for their children; supporting charities for rare diseases and hosting proclamation days for rare diseases – not from a pitiful mindset, but from a dutiful mindset – to raise awareness and increase representation, to normalize disability and celebrate diversity; asking parents what reasonable accommodations, accessibility, and inclusivity would look like for them/ their children; creating access to more identity-affirming spaces; creating opportunities for families in similar situations to connect with each other and share and learn from one another; taking concerns of unequal and inequitable treatment seriously; encouraging reflection on ableism, privilege, and oppression in healthcare, education, community, and workplaces; being open to learning from children with rare diseases and their families; recognizing caregiver work as real work; consider compensating caregivers for their work; asking children with rare diseases and their families what they need instead of making assumptions; ensuring all public spaces are accessible and in working order, and being open to revisiting and modifying the concept of accessibility and what it might entail as we learn more. There are many things that can be done to support children with rare diseases and families of children with rare diseases – all we need to do is begin. It is said that parenting takes a "village"—following these recommendations can be a pathway to creating those villages.

Conclusion

As this research paper demonstrates, there are numerous barriers and limitations faced by mothers and families of children with rare diseases, including having to navigate ableist social, political, and economic barriers on their own, being forced to endure the struggle alone, and assuming the roles of parent, caretaker, expert, and manager all in one. Collectively, these structural and systemic barriers and limitations result in mothers and families struggling to fully support their children. These barriers and challenges end up taking a significant toll on the mental health of mothers and families, resulting in their faith in social justice being diminished.

Parents end up internalizing and accepting that stress, frustration, and inadequate support are all things they just have to live with.

It is not our children's conditions that burden our lives, but the severe lack of care, concern, and support on account of ableist healthcare, social, political, and economic systems. It is the sheer disrespect we face from society when we stand up for and advocate for our children; the phoney 'inclusion and acceptance' spiels; the outright dismissals of mothers' concerns from healthcare practitioners; the absolute lack of social and political interest in researching rare diseases, and using our children's "rareness" as an excuse for not taking accountability or learning how to implement what they need to live a peaceful life; and the sheer unwillingness of society to alter the way it views and talks about disability. The ableist societal structure is what serves as the true 'burden of care' (see Currie & Szabo, 2019b) as it hinders and sabotages our ability as parents – as mothers – to effectively care for our children.

While I intended to focus on how navigating limitations and barriers to community and healthcare services impacts the mental health of parents, the narratives of the participants of this study revealed that there are many more hardships that children with rare diseases and their families have to face. Navigating and fighting ableist structural and systemic barriers also has the potential to significantly impact our mental health as parents. We are routinely scrutinized, judged, and tracked (see Hiranandani, 2005). And as if our families don't already have enough stress battling ableist systems, mothers are forced to become teachers, educators, healthcare providers, resource specialists, and therapists for their children. While they may not have the "academic credentials" of all these professions, they are forced to take on these roles without any kind of recognition or compensation because social and political systems have failed, and continue to fail, to take accountability.

As members of society, we need to understand that while ableist social, political, and economic systems might sustain the discourse and ideology of the 'individualization of disability' and make it the parents' "issue to deal with," the reality is that the barriers, limitations, restrictions, hardships, and struggles that come with these structures and systems prevent us from fulfilling our responsibilities. They prevent us from being 'okay.' Consequently, if we as mothers and families of children with rare diseases are not okay, then how can we manage to take care of and nurture our children? We are trapped and oppressed. We are not supported. Our children are not supported. When we 'dare' to advocate for our children, we are quickly told to get back in our lane. The disability of our children is not what holds them back, it is the ableist obstacles that 'disables' and casts them to the margins of society (Clare, 2017; Dellve et al., 2006; Hiranandani, 2005).

Disability, and what it means to be disabled in this ableist society, is a social justice issue and a political issue. Rare diseases are just that – "rare" and sometimes unexplained – but that does not mean they do not exist. It also does not mean that these "rare" genetic occurrences will never happen again. For the sake of our children and all children to come, we need to publicize the barriers, limitations, and restrictions of ableist structures and systems, call them out for what

they are, and advocate for justice, recognition, inclusion, and acceptance of our children. As parents, we need peace of mind so our children can grow up in a world where they are recognized as valuable and celebrated in their diversity—"rare" not because of their rare diseases but because of their beautiful uniqueness. Our "rare" children deserve better.

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