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# Aging out of Children's Hospitals - Exploring the Transition Out of Pediatric Care for Youth with Rare Medical Conditions and their Social Support Networks

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# Abstract

This is an anthropological investigation of social support networks for Persons Living with a Rare Disease, their experiences within children's hospitals and health systems, as well as their transitions from pediatric to adult health systems in Canada. Drawing on the virtual semi-structured interviews with PLWRD and their social support network, this thesis investigates: 1) who participates in the social support network, 2) the roles social support networks play in the transition to adult care, 3) what new social support networks form during and after the transition, 4) the supports and challenges. This research will be a unique contribution to disability scholarship and anthropology by highlighting the voices of my participants of their shared lived experiences of transitioning out of pediatric care, the unique challenges they face, the importance of having a strong social support network, and raising awareness for the overlooked population or rare diseases.



**Keywords:** Rare Diseases; Disability; Children; Adults; Social Support; Networks; Transition; Pediatric Care; Adult Care; Children Health Care; Adult Health Care

# Summary for Lay Audience

One in 12 Canadians has a rare disease, which is defined as a life-threatening, chronic, or weakening condition that affects a small number of individuals. Yet, as rare diseases affect only a small proportion of patients, doctors often struggle to identify possible diagnoses and treatments. Social support networks are essential to coping with experiencing rare diseases. Social support networks include family, friends, and relationships with individuals and groups found through institutions such as hospitals, research institutes, and community groups (in-person and online). My research is an anthropological investigation of social support networks for Persons Living with a Rare Disease (PLWRD), their experiences within children's hospitals and health systems, as well as their transitions from pediatric to adult health systems in Canada. My research investigates: 1) who participates in the support and social network, and what do they provide, seek, find and find lacking in terms of support through their participation 2) what role do social support networks play in the transition from pediatric to adult care 3) what new social support networks and support needs take form during and after this transition 4) what are the supports and challenges PLWRD and their social support networks face in pediatric care that they continue to face in adult care? What new challenges and support emerge in adulthood? I conducted virtual semi-structured interviews via Zoom with PLWRD, who have previously experienced support from children's hospitals, family members, and various other services in Canada. I interviewed individuals within the patient's network as well. My research will be a unique contribution to disability scholarship by enhancing the sharing of lived experience narratives about transitioning out of pediatric care. It will create and share new insights from PLWRD experiences and support the development of new strategies and approaches to increase awareness about rare diseases, provide a deeper understanding of the lives of PLWRD as they transition into adulthood, and advocate for needed support and services.

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#RareDisease

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# Chapter 1

## 1 Introduction

My research interest is to explore Persons Living with a Rare Disease (henceforth PLWRD), their social support networks, their experiences in the healthcare systems, and the transition period between pediatric and adult healthcare. My interest in this field grew due to my supervisor, Dr. Pamela Blocks' course on *Cultures of Health, Illness, Disability and Healing*. My main interest in this course was *rare diseases*, specifically beginning in childhood. I recognized that children have different perspectives on their experiences that get overlooked. I learned that the PLWRD is the only person who knows what it is like to experience and manage a rare condition and how important it is for the PLWRD to be involved with their health decisions. The main reason why I am interested in this specific topic is that my little sister is one of several children who was born with an extremely rare disease. I have observed the interactions between her and the medical professionals and how she stayed strong throughout her journey. However, from Dr. Block's course, I learned that even though I have been with my little sister through her medical journey, she is the only person who knows what it is like to manage and suffer through a rare disease and how important it is for the child to be involved with their health decisions.

Rare diseases are life-threatening, chronic, or debilitating conditions that affect a small number of individuals, and for some, symptoms begin to show early in life, such as at birth or in early childhood[1] (Casey, 2019; Larotonda, 2016; Rare Diseases Working Group Report, 2017; Zurynski et al., 2008). About 7000 identified rare diseases; about 80% of the identified rare diseases are genetic, and 94% of rare diseases have no treatments available (Casey, 2019; Picci et al., 2015; Rare Diseases Working Group Report, 2017). Three million Canadians have a rare disease, but as each rare disease affects a small proportion of patients, health professionals frequently struggle to identify

[1] Did you know that "one-third of children with rare diseases die before their fifth birthday" (Casey, 2019, p. 11).

possible diagnoses and treatments when patients get referred to them (Rare Diseases Working Group Report, 2017). This can affect the patient's health as the diagnosis may be incorrect, or delays in care can lead to complications (Esquivel-Sada and Nguyen, 2018). My research proposes to explore the lived- experiences of the PLWRD and their social support networks, which I have interviewed.

The process of the time and effort it takes to find a diagnosis and treatment is called the "diagnostic odyssey" (Rare Diseases Working Group Report, 2017). The diagnostic odyssey "is lengthy, and the delay in obtaining appropriate clinical care, including treatment, may have serious consequences on the patient's health. Some patients with diseases that have not yet been described, and without a diagnosis, can endure the debilitating effects of the disease process" (Rare Diseases Working Group Report, 2017, p. 5). Also, there is limited access to and availability of support in communities and clinical trials and some patients struggle to find therapists and medications, which tend to be expensive (Rare Diseases Working Group Report, 2017). This problem is in several countries, so in Canada, plans were made to improve care access for Person Living with a Rare Disease (PLWRD) by addressing their needs, such as giving access to care providers and creating and revising existing policies (Rare Diseases Working Group Report, 2017). For example, Ontario began making improvements in its clinical programs and services. However, challenges still occur, such as inequities between available pediatric and adult rare disease programs and service access among urban and non-urban areas (Rare Diseases Working Group Report, 2017). My project proposes to look at systematic supports through conceptual lenses of advocacy.

Furthermore, Esquivel-Sada and Nguyen's (2018) study took place across Canada in three provinces to fill the knowledge gap through "an analysis of the impact of the diagnosis on the lives of RD patients according to their personal experiences" (p.37). Their study revealed that the impacts of rare disease diagnosis for pediatric and adult patients are "multifold" spanning from social to personal and to medical impacts that are either positive or negative (Esquivel-Sada and Nguyen, 2018). Even though there is increasing public awareness of medical and social issues around rare diseases, inequality in health care and supplementary support services continue to be the reality for PLWRD (Esquivel-

Sada and Nguyen, 2018). Due to the development of new treatments and technologies, many PLWRD who used to die in childhood make it to adulthood. This means they must transition out of pediatric healthcare systems and into the adult healthcare systems (Corkins et al., 2018; Sandquist et al., 2022; Mazzucato et al., 2018).

The transition period comes with challenges as patients encounter health professionals who lack knowledge about childhood experiences with rare diseases. The family of the patient faces challenges. For example, families face financial problems, such as the cost of medication, services, and equipment, and they are not well supported (Zurynski et al., 2008). However, there is a lack of studies around the ***transition of PLWRD*** out of pediatric care. Mazzucato et al. (2018) are one the few scholars who studied the transition of PLWRD into adulthood, and state, "despite the fact that a considerable number of patients diagnosed with childhood-onset rare diseases (RD) survive into adulthood, limited information is available on the epidemiology of this phenomenon, which has a considerable impact both on patients' care and on the health services" (p.1). There is a growth of PLWRD surviving into adulthood, but it is after the transition when complications happen and put the patients at risk due to the lack of appropriate support. A term created for the process called "transitional care", which means "the purposeful, planned movement of adolescents and young adults with chronic physical and medical conditions from child-centered to adult-oriented healthcare systems" (Mazzucato et al., 2018, p. 2). My research will explore how the PLWRD and their social support network navigate the transition period using theoretical lenses of care theory to analyze the importance of care for the PLWRD. Also, the conceptual lenses of systemic support analyze the experiences of solitude versus having support through the transition period.

The transition process for PLWRD is challenging for the patient and their family because they spent 18 years with the same people, created relationships, felt comfortable and safe, and had a routine in place by knowing where to go for an appointment, treatments, etc. Once the transition happens, everything changes, and it becomes challenging for the patient and the family to readjust, especially if not given the same level of welcome, care, and support as they were receiving in pediatric care. Some consequences appear with an unsuccessful transition that can result in a risk increase of emergency admission, severe

health complications because of scarce follow-ups, etc. (Mazzucato et al., 2018; Sandquist et al., 2022). Furthermore, there have been studies on the topic of the transition out of pediatric care, but the children they are referring to have chronic conditions and special healthcare needs, which are not similar to rare diseases. Mazzucato et al. (2018) mentioned in their study that their study is the first "proving epidemiologic data quantifying the magnitude of this phenomenon in RD patients, using data from a population-based registry, ongoing since 2002 and monitoring a consistent number of RD entities" (p. 7).

With this thesis, I will add to the current knowledge around the transition PLWRD goes through to increase awareness, data, and the overall importance of the transition period. My research builds on the existing research about the transition of PLWRD from pediatric healthcare systems into adult health systems in Canada, as other scholars have focused on the United States, Brazil, and Europe. Collecting lived experience narratives about transitioning out of pediatric health systems will contribute to disability and medical anthropology and the scholarship on care, advocacy, and self-advocacy. My research will create and share new insights about PLWRD experiences and support the development of new strategies and approaches to increase awareness about rare diseases, provide a deeper understanding of the lives of PLWRD as they transition into adulthood, and advocate for needed support and services.

## 1.1 Overview of the Research and Thesis Structure

My master's research is an anthropological investigation of social support networks for Persons Living with a Rare Disease (PLWRD), their experiences within children's hospitals and health systems, as well as their transitions from pediatric to adult health systems in Canada. My research investigated: 1) who participates in the support and social network, and what do they provide, seek, find and find lacking in terms of support through their participation 2) what role do social support networks play in the transition from pediatric to adult care 3) what new social support networks and support needs take form during and after this transition 4) what are the supports and challenges PLWRD and their social support networks face in pediatric care that they continue to face in adult care? What new challenges and support emerge in adulthood? Theoretical and conceptual

lenses that will be used theories of care (Hartblay, 2020; Ginsburg and Rapp, 2013, Ginsburg and Rapp, 2024; Manderson et al., 2016), theories from disability, medical anthropology, and disability anthropology (Black, 2018; Block et al., 2015; Kittay 2002, 2011, 2019, 2020; Mattingly 2013, 2014; McKearney and Amrith, 2021). Also, theories of support and advocacy (Currie and Szabo, 2019; De Morais et al., 2018; Green et al., 2011; Kittay, 2020; Manderson et al., 2016; Mazzucato et al., 2018; Picci et al., 2015; Sandquist et al., 2022; Silva et al., 2017; Titgemeyer and Schaaf, 2020).

Drawing on eleven semi-structured ethnographic interviews conducted over Zoom with PLWRD and their support network members, I found 19 recurring themes and categorized them into five groups (Table 1). I document the lived experiences of individuals living with a rare disease and their family members who have received support from children's hospitals. Also, how the support and relationships change after the transition into adult health services. The thesis outlines the different transition stages, the support dynamics, the challenges and the supports, and how powerful advocating is, whether it is parental advocating or self-advocating.

There are four main chapters. Each chapter focuses on a theme and will include analytical reflections from personal experiences shared by my participants during their time in pediatric care and transition to adult care.

The first main chapter (chapter two) explores the transition stages that PLWRD go through during their time in the healthcare system. The first stage is the "Before" stage, so their time in the pediatric care. The second stage is the "In-between" stage. The third and final stage is the "After" stage, where the PLWRD enters the adult healthcare system.

In chapter three, I discuss the support dynamics. I go into the different support dynamics and roles of the parents (mothers and fathers), siblings, and friends. Also, I discuss the change of power and control dynamics after the PLWRD transition to the adult care health system.

In chapter four, I dive into the challenges and supports. I discuss the challenges and supports the PLWRD and their support network members have gone through, are still going through in adult care, and are currently going through.

In the final chapter, I explore the power of advocating. I look into the different types of advocacy work my participants have done, share some stories showing the perseverance of PLWRD, and contextualize the advice shared by PLWRD and their network members.

I found it important to get the voices of the PLWRD and their support network members. I learned that the PLWRD is the only person who knows what it is like to experience and manage a rare condition and how important it is for the PLWRD to be involved with their health decisions. It truly takes a village to get through the medical journey because no one knows everything, especially about rare medical conditions. Also, once the transition happens, everything changes, and it becomes challenging for the patient and the family to readjust, especially if not given the same level of welcome, care, and support as they were receiving in pediatric care. Some consequences appear with an unsuccessful transition (Mazzucato et al., 2018; Sandquist et al., 2022).

**Table 1- Themes**

| **Themes** | |
|---|---|
| **The Transition Stages** | - Before: Pediatric Care<br>- In Between: The Unknown<br>- After: Adult Care |
| **The Support Dynamics** | - Roles of Social and Social support networks<br>- Family Dynamics<br>- Systemic Support<br>- Control/Power |
| **The Challenges** | - Throughout the Medical Journey<br>- Impact of COVID-19 |

| | |
|---|---|
| | - Online & In-person Communities<br>- Encounters with Health Professionals and Service Providers<br>- Expertise |
| **The Supports** | - Throughout the Medical Journey<br>- Building Meaningful Lives<br>- Online & In-person Communities<br>- Encounters with Health Professionals and Service Providers<br>- Extraordinary Measures parents take |
| **The Power of Advocating** | - Advocating<br>- Perseverance<br>- Missing Pieces in Healthcare System<br>- Advice |
| **Storytelling**<br><br>**(Memorable Experiences)** | - Powerful positive and negative examples |
| **Emotions** | - Worry, Frustration, Angry, Loneliness, Stress, Love, Loss, Relief, Happiness, Fear, Worth, Faithful, Anxious, Compassion, Uncompassionate, Valued, Overwhelmed. |

## 1.2 Positionality

My positionality in this research is my interest in *rare diseases*, but most importantly, I have the lived experience of having a sibling with a rare condition. At the age of 1, my little sister, Diana, had a rare kidney cancer, but a very rare case of this already rare condition because she had stage 5 bilateral Wilms Tumor, which spread all over her stomach. So, she had a tumour wrapped around each kidney, and then they began to

spread. She went through only chemotherapy since she was too young to do radiation, but it did not help control the tumours and their spreading. So, they had to perform surgery to take out one whole kidney and half of the second one that they were able to save. She lived with half a kidney for nine years until she went into an end-stage renal disease because the half kidney she had left could not support her body anymore and failed. They took out the half kidney, and she went on dialysis for a year and two to three months until she received a kidney. April 10th, 2023 will be ten years since her transplant. A lot of people from her support network, such as our parents, family friends, and even one of her elementary school teachers, tried to give her a kidney. My sister ended up receiving a kidney from an anonymous donor.

However, even though she had a successful kidney transplant, complications occurred afterward due to a nurse giving her more fluids than she could handle without noticing. The left side of my sister's heart and lungs collapsed, and she almost lost her life that night. She was put on life support until the team decided to put her back on dialysis to drain out the extra fluid in her body. She spent a week in the ICU until she got better. No one will ever understand or know how it feels to grow up in a hospital or how the interactions with medical professionals are, except for the one who lived through it.

In March 2021, my sister turned 18, and she had to transition out of the Victoria Hospital Children's Hospital, where she grew up, knows everyone, and everyone knows her (from the doctors, nurses, staff, service workers, etc.). It was a hard transition, especially since it was during COVID-19.

*"To be honest, the transitioning was hard for me, so I wish they had like a section, you are going out of PMDU from a child scenery to an adult life. I have not seen anyone my age there yet, and you are with a bunch of older people, and it's hard to even be comfortable in that setting. I wish they kind of had a place in between, from the PMDU to a section that's where between 18- to 25-year-olds. Where you are getting ready for that transition more". – Diana Kassem*

Diana was one of the lucky ones. Her primary doctor told her what to expect when transitioning from pediatric to adult care. Her doctor talked to her about how soon the

transition is, that our parents will not be joining her at appointments anymore, the environment is going to be different, she will not know the people, have many frequent follow-up appointments, and not be able to create the same connections she had made in pediatric care.

*"So, I wish there was a kind of middle ground where you get to connect with other people your age, you talk to them, and it's a smaller section where you connect with those nurses and doctors, and they will actually help you furthermore". – Diana Kassem*

Overall, Diana and my family have dealt with the very issues highlighted in my and others' past research and continue to do so as she transitioned into the adult healthcare system three years ago. And my older sister, my parents, and I have been and still are a part of her support network. The fact that I am embedded in these networks I have been studying assists and gives me an advantage when connecting with participants and communities.

## 1.3 Literature Review

As mentioned briefly above, I explored three theoretical approaches: 1) illness as social (disability/ medical anthropology), 2) care theory, and 3) theories of systemic support, parental support, and self-advocacy. First, I will discuss illness as socially constituted using theories from disability, medical anthropology, and disability anthropology (Hartblay, 2020; Ginsburg and Rapp, 2013, 2024; Manderson et al., 2016). Secondly, I will discuss theories of care including anthropological theories of care, and how care is articulated by disability studies scholars (Black, 2018; Block et al., 2015; Kittay 2002, 2011, 2019, 2020; Mattingly 2013, 2014; McKearney and Amrith, 2021). Finally, theories of support and self-advocacy, from the parent's perspective (caregivers/ support network), and the children's perspective, and then specific to the transition period (Currie and Szabo, 2019; De Morais et al., 2018; Green et al., 2011; Kittay, 2020; Manderson et al., 2016; Mazzucato et al., 2018; Picci et al., 2015; Sandquist et al., 2022; Silva et al., 2017; Titgemeyer and Schaaf, 2020). In considering how I might incorporate these three theoretical approaches into my upcoming research, I consider that in research about PLWRD and their parents, there are deficits in systemic support, and the transition

process is often unclear, which is critically important because systemic supports changes with the transition. Upon exploration of all three approaches, I would consider incorporating all three since they are interconnected. If disability is about the relational category, care and support are central. When care is central, that means PLWRD is being listened to and supported.

I am putting the three theoretical approaches as separate things, but they are not necessarily separate. They are essentially complementary to each other because there is an overlap between them. In that Ginsburg and Rapp are medical anthropologists, so is Mattingly. There is a new emerging field, disability anthropology, that Ginsburg and Rapp are moving into, as well as many other theories, such as Pamela Block and Cassandra Hartblay. Also, Mattingly is a medical anthropologist. However, she engages in the theory of care, such as Eva Kittay, who uses different theories of care in her work.

**Illness as Social / Disability Studies**

Medical anthropology is a big field that is continuing to grow, and there is no clear definition for it. Since there is no clear definition, I will be using Manderson et al. (2016) definition, “medical anthropology helps make sense of suffering as a social experience, but it does much more than this. It carries into refugee camps, birthing centers, factories, boardrooms, goals, rehabilitation centers and schools, across countries and between communities.… it is also a field of great privilege; medical anthropology takes us into the most intimate aspects of people’s lives, and the most intimate expressions of their joy, anxiety, grief, and tenderness” (p. 2). Therefore, I plan to do is to focus on how the disease is a cultural, social, and intersubjective experience because it is not simply biological.

Moreover, I will be taking cues from disability anthropologists, such as Cassandra Hartblay and Ginsburg and Rapp. Hartblay (2020) states that disability anthropology has been around for a while because there is a long history in anthropology and “qualitative ethnographic inquiry” from sociocultural anthropologists’ explanations of disability. Also, Hartblay (2020) defines disability as “a chronic impairment that significantly impacts the daily life of a given individual, is a complex category with culturally

contingent political and social meanings" (p. 26). Ginsburg and Rapp (2013) add to this by stating that disability is a "relational category, shaped by social conditions that exclude full participation in society" (p. 53). Also, they invented the term *New Kinship Imaginaries*, which emphasizes, "the reality that families are not only flesh-and-blood collaborations but also acts of cultural imagination, encompassing or excluding the fact of disability within family narratives" (Ginsburg and Rapp, 2024, p. 55).

Additionally, disability scholars' recent approaches show that disability is not something "lodged in the body", but it is created by social and material conditions that ""dis-able" the full participation of those considered atypical" (Ginsburg and Rapp, 2013, p.53; Hartblay, 2020). Moreover, Ginsburg and Rapp's (2013) article on "Disability Worlds" is the first ever article in the Annual Review of Anthropology addressing disability. Ginsburg and Rapp (2013) state that for a long time, disability has been labelled as a term that "dehumanizes populations", but once anthropologists began researching how disability is bordered through social organizations in a person's life, being able to understand personhood, and governmentality. Also, Ginsburg and Rapp (2013) argue "that this insight is so foundational yet so often neglected that anthropologists should be encouraged to integrate disability into virtually every topic they study and teach. Although the discipline has been a latecomer to this growing field, anthropology has landmarks that established what we might contribute to this emerging area of study" (p. 55). I will integrate disability anthropology approaches into my research since most rare diseases can cause disability and thus my research will contribute new insights to the emerging field of disability anthropology. Overall, if disability is about relational category, then care is central.

I will be using medical anthropology and disability anthropology theories to provide the framework for understanding health and disability through the lived experiences of my participants in the pediatric and adult healthcare systems. I will be able to analyze how medical professionals' behaviors intersect with their lived experiences. It will lead me to reveal a deeper understanding of the systemic barriers and lived experiences of the PLWRD. Furthermore, using these theories will provide the framework for understanding health and personhood through experiences of the social support networks experiences. I

will be examining the experiences of social support network members who are caring for PLWRD, so I will be analyzing how cultural beliefs influence family dynamics. I will explore societal expectations and how roles as social support networks are formed. So, I will analyze how social structures (family and kinship) can impact health and disability experiences and influence their access to resources.

## Care Theory

Care is universal and can mean something different for everyone. McKearney and Amrith (2021) state that humans support each other through acts of care "because care is a relationship, rather than a biological quality of individuals, this universal varies along with other forms of social variation" (p. 1). McKearney and Amrith (2021) continued by noting that "care is a human universal. But, humans universally structure, practise, and imagine it differently, creating vital differences in people's lives" (p. 12). In my research, care emerges in the lives of families with PLWRD and is a significant part of how social networks are built. Moreover, anthropologists have been talking about and studying care for a while now, but each anthropologist approaches it differently. Black (2018) states that for some anthropologists, care is "a shifting and unstable concept". From a phenomenological approach, communicative activities of care are embodied experiences and social actions (Black, 2018). Black (2018) stresses that morality/ ethics is embedded in social actions, and essentially, embodied communication is the centre of care activities and the establishment of moral/ethical care. Care activities include bodily care, labour to ensure supplies, medications, services, etc. Additionally, increasingly advanced technology is frequently used in care work. Black (2018) states that anthropologists understand that "new technologies shape the adaptation of caregiving enactments, how these enactments are morally/ethically evaluated, and how cultural/socioeconomic distinctions impact uptake, use, and availability of technology" (p. 84). With the use of technology, it can help PLWRD who are non-verbal communicate with others, and vis versa, help parents find support online when they are not able to receive any in-person, and overall, Block et al. (2015) concludes that technology has been introduced to "enhance quality of life" in health care.

Furthermore, to be able to understand how to care for an individual, you must listen to them. In my research, listening to children, especially during the transition period, is important so that they can explain what they need and do not need. This is why I will be looking at disability notions of care[14-17]. Kittay (2020) states that there needs to be an ethic that will guide care relationships between the care provider (social network) and the person with a disability in need of care. Kittay (2020) appreciated learning about giving care from her disabled daughter. Kittay (2020) argues that care should be observed as "compatible with respect" instead of "compatible with paternalism". She is making an argument that a few scholars disagree with, which is that people do not need to prove that they deserve care and respect, especially individuals with a disability. Kittay (2020) stands by her stance because she knows her daughter will never be able to care for herself, yet she still deserves care and respect. In other words, this is essentially about society deciding who should stay and who should not. Additionally, Mattingly's (2013) study on moral laboratories adds to this decision because her study examines African American families' moral work caring for disabled children and observes the experiences which are considered "experiments". Mattingly (2014) shows each moral experiment through a narrative, so the reader will understand what the children went through, and the struggle the families went through to create a good life. I am going to adopt this method from Mattingly of showing the "experiment" through narratives in my work since I want the voices to be the focus. Mattingly's (2014) work shows how parents would do anything they can, so their child can have a "mortality good life". However, some parents take it too far to the point where they lose their role as a parent because they begin treating their child as their patient, or even worst, the disease. Overall, Mattingly's (2014) focus on moral laboratories showed that a moral life is about the parents and health professionals fighting for what they think is right for the child. The parents and doctors care for the child and want the best outcome for them. But both have different opinions of what that looks like. The child gets put into the middle, not knowing what to do. Even though the child is the essential reason why they are in the hospital, they get placed on the side without getting a say. By focusing on how the parents and health professionals are the ones going back and forth, the children get overlooked, which is the deeper

problem. Social support networks are supposed to show care and support towards the child, but in this case, they are not even acknowledging the child in the first place.

"Care is a way to orient ourselves and direct our energy toward something or someone" (Nishida, 2022, p. 9). Care is used in many ways, so in this thesis, I will use Eva Kittay's care theory and feminist theory of care, and Cheryl Mattingly's moral laboratories theory that engages in the care theory of family's moral work of caring for disabled children. I will use both ways of care theory as frameworks to examine the care relationships between PLWRDs and their social support networks. I will analyze how family members form their roles and the societal expectations of how to care for the PLWRD (e.g., the mother's taking on the primary role).

In this thesis, I use the term "care" very frequently, and since care is used in different ways, let me clarify how I am using it. When I describe my data and use the term "care", I am speaking about the everyday usage of care, the different care relationships, healthcare, personal care, caregiving, and caregivers. When analyzing my data, I will use the term "care" in the care theory used. When using Kittay's care theory, I will be using the term not just as an action but also as a relational practice. For example, I will be highlighting the care relationships between PLWRD and their social support networks and health professionals. I will stress the value of care using Kittay's theory to show the social significance of care because it is an important part of a human being's life. Lastly, I will underline the care support that the PLWRD and their social support network members give each other (e.g., mutual support between each other). When using Mattingly's moral laboratories, I am using the term in relation to caregiving and caregivers through stories and experiences for those caring of the PLWRD. Care is controlled by social and cultural factors, which shape how it is given and received/experienced. Also, I will be using the term in relation to care relationships since care is being given and received both ways, so both sides have their roles in how they are providing care.

**Support~ Parental, Advocacy, and Self-Advocacy**

I will be looking at three different categories of advocacy: 1) systemic, 2) parental, 3) self-advocacy. Systemic support involves educating the public and working towards making long-term changes that will help a group of people. Parental support involves parents advocating for their children with disabilities by using their knowledge on the type of disability to get the appropriate services/support and improve educational access and outcomes. Finally, self-advocacy is when an individual educates themselves on a particular issue to speak up on what should be done and changed.

The unusual circumstances of living with a rare disease usually mean there are fewer established support groups, fewer relevant clinical trials, and a shortage of resources for affected children and their families. As a result, social support networks are essential to coping with experiencing a rare disease. Social support networks include family, friends, and relationships with individuals and groups found through institutions such as hospitals, research institutes, and community groups (in-person and online), who may provide care and support when the children and their families need it. The types of care that social support networks provide differ depending on whether the disease is short-term or long-term because longer-term needs may be indefinite in nature. Multiple studies around PLWRD and their parents and the transition tend to take place in the United States, Europe, or Brazil (Mazzucato et al., 2018; Sandquist et al., 2022; Silva et al., 2017). However, aspects of this work can certainly be generalized to Canada, and my research will expand our knowledge of the Canadian context.

### *Parental Support*

Before and after the transition, PLWRD and their families do not receive support that they need, but parents develop strategies (online and in-person) to get through the challenging times, however, as much as parents do, there are barriers they will face along the way. Several studies discuss the strategies parents develop to get through the challenging times and create communities (Currie and Szabo, 2019; De Morais et al., 2018; Green et al., 2011; Kittay, 2020; Manderson et al., 2016; Picci et al., 2015; Titgemeyer and Schaaf, 2020). Green et al. (2011) acknowledge that parents are successfully developing strategies to create communities for their children and

themselves. Green et al. (2011) adds on to explain that it is the children who are helping make these communities come about by serving as "connecting links" (p. 137). Similarly, groups of parents continue to create support groups/communities but have been doing it online instead of in person, especially using Facebook. Titgemeyer and Schaaf (2020) emphasize that "Facebook is widely used as a tool for support groups for individuals affected by rare pediatric diseases… for approximately every fifth rare pediatric disease, one can find an existing Facebook support group" (p. 6). Parents are turning to Facebook support groups because they are feeling lonely, isolated, and disconnected from their society because not everyone knows how it feels to be a parent of a child with a rare disease (Titgemeyer & Schaaf, 2020; Titgemeyer & Schaaf, 2022). Facebook support groups are allowing parents to connect from different parts of the world, learn, share advice, and listen to what each other has to say. Titgemeyer and Schaaf (2020) report that their study confirms that Facebook increases accessibility for parents to join support groups since it is easier than attending an in-person one. Especially during the COVID-19 pandemic, Facebook support groups must have helped several parents get through the challenging times. They all can relate to the fear and anxiety of either their child or themselves catching the virus and knowing they have somewhere safe to speak with others going through similar situations. I will use Facebook support groups to recruit participants because I feel it is the most effective way to reach out to families who may like to participate and spread the word to others they know.

Currie and Szabo (2019) conducted two studies using "interpretive thematic analysis" and identified seven challenges and issues parents have faced while taking care of their children with a rare disease: parents having more knowledge, taking on several roles, feeling silenced and disconnected, having to work hard to be heard, having to make sacrifices, lack in healthcare system organization, and lack of government support. In addition to the seven, I would add language barriers, religious priorities, the shift towards the two-tier health care in Ontario, and COVID restrictions, and I predict these themes will appear in my research data as well.

***Self-Advocacy***

Sandquist et al., 2022; Mazzucato et al., 2018; De Morais et al., 2018; Silva et al., 2017 studied PLWRD and social support networks. Ultimately, listening to the child is the most important step in this research area. Silva et al. (2017) conducted a study to identify key networks and supports from the perspective of a child with a chronic disease; and two themes were found: 1) strong social network and 2) weakened social network. In the first theme, children have strong social support networks around them who are knowledgeable about their medical condition and treatments. Whereas in the second theme, children have weakened social support networks meaning they do not receive the proper amount of support and care as they should. It is unfortunate to learn that some children with a rare disease are living with weak social support networks because as Silva et al. (2017) state, "children need unique and humanized professional care, which cannot be fragmented, technical or lack dialogue, however it must consider extended care" (p. 7). While there are studies that focus on the child, studies are lacking about the transition period that PLWRD experience. Sandquist et al. (2022) acknowledge that there are challenges during the transition to adult healthcare, and only a few PLWRD and their families receive transitional support. Understanding the challenges that come with the transition period is critical. Once we know the challenges, then improvements will begin to happen because there is a large amount of PLWRD experiencing health inequalities during the transition (Sandquist et al., 2022). Also, Sandquist et al. (2022) indicate the barriers during the transition, such as "the loss of ancillary staff common in pediatric settings, healthcare, culture differences, and even simply the work involved in transferring medical records" (p. 3). Overall, there is no support, parents run into all types of problems, and most studies focusing on the transition, are about people with a commonly experienced disability or chronic condition, not a rare disease. Our understanding of the transition period remains incomplete because research in this area is minimal, especially in Canada.

## Conclusion

In summary, this literature review was used to create a theoretical background for the investigate of the social support networks of PLWRD, their experiences within the pediatric health system, and then their transition to the adult health system in Canada. In

this thesis, I will use three theoretical approaches: Illness as Social, Care, and Support. To decide if I might incorporate these three theoretical approaches into my research, I have to take into account that I know there is research about PLWRD and their parents, there is no support given, and the transition period is still not clear, which is important and critical because supports change with the transition. From exploring all three approaches, I consider incorporating all three since they connect to one another. As previously mentioned, if disability is about relational category, then care is central. When care is central, that means the PLWRD is being listened to and supported the way they should by their social network.

Overall, what my thesis will show, and present is how my anthropology includes disability anthropology, medical anthropology, and care theory. This is my particular contribution to the broader anthropological project of how we as anthropologists look at health, disease, bodies, kinship, and personhood. My anthropology refers to a unique perspective and understanding of social relationships and human behaviors that shape the individual's experiences and beliefs. It also contributes to the understanding on how societies understand and manage disability, health, and disease.

## 1.4 Methodology

The fieldwork for this research was done from May to December 2023 in Canada. Due to COVID-19 still spreading, I opted to conduct my fieldwork remotely. My fieldwork was supposed to be from May to August 2023, but some of my participants either wanted follow-up interviews, needed to reschedule the first interview due to health matters or had some new participants reach out wanting to participate.

### 1.4.1 Recruitment

I conducted my research in Canada, so my goal was to recruit participates from different provinces to get a range of perspectives and experiences. All but two of my participants are from Ontario, and two of my participates are from Saskatchewan but had to travel to British Columbia. Even though most of my participants were from Ontario, all were from different cities/towns, I still got a range of perspectives and experiences.

When it came to deciding who to include in my research in terms of only allowing people 18 years old and older to be interviewed, or people of all ages. I came to the discussion of not allowing participants under the age of 18 years old be included because I conducted everything remotely, and my methodology plan to include children would not have worked effectively. My original plan of including children was to use the method of drawing-story process combined with interviews about what they drew, which Silva et al. (2017) has used.

I would consider the drawing-story process an approach under art-based research (ABR), which gets used to "inform or generate knowledge about the human condition" and recognize things that we don't usually see (Casey and Murray, 2022). Art itself is such a powerful skill that many people use to express themselves and share their thoughts and stories through their drawings. ABR may sound questionable to some people, but art as a methodology in qualitative research methodology has been accepted to be "valid" research since it expands on the understanding and exposure of live experiences (Casey and Murray, 2022). Overall, art as the method offers "more nuanced and unsettled meanings", it urges the readers to create their interpretations apart from what the researcher presents (Casey and Murray, 2022).

Furthermore, combining the drawing-story process with interviews, the interviews provide the children's narratives and stories. The use of narrative methodology (in qualitative research design) is for exploring, uncovering, and understanding the individual (Rau and Coetzee, 2022). Narratives are events that get told in a sequence which essentially makes up a story (Rau and Coetzee, 2022). The goal of narrative work is to "generate comprehensive, layered, nuanced understandings of human experience and meaning-in-context" (Rau and Coetzee, 2022, p.5). Silva et al. (2017) did a great job using the drawing process to allow the children to express their experiences and then get them to tell their stories through them. The reason why they chose to speak with the children and hear their stories is that "the stories they tell and how they understand their life contexts are challenged by memories of past events and present and future representations" (Silva et al., 2027, p. 2). Overall, the drawing-story process combined with interviews is a great method used in research like this when children are involved

because it makes the experience less formal and stressful. Children get to express what they feel and went through in a way that they are in control of, which I feel is the right way to go about it. I hope I can use this method in my Ph.D. research, so I can include participants under the age of 18-year-olds.

I recruited participants through community groups, including online communities on Facebook and X (formerly known as Twitter) through snowball sampling. Snowball sampling is considered to be a popular method in qualitative research (Parker et al., 2019). This method of recruiting helps access “hard-to-reach populations”, which for my research is beneficial since the population of people I am studying is small and rare (Parker et al., 2019). I used snowball sampling by distributing my recruitment material (e.g., flyer) in pharmaceutical spaces with the owner’s permission, sharing it with Western University’s open “Must Know” Facebook page and, on my Facebook, Instagram, and X (formerly known as Twitter) pages. In my posts on X, I added common hashtags such as #raredisease and #chronicillness to reach a larger audience.

Also, I contacted three Canadian rare disease organizations for help with gaining participants. I did not have access to their patient database, but the organizations shared my recruitment information with their network via email and or on their social media platforms, such as Facebook, X, and LinkedIn. The three organizations I contacted were the Rare Disease Foundation in Vancouver, BC, the Canadian Organization for Rare Disorders in Toronto, ON., and the Rare Disease Network of Alberta. However, only the Rare Disease Foundation and the Canadian Organization for Rare Disorders posted my recruitment information on their social media pages.

Lastly, I posted on "RareConnect", a website filled with various rare-condition communities and people from all over the world posting information and stories, resulting in several networks. Unfortunately, I had no luck finding people in Canada interested, instead, I had individuals from Brazil and Algeria contacting me about wanting to participate.

### 1.4.2 Semi-Structured Zoom Interviews

I conducted 11 virtual semi-structured interviews via Zoom with PLWRD (18+) who have previously experienced support from children's hospitals, family members, and various other services in Canada. I interviewed individuals within the PLWRD network as well (such as parents and service providers). Semi-structured interviews allowed the interviewee to feel comfortable and be in control when telling their experiences while still within a structure of questions created beforehand (McGarry and Mannik, 2017). Some questions were sensitive, so the open nature of my approach allowed individuals to focus on topics they feel are relevant and safe for them to discuss (McGarry and Mannik, 2017). By gathering, analyzing and identifying patterns in these accounts of the experience of PLWRD, I will increase our knowledge of the development and roles of social support networks and the changes once the youth (or individual) transition out of pediatric care in Canada.

The interviews were video and audio-recorded and lasted one to two hours. Even though I had prompt questions, the participants were the ones who had full control of the interview. They could stop the interview whenever they felt like it, skip questions and topics they did not feel comfortable answering, and some even asked for me questions! Some family members participated together, so I gave each participant the option to be interviewed separately or together. I ended up conducting three group interviews and eight individual interviews.

I invited each participant to share videos and photos to help me learn about them, their relationship with the PLWRD, their support network, and their medical journey before and after the transition. This was optional for those who were interested, and the main reason was to help support my data collection and analysis and to be included in the dissemination of my research. Each participant who shared photos and videos filled out a "participant photographic release form" and was given a link to a "Photos and Videos" folder, which is stored in my Western OneDrive that I only have access to. Each folder is labelled with the participant's pseudonym. For the participants who shared their photos and videos before the interview, I asked them if it was okay with them if we went through

them at the end of the interview because that allowed them to explain to me what was happening in the photo or video and furthered our conversations.

Throughout each interview, I wrote notes, and once the interview was finished, I made my final notes. One thing I regret is waiting until I finish all my interviews to start transcribing them, even though I planned to begin transcribing after each interview. However, it all worked out in the end because while transcribing, I was able to clarify some of the notes I made during the interviews and write down more that I missed. This process truly helped me lead into my next step, data analysis and coding.

### 1.4.3 Data Analysis and Coding

When it came to how I would be conducting my data analysis, I was conflicted between using software, such as NVivo, or doing everything manually. I ended up doing everything manually. The coding and data analysis process was done in three steps. First, I transcribed all the interviews in a Word document. While transcribing, I was also taking notes of patterns, common themes, similar terms used by the participants, common challenges and supports, etc. Second, I used an Excel document to categorize all the information about my participants and the common themes. Third, I then began color-coding the common themes. For example, all paragraphs or references relating to transition were colour coded with blue, support dynamics were purple, challenges were yellow, supports were orange, and advice and advocating were pink.

By the end of step one, I was left with several pieces of paper because I printed off each transcript to be able to write directly beside certain sections, and all the notes I made were in notebooks. So, I decided to organize my data further by transferring everything to an Excel file. In the Excel file, I had a row for each participant and then columns identifying if they are the PLWRD or members of their network, their rare condition if they have transitioned out of pediatric care, and then a column for each theme. I began transferring the notes I made on the transcripts and notebook into the Excel file, so when I began writing, I did not have to flip through several pages, instead, it was all in one file.

### 1.4.4 Ethics

My research required ethics approval since I will be interviewing people. I submitted my REB application on February 26th, 2023. I had some revisions to make on my application based on the recommendations I've received. On April 27th, I received ethics approval and began my research (Appendix A).

### 1.4.5 Participants

I had 14 participants in total, five PLWRD and nine social support network members (Tables 2 and 3). As previously mentioned, some family members participated together, but I also had solo participants, mainly social support network members. In Table 4, those four PLWRDs were not able to participate because they were either not able to or were under the age of 18 years old (one of the eligible requirements of this research is that you must be 18 years or older).

The participants were all female, except for two males. One of the males was a father, and the other male was a PLWRD. I was hoping for a mixture of genders to get different perspectives since in research, it portrays mothers of being the primary caregiver who does all the work, and the fathers do not do any or much to help. However, in my research, I will show that fathers do help and have significant roles in taking care of and supporting their children. Even though I had one father participate in my research, I did ask the mothers about their husbands' roles during their child's medical journey and was able to get more information.

The age range of the PLWRD who were interviewed was between 19-30 years old, the PLWRD who were not interviewed was between 15- 30 years old, and the age range for the network members was 44- 63 years old. This range of ages provided different and valuable perspectives and experiences because things have changed over the years, such as the advancement of technology, increased research, new treatments, new programs, etc.

Tables 2, 3, and 4 are summarize participants' information relevant to what I discuss in the rest of the thesis. But, when it came down to names, I chose to use pseudonyms

instead of real names to keep my participant's identities private. Every participant was given the option to choose their pseudonym, but almost all the participants allowed me to come up with their pseudonyms.

**Table 2- Participant's Information: Youth**

| **Interview No.** | **Pseudonym** | **Rare Disease** |
|---|---|---|
| 1 | Lucas | Nager Syndrome |
| 2 | Lily | Cutis Marmorata Telangiectatica Congenita (CMTC) |
| 3 | Gabriella | Autoimmune Encephaloradiculomyeloneuritis-<br><br>(Type of autoimmune autonomic ganglionopathy) |
| 4 | Asher | Synovial Sarcoma |
| 5 | Addison | ~ CRPS (Complex Regional Pain Syndrome)<br><br>~USAID (Undifferentiated Systemic Autoinflammatory Disease)<br><br>~REM Behaviour Sleep Disorder |

**Table 3- Participant's Information: Network Members**

| **Interview No.** | **Pseudonym** | **Relationship** |
|---|---|---|
| 1 | Sebastian | Father of Lucas |
| 2 | Tori | Mother of Lucas |
| 3 | Peyton | Mother of Rose |
| 4 | Susan | Mother of Lily |
| 5 | Sarah | Mother of Asher |
| 6 | Harper | Mother of Theo |
| 7 | Zara | Mother of Addison |

| | | |
|---|---|---|
| 8 | Julia | Mother of Ivy |
| 9 | Amelia | Mother of Logan |

**Table 4- PLWRD Who Could not Participate, But Their Network Members Did**

| **Interview No.** | **Pseudonym** | **Rare Condition** |
|---|---|---|
| 1 | Rose | Joubert Syndrome |
| 2 | Logan | Immune Thrombocytopenia Purpura (ITP) |
| 3 | Ivey | Rare Disease Disclosed |
| 4 | Theo | Focal Segmental Glomeruloscerosis (FSGS) |

## 1.5 Themes

I have structured my thesis chapters around the themes that emerged from the interviews. As I previously mentioned, I have found 19 recurring themes and categorized them into five groups: The Transition Stages, The Support Dynamics, The Challenges, The Supports, and The Power of Advocating (Table 1). The two other themes which I will be using throughout the thesis are Storytelling and Emotions.

Even though my thesis is structured around my themes, my four research questions be answered within each chapter.

*1) Who participates in the support and social network, and what do they provide, seek, find and find lacking in terms of support through their participation?*

- The Transition Stages
- The Support Dynamics
- The Power of Advocating

*2) What role do social support networks play in the transition from pediatric to adult care?*

- The Transition Stages
- The Support Dynamics
- The Power of Advocating

*3) What new social support networks and support needs take form during and after this transition?*

- The Transition Stages
- The Support Dynamics
- The Power of Advocating

*4) What are the supports and challenges PLWRD and their social support networks face in pediatric care that they continue to face in adult care? What new challenges and support emerge in adulthood?*

- The Challenges and The Supports
- The Power of Advocating

The theoretical and conceptual framing that I will use to make sense of all these themes and answer my research questions are theories of care (Kittay, 2011; Mattingly, 2014; McKearney and Amrith, 2021), theories from medical anthropology and disability anthropology (Ginsburg and Rapp, 2013, 2024; Manderson et al., 2016), and theories of support and advocacy (Currie and Szabo, 2019; De Morais et al., 2018; Green et al., 2011; Kittay, 2020; Manderson et al., 2016; Mazzucato et al., 2018; Picci et al., 2015; Sandquist et al., 2022; Silva et al., 2017).

# Chapter 2

# 2 The Transition Stages

*"Without adequate support during the transition, there is an increased risk for inadequate follow-up and poor self-management leading to poorer quality of life for the patient, with increased hospitalizations, increased rehospitalizations, and increased healthcare costs" (Green Corkins et al., 2018, p. 82)*

In this chapter, I take you through the three stages of transition: the before (pediatric care), the in-between, and the after (adult care). Transitioning to the adult care health system from pediatric care is a difficult period, with challenges PLWRD[2] and their support network face. Some families are fortunate to receive transitional support, but some are not. PLWRD experiences health disparities in transition and adult care (Sandquist et al., 2022; Reiss and Gibson, 2002). For example, there is a lack of available treatment services in adult care, a lack of knowledge, and a lack of acceptance of PLWRD for treatments. No specialists are willing to take a PLWRD as a patient in adult care because of their disease, and that is a struggle for pediatric doctors who want to make referrals before PLWRD transitions out (Reiss and Gibson, 2002). The other reason specialists do not want to take PLWRD as their patient is their lack of knowledge about the disease since it is outside their scope of specialization (Reiss and Gibson, 2002). Lastly, there is a lack of acceptance of PLWRD for treatments because the life-sustaining treatments that PLWRD had in pediatric care are only available in a "pediatric treatment setting" (Reiss and Gibson, 2002). Also, the transition involves the change of who is responsible and in control of the healthcare decisions. This transfer of responsibilities is a major and overwhelming event for PLWRD but also the parents because they fear letting go[3] (Corkins et al., 2018).

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[2] People Living with a Rare Disease

[3] This will be discussed further in Chapter 3.

Take a step back for a moment to understand the history of *transition* for this population. By the 1970s and 1980s, medical advances began to form. Before then, the transition to adult care for children with complex medical conditions was not an issue because only a few of those children survived to adulthood (Reiss and Gibson, 2002). In the 1980s, a rise in awareness in the healthcare community began for the need to accommodate the healthcare needs of individuals with complex medical conditions who are making it to adult care (Reiss and Gibson, 2002). In the 1990s, there was progress in recognizing the factors that impact transitioning, creating demonstration programs for transition, defining key components for transition programs, and identifying the needed changes in professional knowledge and skills (Reiss and Gibson, 2002). Overall, as Reiss and Gibson (2002) state in their study back in 2002, which still stands today (22 years later), "changes in both the pediatric and adult health care systems are necessary for health care transition to be a successful process. However, these system changes are long-term and will require many years to complete" (1312-1313).

Of all my participants, a majority expressed how there is little to no support in preparing for the adult care health system in pediatric care (the before stage). This relates to the concept of "unplanned survival" that Pamela Block discusses in her article in Danilyn Rutherford's, *Disability Worlds: Wenner-Gren Symposium Supplement 21* (Rutherford, 2020, p. 74). In the past, children with complex medical conditions rarely lived into adulthood. Those who have are examples of unplanned survival, such as Nick in Block's article who is one of the earliest examples (Rutherford, 2020). Block explains how "advances in medical technologies and care systems have extended life for an increasing number of people. Yet the policy mechanisms that would allow for more than pure survival are not in place. Without the material and social structures of support, such youth are in a holding pattern, surviving but not thriving, with little opportunity to do more than exist. There is no doubt that with the proper supports in place, many could be living in the community, going to school or participating in work or day programs, engaged in meaningful pursuits" (Rutherford, 2020, p. 74).

In the in-between stage, when the PLWRD are 16-17 years old, doctors should prepare PLWRD for what happens when they turn 18 years old. When the PLWRD gets to the

adult care stage, it is a whole new place and runs differently than pediatric care. PLWRD and their families are trying to find a doctor(s) to take them as patients and try to figure out how everything in adult care works since it is a different environment. Also, the PLWRD are now in control of their health, forming their independence, and family members (especially parents) are stepping back from being the point person of the PLWRD health.

Throughout this chapter, I will share the experiences of three of my participants going through the transition stages. The first experience is from the PLWRD who needs to travel to another province for healthcare, the second experience is from the PLWRD who stayed undiagnosed throughout pediatric care until she got to adult care, and the final experience is from the support network member whose PLWRD has not transitioned yet. Also, I will be analyzing the experiences using disability theories of care to explain the importance of care for PLWRD (e.g., healthcare, emotional support, medical care, caregiving), Mattingly's moral laboratories, and using cues from disability anthropologists Ginsburg and Rapp to examine how disability shapes kinship. There is an analysis section (2.4) at the end of the chapter, but I also analyze concepts of structural issues PLWRD face throughout the chapter.

## 2.1 Before Transition: Pediatric Healthcare

*"You get thrown into this massive medical world with no one to hold your hand unless you find someone to help you" – Parent 1 (Currie & Szabo, 2019, p. 98).*

Once a child is diagnosed with a rare disease, their life changes, and their family as well. It is because rare diseases are chronic conditions, they are unpredictable, and there is a constant need for medical screenings, appointments, and medication (Larotonda 2016). However, before any of this, getting a diagnosis is very difficult for many PLWRD and can take a while because sometimes it takes a couple of weeks, months, or years, and for some people, it is not until they get to adult care when they receive an official diagnosis. Usually, PLWRD is referred to numerous doctors and or specialists before getting a diagnosis. This leads to delays in treatments and care, which results in a negative impact on the PLWRD health (Rare Diseases Working Group Report, 2017). When receiving a

diagnosis or waiting for one, PLWRD needs to form a pediatric team of doctors, specialists, and other medical professionals, depending on their condition. This is usually done by the parents who take the lead and control of the PLWRD health in pediatric care.

In Canada right now, there is a crisis where numerous Canadians are struggling to find a family doctor (Tasker, 2024). In 2021, 85.5% of Canadians had a regular healthcare provider, but 14.4% (4.7 million Canadians) did not have one (Statistics Canada, 2023). Also, 7.9% (2.5 million Canadians) reported that their healthcare needs were unmet (Statistics Canada, 2023). An article from 2004 about frequently asked questions about pediatricians in Canada states, "It's estimated that between 30% and 40% of children's visits to a doctor for primary health care are to a paediatrician. The majority, then, see a family physician for their ongoing health care" (Paediatricians in Canada: Frequently asked questions, 2004, p. 432)[4]. This was because there were only 2300 practicing pediatricians, but the availability of pediatricians depends on the region (Paediatricians in Canada: Frequently asked questions, 2004). For example, in cities such as Ottawa, Toronto, and Winnipeg, and provinces such as Quebec, children see pediatricians for their care (Paediatricians in Canada: Frequently asked questions, 2004). Whereas in British Columbia, the Maritimes, and large urban areas, children see family physicians and children with complex needs then get referred to pediatricians (Paediatricians in Canada: Frequently asked questions, 2004).

In this section of the chapter, I will take you through three different experiences in the pediatric care stage. First will be Lily's experience travelling to a different province for care, second is Gabriella's experience of not receiving an official diagnosis during pediatric care, and the third experience is Theo, who has not transitioned yet into adult care. This experience will be from the mother's perspective, Harper.

[4] I researched for updated statistics but could not find any.

### 2.1.1 Saskatchewan to British Columbia Lily Goes

Lily was born with CMTC (Cutis Marmorata Telangiectatica Congenita), a rare skin condition. It is a vascular malformation that affects the capillaries. Lily has a capillary malformation down the entire right side of her body, on the left side of her face, neck, and skull, and in some of her internal organs. For many PLWRD, the reality is where they live; there may not be doctors, specialists, or treatments for them, and that was Lily's case. Growing up, Lily and her parents lived in Saskatchewan and had to fly to British Columbia to be treated, specifically at the clinic that specializes in vascular anomaly. Even though the clinic was Lily's primary area of knowledge, they were not considered her primary doctors because she lived in Saskatchewan. So, Lily and her parents flew every 2-3 years to the vascular anomaly clinic to see the doctors for a couple of days and get all the information on what should be done for the next couple of years back to Saskatchewan to tell the doctors.

Lily shared how since she had many little issues that required for her to have different doctors. This resulted with her forming a great team who listened and communicated to her, but also to each other. So, they were a big part of her social support network in pediatric care. But there were two doctors in particular who Lily called her **quarterbacks**. They cared for her alongside her family doctor, so they handled getting her referred to different specialists that she needed to see. *During the interview, when she called them her quarterbacks, I was curious to know why she called them that and the significance behind it*. It was because her dad was a sports coach, which created the bond between her dad and the doctors. Also, it helped Lily, and her dad make sense of things. Quarterbacks are usually the ones who call the plays and make the decisions on the field. So, Lily was the 'ball', the treatment was the 'end zone', and the quarterback was the one who was responsible for trying to get Lily to where she needed to go. Also, they make 'plays' with the other 'players' (the other doctors on the team) to make it happen[5].

[5] This is a great analogy for a kid!

*I found this to be amazing because it truly helps the child, and the parents understand everything more when using something they know. Also, this shows the level of care and support those two doctors gave Lily and her family to get referred to as "quarterbacks", which is not common for many PLWRDs.*

The other half and a big part of Lily's social support network in pediatric care was her parents. Her mom and dad were constant supports during the pediatric stage, such as going to all the doctor appointments, flying to BC, doing their own research, etc. Most importantly, they were there for her as she began to prepare for the final stage, adult care, which was not easy.

### 2.1.2 Undiagnosed During Pediatric Care

Gabriella was 14-15 years old when she began getting sick and was admitted to the Children's Hospital. She was undiagnosed all throughout pediatric care and officially got a formal diagnosis when she got into adult care. Her diagnosis was a type of autoimmune autonomic ganglionopathy called "autoimmune encephaloradiculomyeloneuritis". In pediatric care, they did not know what the future would look like for her, whether she would need more or less support, and if she would get better soon. So, it was up in the air in pediatrics, which made things hard for everyone. Gabriella was consistently an inpatient during her time in pediatric care because she would spend a few weeks at home and then a few months in the hospital. She basically did all her high school from the hospital.

How Lily had to travel to BC, Gabriella had to travel for specific tests. Gabriella is from Ontario, and the hospital she was at was good, except for the pediatric side, since they did not have all the tests she needed. She had to travel to Boston for tests because Canada did not have it, and another time, she had to travel to Edmonton, Alberta because Ontario did not have it.

Throughout Gabriella's time in pediatric care, her main support network was her mom, sister, and the friends she made at the hospital. Her mom stayed by her side throughout everything, and her younger sister too. Also, the friends she made in the hospital, she

stayed good friends with and were good support for Gabriella because even though they were all going through different things, they all had similar experiences.

*I asked if Gabriella if there were any organizations or support groups/ programs available at the hospital or in her city during the time she was in pediatrics. Would it have been beneficial for her to join because she would have been able to connect with others and have people around her almost every day?* Gabriella explained how it would have been great to have some type of support groups/programs because in the States, they had many programs, and she tried to join some of the virtual groups, but they were never the same. Overall, there was nothing like the programs in the States available during Gabriella's time in pediatric care in Canada. However, there was a social worker in the main clinic for the patients who would connect Gabriella with different organizations, get her tickets to hockey games, and even be there for her when she needed someone to talk to for support because the social worker knew her medical team and would talk to them for Gabriella when she was struggling with things.

### 2.1.3 Beginning to Think of the Transition!

Theo was 12 years old when he was diagnosed with a chronic kidney disease called FSGS, which stands for Focal Segmental Glomerulosclerosis. Since Theo is under the age of 18, he was not eligible to participate in this project, so Harper, his mother, participated solo and shared his medical journey. Harper shares that FSGS in kids is rare, and a lot of the kids that do have FSGS are nephrotic, so they also have nephrotic syndrome, which causes them to retain a lot of fluids and swell. Theo was less than 20% of people who have FSGS who are not nephrotic, so he's rare by having FSGS, and even within that subset, he's even rare to not be nephrotic.

At the age of 12 began Theo's medical journey towards receiving a kidney transplant. Theo began peritoneal dialysis when he was 13 years old and did it for six months because Theo's father was a donor match, so he was able to donate his kidney. When Theo was 14 years old, he had the transplant, and that alone was the most memorable moment for the entire family.

Just like how Lily had to travel to get treated, Theo did too. The city Theo lived in did not have nephrologists, so they were automatically referred to a city an hour away from them. So, they must travel an hour every time Theo has an appointment, bloodwork, and even for his transplant. The positive side of this is that the city they were referred to, Harper's whole family lives there, so they have a lot of support there.

Theo's transplant occurred six months into COVID, so there was a lot of extra strict protocol. Theo was at the Children's Hospital, and his father was at the adult hospital across town. The father had to be dropped off at the door with nobody allowed to be with him. Whereas for Theo, Harper went in with him, and she had to advocate to get special permission for her mother to be there because it was only allowed one caregiver during COVID-19, but she was able to get her mother with her. At around noon, Harper got word that her husband's kidney was being driven to the Children's Hospital. What made this moment even more memorable was that Harper's cousin's husband was the police officer who escorted the kidney to the Children's Hospital. Once the kidney arrived, he sent Harper a picture of the cooler in front of the hospital.

So, during Theo's time in the pediatric care stage, he had a great social support network. Harper and her husband are the primary people and their daughter. She was there during dialysis, and her job was to distract Theo by hanging out with him, playing video games, chatting, etc. During the time of Theo's transplant, it was hard because of COVID-19, and she could not come to the hospital or even visit. Also, Theo had his immediate family there for him as well, such as his grandparents, aunts, uncles, and cousins.

## 2.2 In Between the Transition

*"Just because a doctor/pediatrician/ER staff had not heard of a disorder does not mean it does not exist." - Brittany Lazechko Alley*[6]

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6 [6] *(Brentano, 2024)*

Transition is “more than the simple physical transfer of a patient from one practice or hospital to another, but instead, as the designed effort to ensure healthcare independence, preparation, and the completion of this transfer” (Sandquist et al., 2022, p. 3). During this stage, it is time to for PLWRD and their social support networks to begin addressing what comes next, especially once the PLWRD turns a certain age. The age of 18 years old is usually the cut-off age for children to transition from pediatric care to adult care. It was surprising to learn that in Canada, the cut-off age which provincial/territorial funders mandate for the transition to adult care ranges from 16 to 19-years old (Toulany et al., 2022). Also, there is a recommendation for “flexible age cut-offs” for transitioning to adult care with consideration of the individual’s development stage, circumstances, and needs (Toulany et al., 2022). In general, there needs to be specialized training and education about “transitional care issues” to ensure that health care providers are prepared to accept and care for individuals living with a rare disease and complex medical conditions (Toulany et al., 2022). No PLWRD and complex conditions should be rejected care because a medical professional is not prepared or knows how to help.

In this section of the chapter, I will continue taking you through Lily’s and Gabriella’s medical journey towards their transition to adult care, but now during the in-between stage. At this stage of their journey, Lily’s pediatric team began to help her as much as they could with her transition, and Gabriella did not have as much help or time to prepare for her transition. Also, I will finish sharing Theo’s experience since this is the stage he is currently in.

### 2.2.1 “Stretching the limits of what an adult is”

During this stage of Lily’s life, she was in school in Vancouver and had turned 18. The vascular anomaly clinic was such a big support to Lily because they tried to stretch the limits of what an adult is as far as they could. They allowed Lily to stay with them until she was 21 years old, which they used as the cutoff for her. So, the vascular anomaly clinic was Lily’s support system for nine years, and as she transitions into adult care, she is losing her incredible pediatric team. However, their support did not end there because they tried their best to set her up for success in adult care by making referrals to a handful of doctors they thought could work collaboratively. This is what should be done while

PLWRD is still under the care of their pediatric team because involving medical professionals from adult care before the transition will ensure better communication, care needs, the handover of health history and issues will be smooth and allows for the adult care medical providers to get educated further on the care needs of their soon to be patient (Toulany et al., 2022). However, Lily expresses how there were plans put in place for her by her pediatric team who were helping her with the transition to adult care to set her up for success. However, those plans got destroyed by the adult world because they it was demoralizing for Lily being told 'Oh my God! What is this?' and being dropped as a patient due to her complex condition.

There was one doctor in particular that Lily's pediatric team referred her to, who works with many different types of doctors, and they thought he would build a similar team as she had in pediatrics. Unfortunately, that did not happen because the doctor took one look at Lily and told her that her condition was 'so beyond my scope of knowledge, I could not even imagine what I could provide you'. So, he set up a couple of tests for her to do, which ended up getting cancelled. When Lily tried to get in contact with his office, they told her that she had been referred to somebody else and they would let her know when the referral process was ready, but that never happened. It is essentially medical disownment or social abandonment. In Biehl's (2013) book, Vita*: Life in a Zone of Social Abandonment*, the concept of "social abandonment" refers to individuals being neglected and abandoned by society, which has an impact on them. So, people with disabilities, especially PLWRD get overlooked and eventually neglected by those (health professionals) who they need the most to help. In general, disabled people should be valued and respected in society as non-disabled people are; no one should have to prove that they deserve to be valued and cared for (Kittay, 2020).

Moreover, a positive experience throughout this time was when she got referred to a cardiologist, but he told her right away that he deals with heart attacks, so he could not help her. However, he did not just let her go. He told Lily about a clinic that deals with complex heart conditions that would be a better fit. More doctors like him are needed because when PLWRD gets referred to doctors that may not deal with the condition that the PLWRD has, they should make the effort to at least find someone else. Lily expresses

how much respect she has towards this doctor because even though he personally could not help her, he made the effort to find her someone who could and just did not abandon her. The clinic the cardiologist referred her to, she still sees today because they were a better fit, just like he told her they would be.

It is already challenging for PLWRD to find doctors, especially in adult care. So, Lily expresses that she had the utmost respect for doctors that realize when they're in over their head and are like 'I cannot help you but let me find someone who can'. But, when it is like the first doctor who essentially cut ties with her without assisting her with further help like he said he will and made Lily go out and find someone else, "sucks" as Lily put it.

### 2.2.2 Not Enough Support

I asked Gabriella whether her doctor prepared her for adult care or even discussed with her what would happen when she transitioned. She told me they did not discuss anything except that she should start finding specialists in adult care when she turned 17 and had a meeting with a Children's Hospital psychologist about how to advocate for herself. However, the psychologist did not know anything about the adult hospital world.

So, Gabriella had barely to no support in preparing for adult care. She did not know what to expect, and even the person who was helping her how to advocate for herself did not know what to expect either in the adult care world. Even though it was good that Gabriella was receiving help on how to advocate for herself, she should have met with a psychologist from adult care. They not only would have helped her with advocating since they work in the adult care world, but they also would have been helpful with letting her know what it is like and how things work. Also, Gabriella's doctors could have set up meetings with other medical professionals and staff to speak with Gabriella, especially since she spent almost all her time as an inpatient, all the way up to her time transitioning to adult care.

### 2.2.3 What Comes Next?

Right now, Theo is 17 years old, so he is currently in the in-between stage, getting prepared to transition to adult care next year. I asked Harper how Theo feels about the transition, and she expressed how he feels like it is looming, is kind of worried, and sacred of not knowing where to go because it is intimidating to go somewhere else.

This stage is not easy because it is a time when everything changes, such as the hospital, doctors and staff, support relationships, and who has control. For most PLWRD, this stage is very challenging, as we saw in Lily's and Gabriella's experiences. In Theo's case, he is being consistently mentally prepared. He was told that he would be moving from the Children's Hospital to the adult hospital, which is across town from each other, having a whole new team of doctors who would not be seeing him as frequently as he was being seen in pediatric care and would not be getting the same immediate attention. This was a surprise for Theo and Harper when they were told that the doctors in adult care only see patients every six months. Also, it is time for Harper to step back and allow Theo to become more independent. When Theo started out in pediatric care, doctors would communicate with him and ask him questions, but when he was hesitant, Harper would jump in to answer. As he grew up, he gained confidence because the doctors and nurses continued to try and ask him questions and include him in conversations, so he developed those skills and got better at talking. He appreciates that the doctors and nurses talked and addressed him throughout his time in pediatrics. *This is what more doctors and nurses should be doing to help PLWRD prepare for adult care. But also, at this stage of the PLWRD life, parents need to help increase their child's independence by stepping back, loosening their control, and remaining supportive* (Van Den Akker et al., 2010; Corkins et al., 2018).

Many PLWRD have never received, and some still do not receive proper preparation for the transition into adult care. In the US, a parent navigator, TjaMeika Davenport, at Children's National Hospital and a community advisory board member for Got Transition described the transition for children with complex needs to be "the most difficult challenges to navigate as a young adult in the US healthcare system" and "is uniquely difficult to counter" (Sandquist et al., 2022). Davenport and her team made

several efforts to improve the transition for adolescent patients and their families, and one of those supports is the “warm-handoff strategy” (Sandquist et al., 2022). This support includes the adolescent meeting with the pediatric team they are leaving and the adult specialists they are transitioning to to create a support network. *This is something I want to have around Canada, and I was happy to hear from Harper that there is a similar program in place.*

Theo and Harper were told that his nurse practitioner would be taking him to his first appointment at the adult Hospital and would introduce him to his new team, be there for support, and have a familiar face around. Also, Theo has been referred to adolescent medicine, and there is a new program in the hospital Theo joined, Harper explains that,

> “They see kids ages, I do not know if it starts at 16 maybe, and they see them to 25 potentially. So, it's like this overlap between the two, peds and adult world, and to help them with the transition and to help them with any kind of other, other things that come up during those teenage years. So, they're filling a gap. So even though he may be switching hospitals and that'll be scary, having the same person that he can still visit, and they can do a little bit of mental health. They can do referrals. So, it's still like a slight connection with the children's hospital even though he's now moved to adult”.

Theo has been in the program for six months, and during the first meeting, Harper went with Theo, and they explained how it all works. However, when the doctor came out, he only called Theo back, and the two talked over everything. After they finished, Harper was brought in and given a summary of what they had discussed. Harper expresses how this program gives Theo a chance to build up his confidence by getting to talk to the doctors on his own and feels reassured that he has this program until he is 25 if he needs it. Overall, from all of my participants, Theo is the only one who had the opportunity to join a program that will help him with his transition to adult care since it was offered in the hospital where he was receiving care.

## 2.3 After Transition: Adult Healthcare

*"When you have a rare disease, you face two battles. One being the illness itself, and the other, living in a world where so few people understand what you're up against" -Unknown*[7]

The growing attention on the topic of transition is not because of the increasing population of individuals transitioning into adulthood but instead due to the additional burden that this causes for patients, their families, and even some of the healthcare services (Mazzucato et al., 2018). So, it is an important period to investigate. In this final section of the chapter, I will conclude with Lily's and Gabriella's medical journeys into adult care, showcasing how different their transitions were and their experiences in adult care.

### 2.3.1 Adult Care is Here, But Without Lily's Quarterback

Most PLWRD form such a great team in pediatric care who listen and support them, but when the time comes to transition, they must start all over to form another team or at least find a doctor who will take them as a patient. The pediatric team may help make referrals and suggestions, which was the care for Lily, but some doctors in adult care do not want to take the responsibility of taking on a patient with a rare disease because the condition is beyond their scope of knowledge, so they truly do not know how to help. When I asked Lily how the transition was, she expressed that it was rough, and she had to figure out how to be her own quarterback now.

When Lily entered adult care, she had just lost her peds team, which in the past was super beneficial for her because they were like her comfort blanket. Even with the help from her pediatric team, as explained in the last section, was not as successful as they wanted it to be, Lily eventually found a team of doctors in

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[7] (*NCAN's Inspiration Corner, n.d.*).

adult care. However, they do not communicate with each other, so she must be the messenger between her hematologist, family doctor, and dermatologist, which is another challenge.

### 2.3.2 In a Blink of an Eye, Gabriella was in Adult Care

As mentioned, Gabriella was consistently an inpatient, so when she turned 18 years old, she was an inpatient in pediatric care. Gabriella explained how she woke up in the Children's Hospital, got to leave the hospital for a bit to celebrate her birthday, but then she got the call that a bed is available for her in the adult hospital. So, she came back, packed her stuff and was sent to the adult hospital. Gabriella expressed how terrifying it was because this was a very quick transition with the limited to no preparation Gabriella received in pediatric care and was given no information or notice before she went out for her birthday about moving over to the adult hospital.

Nevertheless, as Gabriella entered adult care, she began noticing differences between it and her time in pediatric care. She saw how pediatric care was more centered around what the doctor's thought was best for her, but in adult care, it was more centered around her quality of life. So, Gabriella appreciated that the focus now is on the goals she wants to achieve instead of the goals the doctors want to achieve for her. In pediatric care, she butted heads because they were not listening to her, but now in the adult care, they are listening to her. However, even though Gabriella's doctors are listening to her, they do not listen or even communicate with each other. Similar to Lily, Gabriella noticed another difference between pediatric and adult care, which is that in pediatric care all her doctors communicated with each other, but in adult care, none of her doctors communicated with each other. "In the adult world, I do not think any of my like separate doctors have actually like met together about me like, so it seems very disconnected in the adult world a lot more". 75% of the time of her appointments, she spends explaining what her other doctors have said, and the notes that the doctors would write are always accurate or detailed. Overall, if her doctors talked to each other, then everyone would be on the same page.

## 2.4 Analysis

In this section, I will analyze the experiences through the three stages of transition using different theoretical and conceptual frameworks. I will examine how disability shapes kinships using concepts from disability anthropologists Ginsburg and Rapp, the importance of care relationships and caregiving; looking for it with notions from disability theories of care from Eva Kittay, and the way doctors behave using Mattingly's moral laboratory of care.

### Kinship Care and Extended Kinship Care

In anthropology, kinship care in families and extended kinship is interesting because it reflects how societies and networks are organized, how care is organized within them, and the support structures. Disability anthropologists Ginsburg and Rapp made significant contributions to their research about kinship and understanding the forms of kinship within families across societies. In their new book that came out this year, "Disability Worlds", chapter two focused on "New Kinship Imaginaries and their limits". They invented the term *New Kinship Imaginaries* to "signal the potential for a more capacious understanding of the diversity and complexity of how disability shapes familial life, whether with biological kin or chosen families" (Ginsburg and Rapp, 2024, p. 54-55). This term emphasizes "the reality that families are not only flesh-and-blood collaborations but also acts of cultural imagination, encompassing or excluding the fact of disability within family narratives" (Ginsburg and Rapp, 2024, p. 55). PLWRD have their biological kin and form extended kin to create their social support network. The extended kin are friends (parents and PLWRD friends), medical team members, paid workers, etc. New imaginaries also have their limits, which occur from societal expectations, economic limitations, and personal boundaries.

Additionally, Ginsburg and Rapp state that families often feel obliged to create new kinship imaginaries, and families usually find support through schools, community activity groups, and worship spaces (e.g. churches, synagogues, and mosques) (Ginsburg and Rapp, 2024). They feel obliged to accommodate the shifts that occur in their daily

life and the parental expectations of their child's future (Ginsburg and Rapp, 2024). All these changes create disability worlds and transform the way families see themselves.

In Lily's journey through the pediatric stage, she had two doctors who she considered her quarterbacks because of the levels of care and support they gave Lily and her family. They bonded not just with Lily but also with her parents, especially her dad. Also, they never gave up on her, even when the time came for her transition. They wanted to set her up for success in adult care, so they made her referrals to doctors that they thought would create a similar environment as she had in pediatric care. So, these two doctors (the quarterbacks) became a part of Lily's social support network and essentially a part of her false extended kinship. The reason why they will be considered part of her false extended kinship is because in real kinship, people do not disappear or abandon each other when they reach a certain age. Moreover, they were in Vancouver, and Lily, growing up, was living in Saskatchewan, so she had to form a team in Saskatchewan. I was fortunate to have interviewed Lily's mom, Susan, and she voiced how their experience would have been different if they lived in a city that had all the specialists, services and resources all the time. But they were not, so they tried to create one in Saskatchewan. So, even though Lily and her family did not live in a city with all the people and services Lily needed, they did travel to it when they had to. But they knew they had to create a team in Saskatchewan.

As anthropologists, Ginsburg and Rapp turn to kinship to understand what they are learning because it is a foundational concept within anthropology (Ginsburg and Rapp, 2024). However, they argue that the rise of new kinship imaginaries ranges beyond disability because new concepts of kinship are formed in fields such as sexuality, family formation, and medicalized reproduction (Ginsburg and Rapp, 2024).

Other anthropologists observe the concept of kinship differently. Anthropologist Marshall Sahlins, in his overview of "What Kinship Is… and is Not", states, "The specific quality of kinship, I argue, is 'mutuality of being': kinfolk are persons who participate intrinsically in each other's existence; they are members of one another" (Ginsburg and Rapp, 2024, p. 55). Ginsburg and Rapp argue "that the incorporation of disabilities into

family life is part of a contemporary reanimation of the concept, instigating new kinship imaginaries, from family rituals to household economies to challenges to state bureaucracies. Indeed, when you look for them, disabilities are everywhere in new kinship imaginaries, including in the past" (Ginsburg and Rapp, 2024, p. 55).

I agree with their argument because disabilities play a big role in shaping family relationships. Disabilities are becoming more integrated and noticeable within new developments of kinship imaginaries, and this truly indicates that disabilities have always been present in family contexts. Disabilities cause families to rethink their choices when it comes to finances, encounters with governmental systems, and their living situations.

**The Importance of Care**

If there is a key component when discussing PLWRD, it is **care**. Care is significant for PLWRD because they will feel supported, loved, and most importantly, not alone. Social support networks provide care to PLWRD when needed and on their terms. No one will truly understand what the person you are caring for needs more than the individuals themselves. You must be patient and respectful until they tell or show a sign of what they need. In this research, listening to PLWRD, especially during the transition period, is important so they can explain what they need and want. This is why care is a key component and why I will use disability notions of care from Eva Kittay to analyze the importance of care during the transition stages.

Eva Kittay is a philosopher and is known for her work on feminist ethics, disability studies, and ethics of care. Her understanding of philosophy was challenged by her daughter's cognitive and physical disability. Kittay expresses how "disability is in search of an ethics that will both articulate the harms faced by people with disabilities-discrimination that threatens dignity as well as well-being- and offer moral resources for redress" (Kittay, 2011, pg. 49). She learned about disability from her daughter who is non-verbal, but also found "appreciation of care as a practice and an ethic" from her daughter and her caregivers (Kittay, 2011, p. 52). Kittay had to turn away from her own understanding of what a good life is and "a just society" because since she is a philosopher, they see that although children with disabilities lack "intrinsic properties that

philosophers have defined as essential to equal moral standing" they are "no less a mother's child and no less entitled to become a member of our moral community than any other child" (Kittay, 2020, p. xx).

Nonetheless, Kitty's care theory centralizes care in moral life and values giving the right type of care to others, especially to individuals who are not able to care for themselves. Care is observed as "compatible with paternalism", but instead, Kittay insists that it should be "compatible with respect" and that we respect the person who we are caring for (Kittay 2020). Overall, Kittay argues that people do not need to prove that they deserve care and respect, especially individuals with a disability. She stands by her argument because she knows her daughter will never be able to care for herself, yet she still deserves care and respect as any other human being.

This is relevant to this research around PLWRD because a majority of PLWRD do not receive the level of care and respect they deserve in pediatric care or adult care, so many of them go out and look for it. When Gabriella was in pediatric adult care, she butted heads with her doctors because they did not listen to her and focused more on what they thought was better for her. Whereas in adult care, they were more focused on what Gabriella wanted, so they listened to her. While for Lily, when she was in pediatric care, her team listened to her. However, when she was transitioning to adult care, she struggled to find a doctor because the one she was referred to did not give her the chance or even respect her. So, Lily had to go looking for another doctor who would provide her with the level of care and respect she deserves, which she found. Then there are cases like Theo's where his doctors and nurses showed him care and respect by listening and communicating with him all throughout his time in pediatric care, which helped him gain confidence and skills for entering adult care.

Overall, Kittay's theory helped me understand the importance of care and looking for it due to the role it plays when caring for PLWRD. Kittay puts the person and their values first, which is needed when discussing PLWRD. Individuals responsible for caring for PLWRD need to understand the person, listen to them, and respect their needs and choices.

### The Way Doctors Behave

Moral Laboratories by Cheryl Mattingly is created through ethnographic material based on a long-term study of African American families caring for children with disabilities and illnesses. It observes “the moral work of people engaged in trying to create morally good lives for themselves and those they care about” (Mattingly, 2013, p. 310). Parents and doctors fight for whatever they think will result in the best possible life for the child. They want to do the best they can and make decisions that will have the most beneficial outcome. Nonetheless, opinions differ on how to provide a morally good life because the children do not get involved; they must wait while their parents and doctors act on their behalf. From my data, we saw the opposite happen with Gabriella. Doctors treated her like a person and not just a body in adult care because everything was centered around her quality of life and goals. Also, we saw the opposite with Theo. His doctors and nurses in pediatric care would always include him in conversations and ask him questions.

Additionally, Mattingly (2014) notes the type of language and behaviour the doctors use towards patients and parents and the interactions. Most times doctors do not spend time a lot of time interacting with patients and parents, let alone joking with patients. In Chapter 5, Mattingly (2014) saw a doctor taking the time to joke around with his patient to make her feel comfortable and less sacred. After he joked with her, he interacted with her further by explaining in detail what was going on with her health, but in a fun way (Manderson et al., 2016). Similar to Lily’s interaction with her pediatric team when they used the football/quarterback analogy. Overall, the way the doctor behaves with patients and their families impacts them, as you saw through the three experiences and stages of transition.

## 2.5 Conclusion

This chapter has described the three stages of transition- the Before (Pediatric Care), the In-Between, and the After (Adult Care)- to provide the background of the overall transition and what it involves. I analyzed the experiences using theoretical and conceptual frameworks. I used Ginsburg and Rapp’s work on New Kinship Imaginaries to examine how disability shapes kinship (or false kinship). Eva Kittay’s care theory to

explain the importance of care for PLWRD, and Mattingly's moral laboratory care to analyze the way doctors behave. Once the transition happens, everything changes, and it becomes challenging for the PLWRD and the family to readjust, especially if they are not given the same level of welcome, care, and support as they were receiving in pediatric care. I made sure that this chapter was told through three different experiences because my research focuses on the voices that need to be heard who experienced and know what goes on during doctor visits, finding the right doctors, the environment of the hospitals/ clinics, finding/ accessing treatments, and the encounters with medical professionals. But, most importantly, who knows and experienced the transition from pediatric care to adult care.

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When thinking about these transition stages from an anthropological perspective, they are culturally and socially constructed, which is needed to discuss since it impacts the PLWRD and their social support network's experiences in the healthcare system.

The cultural construct is based on cultural beliefs, social roles, and access to resources and services. When thinking about cultural beliefs, it comes to the individual and their family's culture, which determines how they see the disease and influences the actions of care and treatments. For example, some cultures prefer traditional healing practices over modern-day medicine. So, this can play a factor throughout the transition stages because the healthcare system has specific protocols and rules to follow when treating a patient. In my data, this did not come up. Whereas social roles came up frequently. Looking at social roles in a cultural setting about transition stages involves how family members are taking part in the social support networks because, in some cultures, families have a big role in health decisions and planning. As you just read, the transition from stage to stage involves a lot of planning and decisions, especially when not receiving the proper support. In my research, my participants were from different races and ethnicities (Table 5), but they all had similar social roles when it came to health decisions and planning. During the pediatric care stage, the parents took the primary lead in the decision-making and planning, but then they stepped back with the in-between stage, and the adult care

stage arrived, so the PLWRD was able to start to make their own health decisions. Lastly, access to resources and services can impact the transition stages through cultural factors, such as social class, financial status, religion, and language. In my research, my participants and I did not speak about their social classes, financial status or religion because they were not part of the guided questions I had, and they just did not come up in conversations. However, some conversations glossed over finance when the parents were speaking about paying as much as they needed for treatments that would help their child.

Thinking about my family in relation to these cultural factors, we are and were back then part of the medium class level, we had good financial status, we used our religion for support from Allah (God), and language was a barrier for my parents, especially my mom. My dad knew more English than my mom when my sister was beginning her journey, whereas my mom was still learning it. But even though my dad knew English, all the medical terms and information was hard for them to understand. They were never given translators, but we were grateful to have great family friends who went with my parents to translate specific things.

The social construct is based on social beliefs, access to resources and services, and healthcare institutions. Social beliefs shape the way society sees disease, which can influence the way PLWRD are seen and treated, impacts the way PLWRD and their social support networks seek care, and how they will go through the transition stages. In society, rare diseases are not well-known, and are all seen as only “rare”, that is it, but there is way more than the disease being rare. Even doctors do not know everything or anything about rare diseases. This has a big impact on PLWRD and their social support networks throughout each of the transition stages, which was highlighted throughout this chapter and the rest of the chapters. Like how cultural factors can impact access to resources and services, there are socioeconomic factors that impact it. Those factors are social status, education, and income, which can affect the transition. In my data, even though I did not document my participant’s education levels, based on how much research they did and knowledge they have about rare diseases and how the system works, all benefited the transition process. The parents who educated themselves the minute their child got diagnosed and even before they received an official diagnosis

helped them know what their child needed from the doctor while in pediatric care. The PLWRD begin to educate themselves throughout the stages, but mainly when they get to the in-between stage, where they prepare for the next stage. By the time they get to adult care, they know what they need, what types of doctors, medicine, etc. In general, if the PLWRD and their social support network members do not educate themselves about the rare disease, what it involves, etc., then no one else would. Then there are the healthcare institutions, such as hospitals and clinics. Based on how they are structured and what services they provide and do not provide can affect the transition stages. For example, Lily had to travel to BC for care because Saskatchewan did not have all the healthcare services and doctors she needed. Overall, the three transition stages are influenced by cultural and social factors because they shape the PLWRD and their social support network's experiences throughout the healthcare system and stages.

**Table 5: Demographic Information**

| **PARTICIPANT** | **RACE** | **ETHNICITY** |
|---|---|---|
| 1 | Caucasian / White | Eastern European |
| 2 | Caucasian / White | Scottish/ Canadian |
| 3 | Middle Eastern/ Arab | Egyptian |
| 4 | Middle Eastern/ Arab | Egyptian |
| 5 | Caucasian / White | Canadian |
| 6 | Caucasian / White | Canadian |
| 7 | Middle Eastern/ Arab | Lebanese |
| 8 | Middle Eastern/ Arab | Lebanese |
| 9 | Middle Eastern/ Arab | Lebanese |
| 10 | Caucasian / White | Irish/Canadian |

| | | |
|---|---|---|
| 11 | Caucasian / White | Canadian |
| 12 | Caucasian / White | Greek |
| 13 | Eurasian | Bi- racial |
| 14 | Caucasian / White | Canadian/ European |

# Chapter 3

# 3 The Support Dynamics

*"When dealing with a rare disease, solidarity networks are more important than ever, they help keep those suffering from feeling alone and promote the sharing of experiences and advice" - Pope Francis*[8]

Social support networks are essential in daily life because everyone needs people around them that they can count on. Social support networks are made up of people who share strong, special bonds with and who can help you take your focus off the problem for a little while with emotional, material, or cognitive resources (Salvador et al. 2015, p. 667). In some cases, PLWRD has different networks around them, such as families, friends, schools, and hospitals. Their primary network is considered their family, and their secondary network is their institutional network (De Morais, 2018).

In this chapter, I explore the different support dynamics and roles of the parents (mothers and fathers), siblings, and friends. Also, I explore changes in power and control dynamics after the PLWRD transition to the adult care health system. I will highlight examples from the interviews to showcase the different roles and supports my participants, and the people they spoke about have done. The theoretical and conceptual lenses that will be used in this section are care theory (Kittay, 2011; Mattingly, 2014; McKearney and Amrith, 2021) and illness as socially constituted using theories from medical anthropology and disability anthropology, and disability (Ginsburg and Rapp, 2013; Ginsburg and Rapp, 2024; Manderson et al., 2016).

I will first describe the role of mothers and fathers because each parent has a specific role. Both are there supporting their child living with a rare disease, but it is known that mothers are the primary caregivers. All the mothers I interviewed have provided so much support for their children, while they were in pediatric care, and after they transitioned to

[8] (Watkins, 2021)

adult care even though their adolescent children took on the responsibility themselves. Also, I was fortunate to have interviewed a father, and I heard about the other fathers from either the mothers or the PLWRD during the interviews. Their role is equally important as the mothers' because they have their own set of responsibilities. In this section, my research will shed light on how fathers play an important role when caring for their children since fathers' voices tend to be absent in literature on parenting children with disabilities (Green et al., 2011).

Then I go into the role of siblings. I did not interview any siblings, but I did ask questions about them because they play an important part in the PLWRD life. *Speaking as a sibling, I feel like being there for our sibling is the most important thing, such as being there through treatments, appointments, surgeries, recovery, listening to them, talking things out, etc. At the end of the day, we are siblings we stick together.*

Next, I go into the role of friends. From the interviews, I saw two sides of school friends (supportive vs non-supportive), making friends at the hospital, and the help of family friends. Finally, I discuss the change in power and control dynamics after the transition to adult care. Essentially, parents stay as part of the PLWRD network, but they do not have the control and power anymore over their child's health decisions, treatments, going to appointments, etc.

## 3.1 The Roles of Social Support Networks

**Parents**

Once a child is diagnosed with a rare disease, their lifestyle begins to change, meaning the whole family's as well. Also, it means the child will need a strong social support network of people around them while undergoing appointments, treatments, and regularly. Parents are the primary support children have throughout their medical journey, especially when the child has a rare medical condition that involves additional time, care, and effort from the parents.

Every child's social support network, especially the parents, want the PLWRD to have the best treatment so they can live a long and good life. However, health providers and

parents are talking directly to each other and excluding the child from the discussion. Yet, this is still a common situation reported in many studies that the child's perspective gets overlooked. In Silva et al. (2017) study, their objective was to identify the key networks and social supports from the perspective of a child with a chronic disease. The results showed two themes: 1) strong social support networks and 2) weakened social support networks (Silva et al., 2017). In the first theme, they saw children having effective strong social support networks who know their medical condition and treatments (Silva et al., 2017). The second theme they saw children having weakened social support networks meaning they do not receive the proper support and care that they should (Silva et al., 2017).

In Mattingly (2014) moral laboratories work, two issues she highlights about parents are that they essentially become almost as remote as doctors based on how they know more information and how they lose their role as the parent and begin treating their child as the patient or the disease itself. This can impact their relationship with their child because their child already has so many doctors, but the biggest thing that PLWRD need is for their parents to be their parents. They need that comfort, support, and love instead of being observed and monitored 24/7. Mattingly (2014) states about one of the mothers, "Most painfully, she herself sometimes "forgets" that her daughter is her child and not her patient. She loses sight of how to "just be a mother"" (p. 111). Unfortunately, this is the result for many mothers (and fathers) because they are too focused on finding a treatment to cure their sick child.

In my research, I saw the complete opposite. The parents kept their parent role intact and would fall under Silva et al.'s (2017) first theme, a strong social support network. Yes, all of them have done their research and gained a great amount of knowledge of their child's rare disease, which for some, made them more knowledgeable than the doctors, but that did not make them treat their children negatively or see them as their patients. Due to the limited and lack of knowledge of rare diseases and treatments, parents are essentially left to manage the unmet needs and navigate the care on their own (Currie and Szabo, 2019). Some parents even take it as far as going or looking overseas for necessary support for their child (Pelentsov et al., 2015). Three of my participants mentioned that they thought

of it, and one went through with it by receiving information about their child's rare disease from another country. Mothers are known as the primary caregivers who are responsible for doing almost everything for their children. Fathers, on the other hand, tend to be overlooked since they do not do as much as the mothers do, and in literature, the father's voices are absent (Green et al., 2011). Both roles play a part in the result of their child's and family's "morally good life" because they fight for what they think is best for them (Mattingly, 2014). I will highlight a few of the mothers and fathers from my study to show the roles and support they provided for their children, especially showing how fathers are as involved in their children's care as mothers.

*Mothers*

"Oh, sorry, I get a bit carried away when I talk about his condition when someone wants to listen" Amelia expresses after she introduced herself, her son (who was not eligible to participate but asked if he could sit in during the interview, which he did) and went into his medical journey. I did not mind when she or any of my other participants got carried away talking about their or their child's rare disease and medical journey because that is what I want from them. I want them to feel comfortable and safe in the interviews, where they can speak as much as they like and for as long as they like. Amelia shared so much of her role as Logan's mother and the support she provides him, and I would share everything if I could, but I will be showcasing one of the many things that stood out to me from what she has shared. Logan was diagnosed at 10- years- old with Immune Thrombocytopenic Purpura (ITP), a rare blood disorder where the immune system has gone wrong, and his body begins destroying the platelets in his blood. Currently, Logan is 17 years old, meaning he is in his last year of high school and pediatric care. We got into a conversation about Logan's plan for school since he wants to go to a university about 5 hours away from home. I asked what their plan would be if he did go to that university, are they going to think ahead about the hospitals or clinics around the university for his care, etc. Amelia stated,

> "We've always tried to not let it [his medical condition] rule his life, so I do not think we'll start to put it is a priority, but it would not be the forefront of a

> decision. Okay this is where you want to go like maybe you've got A, B and C that you want to go to. Let's go and see what medical supports there are at A, B, C or in the area which will then help with final decisions. I would say would probably be how we would do it."

Ensuring that Logan's rare disease not get in the way of him going to school where he wants truly shows how she puts his wants and needs first.

Another mother I interviewed was Julia. Julia has been preparing her daughter[9] since she was young because a doctor explained to Julia that Ivey's first transition is as early as age 3 or 4 when she is in pre-school or beginning school. There is no framework for this, so Julia built her own with long- and short-term goals in terms of her ability to talk about herself to her doctors, to do her medical management independently, etc. With all the work Julia has done throughout the years, she continuously has been preparing Ivey, and she became independent with doing her own medical care by the age of 5. Also, Julia has created a shared document of Ivey's medical history, supplies, management plans, and OR plans, so Ivey can always have it on hand whenever she needs it, must change or add anything, etc. Lastly, Julia has set boundaries and is respectful towards Ivey's privacy and story. She expresses how there is a switch between her story (caregiver story) and Ivey's story. It starts as the caregiver's story, but then around the age of 9 or 10, it is Ivey's story. So, it becomes very important to respect her privacy and be careful to distinguish the difference between what is Julia's caregiver story versus Ivey's patient story because it is not Julia's story to tell it's Ivey's medical story.

Next is Peyton. Her daughter Rose has Joubert Syndrome, a rare genetic condition characterized by abnormal brain development. She is missing a part of her brain that gets all the information in and out of the cerebellum. They struggled a lot to get a diagnosis because of how rare her condition is. But it was not until Rose was three and a half years old that she received a diagnosis. However, it then took another couple of years until they connected with specialists who were not in Canada, but in the US. Also, they got local

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[9] I will not be explaining Ivey's condition because they want it to be disclosed.

doctors (pediatrician, pediatric ophthalmology, etc.) for everything they needed while getting information from international researchers that Peyton would then bring back to the medical team. Throughout Rose's time in pediatric care, Peyton worked hard to find doctors, get information that would help Rose's doctors, find workers to assist her and her husband with Rose, take Rose to appointments, regulate Rose's emotions, etc.

Another thing Peyton does is keep her daughter in the loop when at a doctor's appointment, where she takes the lead in talking to them. Rose has a developmental disability, but she does comprehend and rationalize with help. So, Peyton and Rose talk through things together; Rose has her signing authority for all her legal and medical things, so Rose is at the level of personal care and advocacy. But people usually talk to Peyton about Rose in front of Rose, including doctors. Peyton acknowledges that she does take the lead and charge when they are speaking with doctors,

> "They do talk directly to me. However, to be fair I'm more forward than a lot of people. So, I may, I might not allow that doctor, patient connection as much as I could. I tend to think and talk quickly, and so, and I'm talking at the level that I'm trying to connect with the doctors with, and that's not who Rose is. Rose uses different information, like different words and different rational paths. So, I kind of cut her off, and I just talk to the doctor, and then I'll say to her so what we're talking about is".

But Peyton always keeps Rose in the loop; she does not isolate her from the conversation and lets her know what's being said and happening.

Overall, all three of these mothers go above and beyond for their children by putting their children's needs and wants first by caring and showing them respect. It relates to Kittay's argument that people with disabilities do not need to prove that they deserve care and respect. As Kittay states, "One does not need to demonstrate autonomy to command respect" (Kittay, 2020, p. 211). She learned that from her daughter because her daughter is not autonomous, but that does not mean Kittay will not respect her wishes and needs. I believe that Kittay's care theory plays a part in the mother's role in caring for their children with rare diseases because they put the child and their values first.

*Fathers*

I wished to have more fathers participate in my study, but I am grateful that I interviewed one and heard about the others from the mothers and PLWRD. Ginsburg and Rapp (2024) state, “Men in heterosexual relationships often deferred to their wives and partners as the primary caregivers for their children, from feeding and dressing to working the bureaucratic structures in pursuit of appropriate services. Nonetheless, some fathers have adapted their work lives significantly, founding small independent businesses relevant to their child’s disability; they often adjust work schedules to be more flexibly available, given the intensity and relentlessness that often characterize required care. Others take on leadership for particular conditions” (p.70). In my research, each father had different roles and supports that they took responsibility for, which was interesting to learn since fathers are known to not do much.

Sebastian, the father I interviewed, has been involved in Lucas’s medical journey. Lucas was born with Nager syndrome, which is a rare disease that affects the development of the face, hands, and arms. Lucas has an underdevelopment of the outer ear that left him with conductive hearing loss, and he has an underdevelopment of his hands. His thumb on his right hand was non-functional, so he had surgery to remove it and replace it with his pointer finger. Sebastian went to doctor appointments out of town, made sure they had a good community, joined a research study with Tori and Lucas, supported Lucas academically, and pushed him toward his goals. Sebastian even moved his job closer to home. He began teaching at the school near his house, which saved him a lot of time when Lucas had appointments after school because he would not have to deal with traffic getting home. His job made things flexible when scheduling appointments, which is not the case for certain jobs. So, when it was March break, Christmas holidays, and summer break, those were usually the times when they would book Lucas’ appointments. Also, Sebastian had care days that he could use to take Lucas to appointments. But Sebastian does acknowledge his wife, “Tori was at home with Lucas, actually with both boys. So, she did a lot of research.… Having Tori at home took the pressure off, and then with my career, I had a lot of leg room to make those appointments”.

Next is Susan's husband/ Lily's father, Greg. Where Tori was the one who took the primary lead and did the research. Susan told me about how Greg was the one between them who was the researcher, took the lead, and at doctor appointments, he would try and find out what things were by talking to the doctors and surgeons.

Then there is Julia's husband, Ben. When Julia was talking about Ben, she mentioned how intelligent he is, even though he does not know all of Ivey's doctors because it is just too much, and that you need only one person being the "project manager". She continues to explain how he is very involved. He sets up and cleans up all of Ivey's medical management, all the health insurance claims, and drives them to appointments. Most importantly, him and Julia are a team who has different strategies to achieve goals sometimes what is needed is passion while other times, what is needed in calm. Also, like Sebastian, he is a teacher, so he has a lot of time in the summer and understands the education system well due to working in it, which helped them be successful with Ivey's IEP.

Overall, each father is/ were very involved in their child's life in different and similar ways. They all care for their child, they use the skills they have and do everything they can to help, change things in their life to align with their child's treatments, and just be there who their child and family. As one of the fathers in Mattingly's (2014) work states, "You have to 'step up to the plate' to raise a very sick child, and this can prove an almost impossible feat" about how "the work of care demands the work of cultivating virtues to be, for example, a 'good enough' parent" (p.5).

**Siblings**

Just like how the parents' lives change once their child gets diagnosed with a rare disease, the sibling's lives do as well because having someone in the family with a rare disease and a disability causes challenges and concerns to everyone (AlexionAstraZeneca Rare Disease, 2022; Meltzer, 2019). A siblings relationship is known to be the longest relationship a person has in their lifetime (Meltzer and Kramer, 2016). Ginsburg and Rapp (2024) explain how parents today are more aware of the "complex ties binding disabled and nondisabled siblings" (p. 65). Also, a significant concern for parents is the

fear of neglecting their other children (Pelentsov et al., 2015). All my participants who spoke about their other children expressed how they did not involve their other children with everything that was going on when they were younger to protect them, how they did not treat them differently than their child with a rare disease, or how helpful their other children are at a young age and when they are older. Even though siblings are not the primary caregivers, they "consciously or not, assume the role of caregiver in some form, be it entertaining and caring for their sibling, helping with their medication or calming them down after a seizure. These young people take on significant responsibility, and many parents rely on them to do so" (Alexion AstraZeneca Rare Disease, 2022, p. 8). Thus, they would be defined as young carers (Brolin et al., 2024; Iacobucci et al., 2022). The following are four examples from my study that show the different roles and supports that sibling gave to their sibling living with a rare disease.

Firstly, Tori and Sebastian's younger son, who they refer to as Lucas' "sidekick". They gave him every opportunity they offered to Lucas, such as enrolling him in every sport that Lucas was in, and they even did activities together like karate and swimming. Lucas and his brother had the typical sibling relationship. Next is Peyton's younger daughter, like Lucas and his brother, who had a typical sibling relationship. Peyton explained they did not demand much from her when she was young, but as she is older now and has a degree in neuroscience and biology, she's been a good help to Peyton by helping her process everything that has happened.

Then there is Amelia's younger son. Amelia and her husband protected their younger son from information about Logan because they did not want him to worry about his brother. However, he knew things had changed with Logan because Logan could not participate in things they used to do together, such as using the trampoline in the backyard that they loved doing together. So, it was hard for her younger son to understand, but as he got older, they began sharing more information with him, and now he is involved with everything. He waits to know what the bloodwork results are every time, and when the results are not good, Amelia tells her younger son before she tells Logan because she would tell him that Logan would need extra support when she tells him the results, and he will be there for Logan. But at the end of the day, they are brothers, and Amelia's

younger son would get frustrated with some of the restrictions around Logan, such as when Logan is tired after school, he will need a nap. Amelia would then ask her younger son to help her, and he would question why Logan was not helping. Amelia would explain why Logan is not able to do much and, even though her younger son understands why, he resents it at the same time. However, her younger son would be the first person there to support Logan when he needs it, and they are brothers, so they have a typical love-hate relationship.

Lastly, Harper's older daughter supports Theo in different ways. They would meet in the kitchen at midnight for a snack and have a conversation with each other. While Theo was doing dialysis, Harper's daughter's job was to distract Theo because he would be awake for a few hours connected to the machine, so she would hang out with him, play video games, and chat. When Theo got his transplant, it was during COVID-19, so it made things hard because Harper's daughter could not come to the hospital and visit. But she took on the responsibility of caring for her father while he recovered after he donated one of his kidneys to Theo. Harper's daughter was there to help whenever she could, but it was very hard on her.

Overall, siblings are an important part of a PLWRD social support network. The roles and supports vary depending on the family because in cases like Harper's family, they included their daughter from the start of Theo's journey. Whereas in Amelia's family, they did not tell their younger son about Logan's rare disease until he got older. Siblings of PLWRD are "unique individuals with extra challenges, responsibilities and limitations in their lives" (Alexion AstraZeneca Rare Disease, 2022, p. 4). They often get overlooked because when it seems everything is about their sibling with a rare disease, they feel isolated and confused (Alexion AstraZeneca Rare Disease, 2022; Ginsburg and Rapp, 2024). Also, research suggests growing up with disabled siblings can infuse the non-disabled sibling with responsibility, patience, and compassion for others (Ginsburg and Rapp, 2024). They may feel inspired to go into a "helping profession", such as medicine or teaching, and some non-disabled siblings appreciate their disabled sibling because they gain an understanding of "the wide spectrum of human difference" (p. 65-66). I do wish that I had the opportunity to interview the siblings because, even though the parents did

tell me about them, they have their own unique perspectives of their role; they are the only ones who can truly express how they feel and explain their experiences growing up with their sibling with a rare disease and how it might have changed once they became older.

**Friends**

Friends can be a positive or negative impact on a person's life. When thinking of a PLWRD social support network, the primary people would be their family members. However, De Morais et al. (2018) state, "Friends are combined with family members in primary networks. Secondary networks are primarily characterized by relationships that are not chosen" (p.7). I planned to interview friends because I knew they would have a role in the social support network of PLWRD. Unfortunately, I did not interview any, but I was told about them.

Friends would be considered relationships outside of rare diseases context due to a lack of knowledge about the rare diseases they have. Generally, people with disabilities suffer discrimination in different domains, such as in the workforce and education (Kittay, 2011; Carey et al., 2020). It even occurs in friendships because the person with a disability feels excluded from the group. From all the PLWRD I interviewed, two participants shared their experiences with their school friends who were not supportive and made them feel excluded. Another participant shared his experience of having a good friend who stayed by his side and supported him.

Gabriella spent almost all her time as an inpatient in pediatric care, which led her to become best friends with her roommates in the hospital who were there for different reasons than Gabriella, the same age as her and spent a long time as an inpatient. As they began getting closer, a child life specialist split them apart because she explained to them that becoming close was not good for their health. They would just want to stay in the hospital. Gabriella became closer to the friends she made in the hospital, with who she is still best friends to this day, rather than her school friends. Since Gabriella was always in the hospital, which was an hour away from her home, she did not see her school friends much. On the days when she returned home and went to school, she would feel left out

because she had missed everything that happened when she was gone, and that was hard for her. So, that is why she got closer to the friends she made in the hospital.

When Lily was in school, there were problems with friends because of the lack of understanding about Lily's rare disease. Lily explains how there were attempts at bullying her about her rare disease because it was something they though was her weakness, but it was not. She expresses how CMTC was never something that people could use to mock her or make her cry because she felt like it was something she owed, and if someone tried to tease her or anything, she would give them an hour-long lesson about her rare disease. It continued as Lily got to high school because her friends still did not understand her rare disease. For example, Lily was allowed to miss class without parental permission to go to the doctor, because her parents would forget to call in. Her friends would confront her about how she is the only person that gets to do that, and it is not cool. It shows how they do not understand and care what Lily has or is going through. Instead, they were jealous about the accommodations she has.

Compared to Gabriella and Lily's experience with school friends, Asher had the opposite experience. Asher was diagnosed with Synovial Sarcoma, a rare type of cancer (he had a soft tissue tumour in his neck), during his last year of high school. While he was going through receiving the diagnosis, treatments, and recovery, Asher had a good social circle of friends who supported him and are still friends with him today. His friends would always visit and hang out with him. After Asher's surgery, he could not speak for a few days, but his friends still came over and hung out with him. They had made several touching memories standing by each other's side since they all cried together and provided comfort to one another because they saw how much Asher was struggling.

Additionally, Ginsburg and Rapp's (2024) work on kinship speaks about how parents turn to in desperate times when familial caregiving arrangements begin to fail. They explain, "Parents may turn to extended family members, friends, and social service supports in search of alternative solutions" (p. 61). However, during their fieldwork, they frequently heard the following comment, "I feel terrible saying this, but I hope [my adult child] dies before I do. Nobody else will care for them the way I do" which addressed the sense of

“irresoluble anxiety” parents have (Ginsburg and Rapp, 2024, p. 61). Of all the parents I interviewed, only one mentioned a person other than family members they would want their child to go to for help.

Since Logan was not eligible to participate, Amelia had a conversation with Logan before the interview, discussing different topics, and Amelia even asked him questions to know his answers. One of her questions to him was “who you would you say your support system is?” Logan said his parents, brother, and Amelia’s best friend. Amelia explains,

> “My best friend is probably the other person in the world other than the medical professionals, that knows more about his condition than most people because she's taking it upon herself to support me and look things up herself. So, when I'm worried about Logan, I know I can talk to her, and she gets it because she's done the research and Logan knows that as well. If I was not around, she's the best person for him to call, and say, help me!”.

So, Amelia trusts her best friend to be the person her son goes to for support and help because she took the time to research Logan’s rare disease and understand it all on her own. That shows how much her best friend cares for Amelia and Logan. This is an example of Ginsburg and Rapp’s kinship imaginaries because they highlight that kinship imaginaries are not just defined by biological ties; it also includes chosen family members. The support and care that Amelia’s best friend offers is the same type of support and care biological family members would offer them from the start of Logan’s diagnosis to now. Especially since Logan himself chose her first, shows how close she is to him and the family, and how valuable she is.

Furthermore, friends can be considered kinship imaginaries since they recognize how people support one another and form strong bonds and relationships. As mentioned above, there are chosen families involved in kinship imaginaries, but there are also support/care networks involved. Meaning non-biological relationships, such as friends. reshape the views support and kinship. Another example of Ginsburg and Rapp’s kinship imaginaries is Asher’s friends, who have stayed by his side until today. They were there with him like his family was, such as while he was getting his diagnosis, going through

treatments and recovery, visiting and hanging out with him even when he couldn't talk. They had emotional moments together while seeing Asher was struggling, and they stuck by his side.

Overall, school friends can either be supportive or non-supportive, long stays in the hospital can lead to forming long-lasting friendships, and even friends of parents are included in the PLWRD social support network because they provide support and care to the PLWRD as parents do.

## 3.2 Changes in Power and Control Dynamics

One of my parent participants expressed her thoughts on the power and control dynamics,

> "And the transition in general was hard for me because I've been like the one in control the whole time, having you know the questions and keeping track of everything and then to be put in that position where literally overnight no longer that person".

The transition of power from parents to youth involves navigating complex power dynamics within healthcare settings. Developing assertiveness skills and building confidence in expressing their needs are essential aspects of navigating these power dynamics effectively. Generally, being actively involved in the decision-making processes regarding their health fosters a sense of autonomy and self-determination, empowering them to take ownership of their medical journey.

Many children grow up living with a rare disease, which is challenging because some of those diseases are life-threatening and/ or not curable. It is a stressful time for not only the children but also the parents because they are the caregivers of their children. Being a caregiver is difficult, mainly when it is your children that you must see suffer. Some parents overstep and take full control over their child's life without discussing or getting the child's input on decisions. According to Manderson et al. (2016), "These acts take their toll on caregivers, and result in frustration for those receiving care, for whom the loss of independence, often tempered with confusion, can be deeply distressing" (164).

Some parents do not understand that children need to be involved and included in the decisions about their health.

“Even when children are subject both to parental and clinical control, they are not invisible” (Manderson et al. 2016, 20). Manderson et al. (2016) discuss how children are powerless when they are sick, so they depend on their parents, which leads to the parents and their child’s lives getting “entwined”. It became a norm for parents to speak for their children about how they feel when talking with the medical profession. However, the parents cannot know what the child is physically or emotionally feeling. Additionally, Larotonda (2016) explains how the disease becomes “incarnated- personified and tangible- on the child, whose physical appearance can be marked by particular phenotypic traits or malformations. This influences the child’s self-perception and how he or she is considered by others, so shaping individual and social experiences” (27). Once the child starts to look at themselves as the disease, it will affect them personally.

Children are not the only ones who might look at themselves as the disease because parents might too. Parents make sure they are doing everything they can to support their children. However, that sometimes becomes too much, which results in the parents seeing their child as the disease instead of their child (Mattingly, 2014). During the child’s time dealing with having a rare condition, they feel powerless and vulnerable, so they depend on their parents to help them (Manderson et al. 2016). It then leads to the parents speaking for and representing their child all the time, which becomes a problem because sometimes it is beneficial for the child to do the talking, especially since it is their health, and they know what it feels like at that moment (Manderson et al. 2016).

Parents overstep the “parental power” they have with expressing what the child is experiencing, such as symptoms, which most of the time they are not certain about (Manderson et al. 2016). At the end of the day, the parents want the best for their child, but boundaries should be set. Children need to feel safe and comfortable speaking about their feelings and know what is happening. Especially as they start to age, they are expected to take on more responsibility for their health (Manderson et al. 2016). PLWRD will be dealing with their disease all their lives, so it is important to know what is

happening with their health, be able to speak with their medical team and understand the next steps.

In this final section, I will showcase how the power and control dynamics move from the parent to the PLWRD possession once they transition into adult care and become more independent. Parents who once had control and power over their children's health decisions stay a part of their networks but are not as involved in their lives as they used to be.

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Lily's main social support network was her parents before and after transitioning to adult care. Susan shared that when Lily turned 18, she started to age out of everything, and she left Saskatchewan for Vancouver to go to school, so the dynamics changed because Susan and Greg were still living in Saskatchewan. Lily needed to do things on her own since she became an adult, such as trying to navigate adult care by finding new doctors. Next is Gabriella and Lucas. Once they transitioned to adult care, they wanted their independence in their health care, so their parents had to step back. But their mothers still wanted to be updated on what happens at appointments, results, etc. Tori expressed how she even saw the shift of control get passed to Lucas during his appointment when the doctor did not even acknowledge her and stayed talking to Lucas only. So, not only is the transition to adult care hard on the parents, but also the loss of their responsibilities and control of their child's life, which most of them have been dealing with for 18 years.

Then there is Rose Peyton's daughter. Rose's main support network was and still is Peyton and her husband, but once Peyton moved into a group home, they lost control and power over her health care because now Rose has a team at the group home who oversees her care. However, Peyton and her husband are still on call and get to know what is happening. Rose moving into the group home was another transition after he transitioned from pediatric care. Once Rose transitioned out of pediatric care, all her connections with her doctors disappeared, so Peyton had to find Rose all new doctors and educate them on Rose's health condition, such as what had happened in the past and what they needed to do to help her. When Rose moved into the group home, all her doctors changed again

because the group home was not in the same city they lived in and where the doctors had been lcoated. So, all of Rose’s medical care has transferred to somebody new where the group home is. Peyton expressed “So that's been hard for me, 'cause I have to let go”.

As Peyton expressed how hard it was to let go, it was the same for the others. These parents have supported and cared for their children for their whole life. They scheduled appointments, treatments, and surgeries, made sure they got the right medication, did research about their rare disease, made sure they were seen by the best doctors, and the list goes on. Then suddenly, all their responsibilities vanish the minute their child turns 18 years old or moves to a group home. Now, they need to let go and allow their children and group home team to be in control.

## 3.3 Conclusion

This chapter looked at the roles of mothers, fathers, siblings, and friends and the change of control and power dynamics through the theoretical and conceptual lenses of care, medical anthropology, and disability theories. Also, when discussing each role, I shared several of my participant's experiences to allow the reader to understand what their role involves. Each member of the social support network has a specific role and support they provide to the PLWRD. When PLWRD get to the age when they are ready to take their health care into their own hands, their social support network, especially the parents must also be ready to let go and allow them to take charge.

# Chapter 4

# 4 The Challenges and The Supports

In this chapter, I discuss the supports and challenges PLWRD and their social support networks face in pediatric care, continue to face in adult care, and any new challenges and supports that emerge after the transition to adult care. I will analyze the experiences of my participants using the theoretical and conceptual frameworks of care, medical anthropology and disability theories, and advocacy support (Black, 2018; Ginsburg and Rapp, 2013; Ginsburg and Rapp, 2024; Kittay 2002; Kittay, 2019; Manderson et al., 2016; Mattingly, 2014; McKearney and Amrith, 2021; Rossetti et al., 2021; Rutherford, 2020).

I begin discussing the challenges that the PLWRD and their social support networks have gone through, are still going through in adult care, and are currently going through. The one challenge I specifically go into is the impact of the COVID-19 pandemic. Then, I discuss the support PLWRD, and their social support networks have experienced in pediatric care and after transitioning into adult care. The one support I specifically go into is the extraordinary measures parents take. In the final section, I will discuss the encounters with health professionals because it has both challenges and supports.

## 4.1 Challenges

PLWRD and their social support network face several challenges throughout their time in pediatric care and the transition to adult care. The challenges range from lack of knowledge from health professionals, insufficient support, unsupportive individuals around them (e.g. doctors, friends, teachers, coaches), and lack of services and resources in their hometown meaning families need to travel to different cities, provinces, or countries. Families struggle to find doctors and specialists, lack transitional support (no help or preparation), lose pediatric team, get to adult care and do not understand how things work there (e.g. in adult care, PLWRD see doctors every six months, whereas, in pediatric care, doctors see their patients in much shorter periods) and PLWRD feeling like they are missing out on things in school and with friends.

Most of these challenges have not only been identified in my research but also in past

literature. For example, Currie and Szabo (2019) have conducted two studies on the parent's perspectives on taking care of their children living with a rare disease, and they have identified several challenges: parents having more knowledge about their child's condition than the doctor; there is no proper structure or organization in the healthcare system (parents must repeat their child's health history at doctor appointments); parents struggle to access and receive government supports; parents take on different roles due to lack of healthcare services; parents being silenced and feeling disconnected.

The challenge of parents having more knowledge about their children's condition than the doctor leads to the parents having to teach the doctors. Two parents from their study stated, "I feel they are learning as I teach them [referring to the medical team]" and "The textbooks they've read are probably 15 years old based on a handful of studies. There is so much more information and research now" (Currie and Szabo, 2019, p.98). It is fundamentally what has been wrong for years and still is wrong with not only rare diseases but also other medical conditions and even worldwide pandemics (e.g., COVID-19). None of this is new because they have been around for several years, so there is previous information surrounding them that can be used to lead to solutions. Yet, the government and individuals in the health care system are not learning from the past to improve the present, even though there is plenty of stuff that needs improvement. Based on my research, this challenge affects the parents and the PLWRD in pediatric care, especially when they get into adult care. A participant expressed,

"Absolutely! I felt like I knew more than everybody that I dealt with basically. I do not think I've had a doctor since I left peds that knew [about my condition]. No, I do not think I've had a doctor since I left pediatrics that actually".

Another participant voiced,

“I think doctors are always gonna assume that the patients know less, less than them. I remember, there's a funny, tweet about do not assume your two-hour lecture on my condition is, no, there was a doctor says, do not assume that you know more than my 15 years of medical school. And then the response was do not assume that your two-hour lecture on my rare condition equals my lifetime of living it.” (Figure 1).

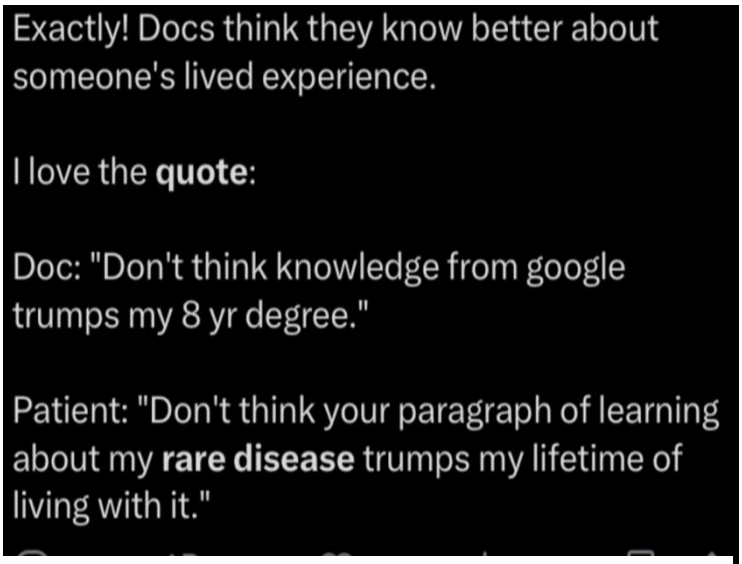


**Figure 1- X (Twitter) Rare Disease Meme**

Moreover, the list above involves challenges that should not be on there, such as the struggle to access and receive the government’s help, the structure and organization of the healthcare system, parents being silenced and feeling disconnected when trying to support their child through a difficult time. The question that I have is, why do families have to keep struggling and fighting when a solution or at least plans for a change can be created? We are responding to similar challenges the same way each year, and nothing is changing, even though we have such different societies now and new types of technology that can further help. Therefore, a change needs to start, especially since we have information and evidence from the past years to help make the change happen.

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Table 6 shows a visual representation of the challenges my participants faced in pediatric care, continued to face in adult care, and new challenges faced. These challenges PLWRD and their social support network face in the healthcare system are linked to social and cultural forces, such as socioeconomic status, cultural beliefs, geographic inequalities, and access to healthcare. In the following section, I will discuss the impact

that the COVID-19 pandemic has had on the PLWRD and the encounters with health professionals.

**Table 6: Pediatric Care and Adult Care Supports bolded ones are the new challenges that occurred**

| **Pediatric Care** | **Adult Care** |
|---|---|
| Lack of knowledge from health professionals | Lack of knowledge from health professionals |
| Insufficient supports | Insufficient supports |
| Unsupportive individuals (e.g. doctors) | Unsupportive individuals |
| Lack of services and resources | Lack of services and resources |
| Struggle to find doctors and specialists | Struggle to find doctors |
| Lack of transitional support and preparation | Lack of knowledge about adult care |
| Lack of knowledge about adult care | PLWRD feeling left out |
| PLWRD feeling left out | Need to travel to other cities, provinces, etc. |
| Loss of pediatric team | Encounter with healthcare professionals |
| Need to travel to other cities, provinces, etc. | COVID-19 |
| Encounter with healthcare professionals | Not being heard by doctors |
| COVID-19 | Online & In- Person Communities |
| Not being heard by doctors | **Struggle to see doctors** |
| Online & In- Person Communities | **Lack of follow ups and monitoring** |
| Missing out on things (e.g. school) | **No communication between doctors** |

### 4.1.1 The Impact of the COVID-19 Pandemic

The COVID-19 pandemic has impacted everyone in the world in several different ways. It had significant repercussions on the healthcare system because it resulted in creating restrictions and re-organizing the system to "concentrate resources needed to the care of COVID-19 patients and to respond in general to this health emergency" (Talarico et al., 2020, p. 1). COVID-19 also had (and still has) big effects on PLWRD. Talarico et al. (2020) stated how "the care of several chronic conditions was in many cases discontinued, and patients and healthcare professionals treating these conditions had to cope with this new scenario. It was the case of the world rare diseases (RDs) that had to face this global emergency despite the vulnerability of people with RDs and the well-known need for high expertise required to treat and manage them" (1).

During the start of the pandemic (April 28, 2020) the Canadian Organization for Rare Disorders (CORD) President and CEO wrote a letter to the ministers of Health and responses about the impact of COVID-19 on rare disease patients (Wong-Rieger, 2020). CORD supported the federal and provincial/territorial government movements to protect Canadians. But they also wanted to raise awareness and attention to the "serious unintended consequence of the pandemic responses, which is now threatening the lives and wellbeing of another cohort of Canadians, those living with chronic conditions, both rare and common. Patients with underlying conditions that compromise the immune, respiratory, and cardiac systems are more susceptible to COVID-19 and its complications" (Wong-Rieger, 2020). Many PLWRD could not access healthcare services, see their doctors, or even have surgeries or treatments done (Talarico et al., 2020).

Each of my participants was affected by COVID-19 in some way; some were affected more than others. One participant had two major events happen to her during COVID, which I analyze in this section because I believe these events highlight the struggles of the healthcare system and society toward PLWRD.

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The COVID-19 pandemic was an eventful time for Lily. When it hit Canada, she was in a different province away from home and an extreme lockdown. Before the city went into lockdown, Lily encountered a difficult situation on the bus. The last week before the lockdown occurred, Lily was bussing back home from work with a sinus infection at that time, which she gets every six weeks, but it was not contagious because it was bacteria that lived inside her that made her sick every six weeks. So, while Lily was on the bus, she was blowing her nose and coughing, and at that moment, it was the first time she thought that she was going to get harmed on public transit because people on the bus with her began to freak out. A man came up to her and started yelling in her face that she was going to kill them all. Lily tried to explain to him that she did not have COVID-19, instead, she had a sinus infection that she gets every six weeks due to her rare disease, but that did not help the man calm down. He was still mad and confused because he did not understand. At the time, COVID-19 was still new and not many people knew about what it truly was, so adding another disease that he did not know about, especially being a rare one, the man panicked even more. So, Lily got off the bus at the next stop coming up and walked the rest of the way home. She expressed how this incident was an eye-opener to her that people were on edge, and if she presented abnormally, she was putting herself at risk.

This incident showed how the understanding and awareness of rare diseases are still lacking in society, but it also showed how people looked at Lily as a disease. She tried to explain to the man that she did not have COVID rather it was due to her rare disease, but that did not change his mind from looking at her without seeing COVID. It is like in Mattingly's (2014) work, where parents and doctors look at PLWRD as a disease, but in their case, the goal is for a better life for the PLWRD. The man on the bus was not thinking about Lily's well-being rather he feared her affecting the others with what she had. So, it comes down to not only parents and health professionals seeing PLWRD as their rare disease, but others do too, but each person's reason for seeing them like that is for different reasons.

Both social and cultural forces are linked to the impact COVID on PLWRD and their experience, and the way society saw PLWRD during it. For example, what happened to Lily was the forces of lack of education and awareness of illness and social isolation.

Additionally, Kittay expresses how "disability is in search of an ethics that will both articulate the harms faced by people with disabilities- discrimination that threatens dignity as well as well-being- and offer moral resources for redress" (Kittay, 2011, pg. 49). The way she learned about disability was from her daughter, which is how most people know as well. Once they learn about disability, they see a whole different world from their perspective. Kittay found an "appreciation of care as a practice and an ethic" from her daughter and caregivers (Kittay, 2011, p. 52). She insists that care is "compatible with respect" and that we respect the person we are caring for (Kittay, 2020). Showing respect to people with disabilities (visible or non-visible) should be extended to everyone in society because it should not be just the people giving the care who need to show and give respect. The man, along with everyone else on the bus, should have shown Lily some respect, especially when she was trying to explain her story. Yes, they were scared due to a global pandemic that everyone barely knew about except that it was killing people, but that did not give them the right to accuse Lily and make her feel unsafe.

The second event that occurred to Lily was regarding getting the COVID-19 vaccine. As mentioned above, Lily gets sick every six weeks, but she was not diagnosed with an immunocompromised condition, she was generally considered immunocompromised or immunosuppressed because of her rare disease, and this caused complications when trying to receive the vaccine. Her doctors did an amazing job advocating for her as a PLWRD, especially her family doctor, who contacted the government to explain that they have all the evidence that she will be unduly harmed by this more than others not living with a rare disease. But then the government asked them for proof of Lily's condition. Her family doctor let them know that there was not any since there was no one with her rare disease living in Canada at that time. There were two people in Europe with Lily's rare disease who got brutally sick, and one did pass away, which they used as evidence instead. However, the government did not accept it because it was not evidence-based in

Canada, specifically evidence from the province she was in. Lily's doctor began to get angry with the government and asked them, 'Do you want her to die? And then she'll get the care she needs?' But unfortunately, nothing worked, so she had to wait to receive the vaccine. An interesting thing that Lily found out when she got back home to Saskatchewan was that she was considered a high-risk patient based on her health history there. So, she got the first vaccine, and then when she went to get her second one, they knew about her rare disease based on the file they had of her health history and told her that she needed to get the second one due to her being high risk. Overall, this just showed how care in Canada is provincially motivated.

The other cultural and social forces that appeared in Lily's second event was the lack of healthcare access and government response to the situation on hand, which interconnect. They interconnect since due to the government denying Lily from receiving the COVID-19 vaccine (so, denying healthcare access). During the COVID-19 pandemic showed gaps in our healthcare system has that affects PLWRD in a negative way, such as Lily's experience.

It was a challenging situation for Lily since the government was not accepting any of the information they were providing. However, Lily was not the only one in this fight. She had her doctors fighting and advocating for her. In many cases, parents and PLWRD do all the advocating, such as in the healthcare system and schools. They have developed a great understanding of rare diseases, policies, and the healthcare system to ensure they receive all their needs (Rossetti et al., 2021). Most times they are advocating against doctors due to the lack of knowledge regarding rare diseases, but in this case, the doctor is the one doing the educating and fighting to ensure the PLWRD needs are met. This also shows the amount of care Lily's doctors have for her to help her out while she is in a whole different province.

Overall, the impact of the COVID-19 pandemic was a challenging time for PLWRD in different ways, and it was a reminder of how rare diseases are overlooked and the healthcare system not always being on their side. I would like to end this section with Dr.

Wong-Rieger's closing statement of her letter because it highlights the importance of something that needs to change, especially if a new pandemic occurs:

> "The problems experienced by rare disease patients and families were not directly caused by COVID-19. Sadly, it has taken a pandemic to bring pervasive deficiencies and dysfunctions in our healthcare system to the surface. We urge governments to address these issues now. As a patient community, we are ready and capable of helping generate and implement changes to serve all Canadians … before the next COVID-19 wave or another health crisis" (Wong-Rieger, 2020).

### 4.1.2 Encounters with Health Professionals (Challenge)

One of my participants expressed, "never being able to come into meetings on a level field as a doctor, and it meant that every appointment felt like going into a battleground".

In the case of rare diseases, where many of the diseases need research, they are instead being overlooked and sometimes completely ignored. In Esquivel-Sada and Nguyen's (2018) study, they state that doctors feel "unethical to announce to a patient the diagnosis of a disease for which nothing can be done" (p. 38). How do they know nothing can be done without looking further into what the patient has? How is it right to not tell the patient about the disease? Is it because it is not in their textbooks, so they do not have to do anything about it? There are several treatments in the world that are not being heard and taken into consideration because no one wants to do any extra work. Doctors, especially new ones, are following the textbook and thinking in only one way, and that is it for them. It is because healthcare professionals are trained to listen to certain things; they go by the textbook and forget the most important things. They forgot to listen to their patients and their families and give empathy.

The PLWRD and their families want and need to be heard, especially when they have information that can help. It cannot be just what the doctors want to hear anymore. They should accept the information the parents are providing. Also, people go to doctors for help, meaning they are putting their trust in them. But how are the patients and the parents going to trust the doctors if they are not listening or even showing that they want

to help? This is the social force of lack of knowledge and awareness from health professionals that PLWRD face. PLWRD and their social support networks trust the medical team to help them, but when they have limited knowledge about the diagnosis and rare disease itself, it impacts the PLWRD negatively (physically and mentally). If health professionals take a bit of time to briefly understand their patients' case, then that is at least a step in the right direction.

Nonetheless, this comes down to power, who has it and who does not. Naturally, doctors have higher power, and they will not listen to anyone below them, especially when parents are telling them information about something they have never heard or know of. Eva Kittay's *Ethics of Care* "addresses the obligations and responsibilities that arise within asymmetrical relationships of situation and power between caregivers and those receiving care" (Kittay, 2019, p. 164). So, the carer in the relationship must be attentive, mindful, and responsive to the needs and wants (Kittay, 2019). In this case, the asymmetrical relationship is between the doctor and the family.

In this final section, I will be discussing three challenges of encountering health professionals. The first one is doctors not being prepared for appointments and not communicating with the other doctors on the team. The second one is having to guide the doctor to the answer. Lastly, parents are being silenced and not being allowed to ask questions.

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On the day of a doctor's appointment, you expect the doctor to be ready to talk to you when you see them, but unfortunately, that is not the case for PLWRD. Lily and Gabriella expressed how adult care doctors are not prepared for appointments and do not communicate with the rest of the team as they were in pediatric care. Gabriella mentioned that 75% of her appointments are just explaining to them what the last appointment with the other doctors was about, and the notes from the doctors are not even accurate or detailed. Lily stated how during her appointments, she does not have the time to go over everything since appointments are 15 minutes only because, by the time she finishes the recap, the appointment is done. So, if each team talked with each other, it would be more

beneficial for everyone because everyone would be on the same page, and the appointment would consist of talking about new things, not previous ones. However, non-communication between teams also occurs in pediatric care. Harper expressed that was the biggest challenge for them. Theo has two teams, one for the kidney and another for the liver, but they do not communicate with each other about the different things they want to do for Theo.

Next is having to guide the doctor to the answer. As Peyton describes it as having to play their game. She explains,

> "Make it as if it was their idea. So, I mean, I've said things like, 'you know, I'm not really sure, because I'm not a doctor, but some of the other parents of people with Joubert Syndrome have, said ABC, and I was just wondering what you think about that'. Right? Meanwhile, before the appointment, I have not just talked to parents, I found this was about behavioral medications for Rose when she was about 10, and I had actually found a pharmacist rep to talk to me about what all the different things were out there right now in Ontario, and what the side effects were, and what they also treated, and what the comorbidity was, etc. So, when I went in there I was very, 'I just really need your help' kind of mom".

The same goes for Lily because she shared that she learned fast that the best thing to do was not give them the answer but to lead them to the answer instead. She would be essentially dropping them breadcrumbs along the way to make them start figuring it out, which then would lead them to the conclusion on their own about what the problem was, and Lily would have to act shocked. Since it is a game of directing the doctor to where you need them, this can get quite frustrating at times for the PLWRD and the parents because it is a waste of their time playing this game. This challenge happens in pediatric care and carries into adult care.

Then there is the challenge of parents being silenced, not being allowed to ask questions or give information, and most times doctors completely disrespect the parents. Many parents do not allow themselves or their children to be disrespected, so they simply just leave them. For example, Peyton stated, "Well, the ones who were not open to listening

to me did not stay on the team", and Tori voiced that the deciding factor for her to leave one of Lucas' doctors was when she came one day to Lucas' appointment with a list of questions. She had a paper with her questions, and the doctor saw that she had a list and told her "I'm not answering those questions". Additionally, Tori and Sebastian take Lucas' care to a hospital and environment they feel comfortable in. I asked them what made those other hospitals and environments different than their local hospital, and they stated,

> "I felt like our voice was more like we had more of a stronger voice; I think. I felt like we were well, there's doctors more willing to listen and hear us. We felt more valued. I personally think we felt more valued as individuals and respected and not just like, you know, maybe they see a hundred people like Lucas, but they know that he was our only and they respected that. They gave us the time and whatever. I did not feel like we were just kind of being rushed out any of any of those appointments and I sometimes felt like that here".

Though only the parent participants were the majority who voiced this challenge, there is a possibility that in the adult care world, the PLWRD will face it as well.

All three of these challenges about encounters with health professionals are disappointing because not only do they start in pediatric care and carry out into adult care. However, these challenges have been known in previous literature, so it is a continuous pattern. Especially with rare diseases, it will be a consistent battle until they stop being overlooked. Due to the limited and lacking knowledge about rare diseases, parents are left alone to manage their children's unmet health needs (Currie and Szabo, 2019). Parents end up becoming the experts on their child's condition due to the amount of work they put into researching and understanding it (Currie and Szabo, 2019). Unfortunately, all their hard work does not get used most of the time because doctors would not listen to them, which is unbelievable. If they are being given information about a rare disease, they do not know anything about, why not take the information and learn about it? Most of the time, parents, such as Peyton, are getting their information from international

researchers. So, for parents, it gets frustrating when the health professional lacks knowledge, would not take the information given to them, or does their own.

This relates to Kittay's (2019) ethics of care, "CARE includes respect" (p.210) because parents work hard to care for their children and respect their needs. However, the doctors do not show that same energy towards their patients. Kittay herself learned from her daughter about disability and found the "appreciation of care as a practice and an ethic" (Kittay, 2011, p. 52). She is a philosopher whose understanding was challenged throughout her time learning from her daughter about what a "good life" is and "a just society" (Kitty, 2020). Overall, Kittay makes the argument in her work on care that people do not need to prove they "deserve" care, but from the identified challenges about encounters with health professionals I found in my data, it seems like the parents and the PLWRD must work hard and prove to the doctors that they are worth their time to be seen and heard.

## 4.2 Supports

PLWRD and their social support network face several supports throughout their time in pediatric care and the transition to adult care. The supports range from support social network duties (e.g., taking PLWRD to appointments, giving them mediation, etc.), health service workers (e.g., nurse, hearing internet), extraordinary measures, accommodations in school (e.g., technology support) and hospital (e.g. extra time on exams). Families find support from faith-based groups and online/in-person communities. Parents help with the preparation for the transition to adult care, and there is now program(s) in place to help as well.

When a child is diagnosed with a rare disease, the family's lifestyle changes, but the size of the change, depends on the type of disease. In some cases, the child's disease requires only one parent to be always present, other times requires both parents, and if the child requires specific types of treatments, the family must move to another city or even country. The result of each scenario involves parents having to leave their jobs, homes, and even family members behind to ensure their children are receiving the proper care they need. The parents' lives become scheduled because of reoccurring appointments,

specific medication times, and in certain cases, continuous monitoring (Salvador et al. 2015). This can impact the parent's dynamic in the beginning stages because they are new to the world of being the primary caregiver/ social support.

Table 7 shows a visual representation of the supports my participants faced in pediatric care, continued to face in adult care, and new supports faced. In the following section, I will focus on discussing two supports: the extraordinary measures parents take and the encounters with health professionals.

**Table 7: Pediatric Care and Adult Care Supports bolded ones are the new challenges that occurred**

| **Pediatric Care** | **Adult Care** |
|---|---|
| Social Support Network duties | Social Support Network duties (depends on rare disease, most PLWRD become independent) |
| Health service worker<br>- Nurse<br>- Hearing internet | Health service worker (depends on rare disease)<br>- **Group home workers** |
| Accommodations (in school)<br>- Technology support (e.g. specific technology in classrooms)<br>- IEP | Accommodations (in school and hospital)<br>- **Extra time on exams**<br>- **Extended limits of what an adult is** |
| Faith-based groups | Faith-based groups |
| Services and Resources Available<br>- Mental Health services<br>- Therapist<br>- Government paying for treatments<br>- Parents making helpful resources for their child | Services and Resources Available<br>- **Information resources from Organizations** |

| | |
|---|---|
| Preparation for Transition<br>- Transition programs in hospitals<br>- Parent's help making sure their children are prepared | Preparation for Transition (continues to adult care)<br>- **Transition programs in hospitals** |
| Travelling (meeting other families) | Creating friends (PLWRD) |
| Creating friends (parents and PLWRD) | Extraordinary Measures Parents Take |
| Extraordinary Measures Parents Take | Online & In- Person Communities |
| Online & In- Person Communities | |
| Part of research studies | |

### 4.2.1 Extraordinary Measures

Parents adapting to their new lifestyle can be challenging because they find it hard to balance work and family (Pelentsov et al., 2015), which leads to many parents take on extraordinary measures. Extraordinary measures are "a form of access intimacy and "people as affordance" that involves major life and quality of life changes that they would otherwise not make" (Ries et al., 2024). The biggest, extraordinary measures the parent participants have made were either: 1) using the flexibility of their job, 2) parents leaving their jobs or 3) taking time off from work.

Parents need to learn what works for them in terms of being organized and prepared, putting their full effort and commitment into being there for their children, and figuring out their financial needs (Salvador et al. 2015). They need to think ahead about situations involving them having to move if their hospitals do not have the proper treatment their child needs. In other cases, families stay where they are, but they must organize their daily activities around the child's care. For example, "the strategy I have found to conciliate the care to my daughter and work was getting a job close to home. Thus, I can go home to take a look and find out how she is" (Salvador et al. 2015). If both parents

work, they must make their work schedules opposite from each other (Salvador et al. 2015).

Sebastian is a schoolteacher, and he physically moved closer to his job. His moving closer to the school he taught saved him a lot of time to make it to appointments right after school without getting stuck in traffic. Also, his job has flexibility since he has March break, Christmas and summer off. So, they would book Lucas' appointments around those dates. Also, Sebastian has a lot of discretionary care days off from work that he uses as needed for when Lucas has appointments. Like Sebastian, Greg also was a schoolteacher and Susan worked in a postsecondary college. She explained how they had the comfort of having a fair amount of time to take off like Sebastian had.

Additionally, some parents, whose jobs do not provide the flexibility described above, must completely leave their jobs once their child gets diagnosed, such as Tori and Amelia. Tori decided the number of appointments and procedures Lucas had, it would be better for her to be home with him, but it was life-changing for her to leave the workforce. Similarly, Amelia decided to step back from her job because it was challenging to juggle everything. Amelia expressed, "It made life a lot easier that I was not juggling work as well. So very, very blessed that I have been able to be here to help look after everything and try and just you know, navigate it all together, the ups and downs". Moreover, some parents take time off from work, and for each parent, the result of their time off leads to different outcomes. For example, Peyton, who is also a teacher, would take time off to bring Rose to all her appointments. Peyton wanted to be a principal, but since she took time off for Rose's appointments, she had questionable attendance which led to her losing promotion opportunities. In general, parents would do anything for their children, especially if their child is ill. It can take parents time to come to terms with their child's diagnosis, but as one of the fathers in Ginsburg and Rapp's (2024) work told them, "It's a marathon, the long run. I want to be there for my son when he crosses the finish line" (p. 49).

Ginsburg and Rapp (2024) found that parents from different diverse classes and cultural backgrounds shift their work focus, and become absorbed in the disability worlds, which

got opened by their children. “The idea of disability worlds offers a framework embracing the challenges and world-making that disability brings to social life” (Ginsburg and Rapp, 2024, p. 5). Parents of PLWRD try their best to make their child’s life as great as it can be. Yes, there may be several challenges along the way, but parents will make sure that their child is receiving everything they need, is happy, and well whatever the cost is. Overall, parents adapting to these new ways of living regardless of how big or little it is, can be a lot on the parents.

### 4.2.2 Encounters with Health Professionals (Support)

There are always two sides to things, and when it comes to encounters with health professionals, there are both sides. I have discussed the challenging side in 4.1.2, and in this final section, I will discuss the support side. The support side involves doctors being more attentive and supportive towards the PLWRD and their social support network

In the previous section (4.1.2), I discussed Lily’s challenging encounters with her doctor, and here I will be discussing the supportive side of her encounters. From Chapter 2, you learned how Lily had, such a great pediatric team who were there for her, listened to, and supported her throughout her time. Lily shared how whenever she had an appointment in the clinic, they would have two meetings: a patient meeting and then a family meeting. In the patient meetings, Lily would talk to the doctors about everything, and when it was time for the family meeting, the doctors would ask her what she would not want brought up in the family meeting. They respected her privacy and gave her a voice to say what she did and did not want talked about in front of her parents. This was not the only thing that Lily had control and say over. She was also asked for her consent when she was only 5 years old! Lily’s doctors were good at explaining things to her, especially treatments because they understood that she needed a voice. When Lily was five, she and her parents went to see a dermatologist for laser surgery for Lily’s birthmark on her face. The doctor explained the whole procedure and Lily was not for it. She told them no and hid underneath the doctor’s desk. The doctor saw that she was not comfortable doing it, so he said he would not do it, it is her face, it is her decision, and she has the power of consent. That was that for the conversation on the laser surgery until Lily was comfortable going through with it.

This type of behaviour from a doctor is needed, especially with rare disease cases. The children and the parents need that to know that the doctor helping them cares enough to listen to their stories, questions, concerns, and in Lily's case, their opinions. The interactions between a doctor and their patient are powerful because they will remember and open to more. For example, Amelia shared that Logan mentioned one particular doctor when she [Amelia] asked him who would be in his support system. The reason why Logan mentioned this doctor is because they formed a strong connection. Amelia expressed how the doctor would listen and remember everything, and never make them feel rushed like Logan's other doctors who would not hear them out about the concerns they had. So, this doctor went above and beyond for them.

In chapter five of Mattingly's (2016) book, she highlights the type of language and behaviour the doctor uses towards the patient and the parent and the interaction between the doctors. The type of language and behaviour the doctor uses towards the patient and the parent is important to look out for because most times doctors do not give a lot of time to interact with their patients, let alone joke with them. For example, in chapter five, the doctor, Dr. Kesen, takes the time to joke around with his patient (Besty) to make her feel more comfortable and less scared (Manderson et al., 2016). After he joked with her, he interacted with her further by explaining in detail what was going on with her health, but in a fun way (Manderson et al., 2016). A doctor interacting with the child rather than the parent is rarely seen in studies because doctors do not usually take a lot of time to talk to the patient, especially children (Manderson et al., 2016). Talking to the child directly is needed because they must understand what is happening with their health since they will be the ones taking responsibility for it soon (Manderson et al., 2016). Nonetheless, this type of exchange makes the situation for the child less stressful and scary, but it also puts the parents at ease.

Overall, the type of language and behaviour the doctor uses towards the patient and the parents and the interaction between the doctor and the patient is important to take note of. As you saw in the previous section on challenges, when doctors do not listen to their patients and parents, it affects them. To end this section with one of my participant's advice to the doctors:

"Just listen and believe the patient's experience. The outcomes that matter to the patient, that's what matters. Just listen! Listen to the person in front of you."

## 4.3 Conclusion

This chapter discussed the supports and challenges PLWRD, and their social support networks faced in pediatric care, continued to face in adult care, and what new challenges and support emerged after they transitioned into adult care. The battles parents and the PLWRD go through is a never-ending process because they continue to fight even when they feel silenced or ignored by healthcare providers and services. PLWRD go through a lot in their medical journey, so it is not helpful when the doctor knows very little about their condition. Also, parents must learn to adapt to their new lifestyle, which sometimes means losing their jobs, moving away, and taking on the role of a full-time caregiver. Parents are the child's primary social support because they can count on them to be there every step of their medical journey. Overall, this is an emotional and tough journey that the PLWRD and their parents must go through, and they will do anything they can to get the doctors to listen and help them live a *good life*.

# Chapter 5

# 5 The Importance of Advocating

In this chapter, I will discuss the importance of advocacy for PLWRD and their social support network in two parts: self-advocacy and parental and systemic advocacy. PLWRD self-advocate for their rights, treatments, services, and what they truly need and want. Parental advocacy is when parents advocate for their children living with a rare disease, using the knowledge they learned to receive appropriate services and support and improve educational access and outcomes for their child. Lastly, systemic advocacy involves educating the public and working towards long-term changes. **Advocacy** is a "multifaceted role with various embodiments within the rare disease community" (Nori et al., 2022, p. 2).

Advocacy itself is connected to a broader dominant discourse and context in different ways, such as power dynamics, social media impact, intersectionality, and social expectations. Power dynamics is one of the dominant discourses because of its influence in society since it can control the voices of those advocating, especially those from marginalized communities (e.g., rare disease communities)! Next is the social media impact, which I found in my data to be either good or bad when using it to connect with others, the way information about rare diseases is being presented, and the advocacy efforts of groups making sure what they are posting is being seen across social media with accurate information. Then, intersectionality and social expectations go hand in hand. They go hand in hand because intersectionality in dominant discourses addresses the unique challenges and experiences different groups are face. Social expectations come in because society has serval stereotypes about gender roles, family dynamics, and race. One stereotype about gender roles that I debunked in my research is that fathers are not involved or do not have a significant role in their children's lives. This stereotype going around the internet and being in society as an expectation impacts the way fathers are seen. There are many fathers, like the ones in my research, who do so much work and are involved, especially when it comes to advocating for their children and spreading awareness about rare diseases.

Overall, PLWRD and their social support network members advocate daily by educating others about rare diseases and everything around them. All my participants are passionate advocators, so each part will showcase their lived experiences advocating. I will analyze them using conceptual frameworks of self-advocacy and systemic and parental advocacy to understand the importance and impact of advocacy (Block et al., 2011; Carey, Block, Scotch, 2020; Green et al., 2011; Nori et al., 2022; MacLeod et al., 2017; Manderson et al., 2016; Plackowski and Bogart, 2023; Rossetti et al., 2021; Silva et al., 2017; Bosi et al., 2019). Also, I will use Cheryl Mattingly's moral laboratory of care and Eva Kittay's care theory to show the connection between advocating and care. In the final section of this chapter, I will share the advice my participants gave when I asked what advice they would provide another PLWRD or parent going through what they went through or are currently going through.

## 5.1 Self- Advocacy

One of my participants expressed about advocacy,

> "I think to like advocate for yourself but to advocate smartly. Like to not just like push, push, and push, cause you're not gonna get what you want with that. It's an art to properly advocate, and to be like, you know, stroke the doctor's ego and be like you're really smart. So, I think, like to really encourage advocacy, and but more just like advocacy for your own quality of life…".

Rare and complex health conditions impact an individual at multiple times in their life, so that requires them to develop an understanding of their health condition and the factors that come with it, such as treatments, medication, how to deal with stigmatization, manage pain, and so much more (Manderson et al., 2016). PLWRD advocate for their rights and accessibility in various domains, including education, employment, medical fields, and social inclusion. From those I have spoken to, their advocacy work ranges from joining accessibility committees at school to help and advocate for themselves and others, working as a patient advocate, attending conferences and webinars, and connecting with other communities. Also, many advocated for things they truly wanted and believed in, even if that meant fighting their doctors and parents.

There have been many researchers and self- advocators who have studied advocacy in different ways, such as developing a conceptual framework on self-advocacy and empowerment strategies, raising awareness for self-advocating, exploring the importance and empowerment of self-advocating, and the challenges faced when self-advocating (Block et al., 2011; Carey, Block, Scotch, 2020; Nori et al., 2022; MacLeod et al., 2017; Manderson et al., 2016; Plackowski and Bogart, 2023; Bosi et al., 2019). I will be relying on concepts from these researchers and self- advocators in this section to analyze the different experiences of advocacy work my participants living with a rare disease have done.

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PLWRD become their own advocates as they grow, and many follow in their parent's footsteps, who have raised awareness and advocated for their needs and rights throughout their medical journey. In chapter 8, *Narratives of Rights*, in Carey, Block, and Scotch (2020) book, *Allies and Obstacles: Disability Activism and Parents of Children with Disabilities*, they state,

> "Building self-advocacy is a crucial component to ensuring rights. Parents and parent organizations have individually and collectively supported self-advocacy in many ways. For example, one interviewee, Mark, attributed his success as a self-advocate in part to the important role played by his mother, who fostered his development as a self-advocate, traveled with him, assisted him preparing speeches, and sometimes participated in events, especially when they were addressing families" (p. 205).

Just like Mark, my participant, Lucas, followed in his parent's footsteps by taking on the role of advocating and helping others. Throughout Lucas's medical journey in pediatric care, his parents -Tori and Sebastian- were strong advocates. I was fortunate to have interviewed them and will go into more detail about their advocacy work later in this chapter, but their work began the minute Lucas was born. Throughout Tori's pregnancy, the doctors never noticed anything off in the ultrasounds, so when Lucas was born, everyone was surprised that there were some medical conditions. Before their first
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appointment with the geneticist, Tori and Sebastian's families were involved in supporting and researching the symptoms Lucas had when he was born. One of their family members narrowed it down to two conditions, and they were right with one of them. When it came down to surgeries and doctors, Tori stated,

> "I was a very, very strong advocate for Lucas, and you know, there were times that he needed a surgery for something, and the one doctor did not have surgery time to do it. I would be calling another doctor in his care to ask if they had time".

Their work did not stop there, when they would have appointments outside their city, they would meet with other families and help educate them on handling situations, etc. So, Lucas grew up watching his parents advocating for him and helping others, and now he has adapted his advocacy skills. Lucas has been very active within his university. Lucas joined his university accessibility committee and is the student outreach director who looks at the accessibility survey results and makes suggestions for the university based on the results. Also, if students have any accessibility concerns, they talk to Lucas. He is also an off-campus soph, so he gives first-year undergraduate guidance and help whenever they need it. Lucas voices about his work, "my accessibility committee, and being a Soph, I've also been able to kinda educate other people and talk to other people and you know hopefully help other people who maybe are in a similar situation or know someone or stuff like that".

Another participant, Asher, was diagnosed with Synovial Sarcoma when he was 17 years old, so he was in his final year of high school, which is a big milestone. Asher was in the second semester of his grade 12 year and had to "drop out" of in-person school due to treatments and surgery. He was thankful to have gotten accommodation through the school to receive a teacher to come to their home and teach him, which allowed Asher to graduate with his cohort. Asher was named class valedictorian, which was a memorable moment for him,

> "It was a very touching moment in my life, just being up, there, being able to tell people my story of how I recently beat cancer, and I'm still able to graduate with

> my peers. It was like a very powerful moment in in my life, and it's a moment I'm very grateful for honestly".

Asher getting the opportunity to speak about his lived experience is an important aspect of self-advocacy. Plackowski and Bogart (2023) work on awareness and self-advocacy of adults living with a rare disease; in their article, they highlight the significance of listening and understanding the experiences of PLWRD and advocating for their rights. Most self-advocates spoke to indicate the importance of interpersonal education to rare disease advocacy and raising awareness (Plackowski and Bogart, 2023). So, PLWRD should educate others by discussing and telling their lived experiences and stories (Plackowski and Bogart, 2023). This is exactly what Asher did with his valedictorian speech "Being able to tell my story of the hardships that I went through, and kind of being an inspiration to my peers that I've known for a few years now it. It was very touching for me". Being there on the day of his graduation after spending the whole semester fighting cancer showed everyone his strength and raised awareness about rare diseases. The increase of rare disease awareness influences access to helpful diagnostic outcomes, treatments, and social support from everyone in a positive way (Plackowski and Bogart, 2023).

Other PLWRDs become self-advocates by seeking access to medical information through their resources and challenging their practitioners/ doctors/ specialists (Manderson et al., 2016). Rare disease self-advocates voice the need for more PLWRD to take part "in all types of media and positions of influence" because then it will lead to an increase and accurate representation of rare diseases that could make a positive impact relating to rare disease awareness, understanding, and even empathy (Plackowski and Bogart, 2023, p. 74).

For Lily growing up, there was a strong barrier between the knowledge she had about CMTC and the knowledge she could not access. There was not a lot of research on CMTC, it was hard for her to find anything, and even if she wanted to research what other people like herself went through, there was not a clear mechanism to do it. When she got to university, she was able to use the University library to access the medical

university library, which when she was a kid, she did not have these recourses. Nowadays, it is easier for individuals living with CMTC and their families to find resources and get guidance than it was for Lily.

Lily is a patient advocate for an organization on their Facebook and Instagram pages. People can message her questions and speak with her. This organization has brochures, documents, and even a book with information a general practitioner (GP) would need when a patient comes to them and is diagnosed with CMTC. None of these resources existed for her, and she did not know half of the information in the brochures and book until she began working with the organization. She was learning more about CMTC and was like “Oh, that explains so many things about why I cannot do that’, Oh, did you know that people do not know left and right with that condition? Often when it's affecting in the brain. That's not just me”. Organizations like the one Lily works for are essential resources for PLWRD and their families. Nori et al. (2022) article explores the contributions and challenges PLWRD, advocates, and caregivers face to shape global efforts in the context of rare diseases. They state, “Organizations established for a few rare diseases have matured over time into important resources for high-quality information and advocacy support for rare disease patients” (p. 1). Organizations that are “patient/caregiver-based” have very limited resources and rely on friends, friends, colleagues, and local communities for support to raise funding (Nori et al., 2022). These advocacy organizations are committed to educating, connecting, and supporting PLWRD, caregivers, researchers, clinicians, the pharmaceutical industry, healthcare providers, etc. (Nori et al., 2022). Also, they help further rare disease research, approvals for new treatments, and guarantee access to care (Nori et al., 2022). However, there are not organizations for all rare diseases. Roughly only half of the identified rare diseases have an advocacy organization, and they tend to be small, meaning they have limited resources and rely on volunteers to keep them going (Nori et al., 2022).

Finally, Gabriella, an advocate who has done a great deal of advocacy work, pushed for more from her doctors and fought for what she wanted. Gabriella has done advocacy work in Canada and some in the US, speaking at conferences, doing webinars, connecting with communities, engages in advocacy around nutrition support since she cannot eat

orally and is on intravenous nutrition. Gabriella also advocates for mental health awareness for people with chronic and rare diseases because she states that many of times, people focus more on the physical side and do not realize the mental toll that it has on people. Gabriella also advocates for more from her doctors, such as for treatments. The treatments she is on now are working and keeping her alive, which her doctors are happy with. However, Gabriella pushes for more to help her take the next step instead of just settling with what she has now.

Furthermore, self-advocacy involves setting and achieving personal goals and using skills to navigate obstacles (Block et al., 2011), which Gabriella did. Before she got sick, she played competitive sports, such as biathlon and water polo. Once she got sick, she had to stop them because she became paralyzed and, in a wheelchair, and could not do those sports anymore. Nonetheless, that did not stop her from playing any sports. Once she got used to being in a wheelchair, she began researching the Paralympics and seeing what sports nobody in Canada does, and she ended up choosing fencing. However, she did not realize that there was no money for it, no resources for support, and no one to train her. But everything worked out because since no one else in Canada played wheelchair fencing, she began getting asked if she would want to travel and compete in different places. When it came down to informing her doctors so they could make sure to have a plan for everything, she did not ask them if she could play, she told them what she was doing and that they had to be on board with it and help her with it. At the start, some of her doctors were not supportive of her doing competitive sports involving intensive exercise and activity. Also, she had a central line in her chest, which was placed in the same spot that is the main target area in fencing. So, Gabriella had to convince the doctors to do another surgery to switch the central line to the other side of her chest. She achieved what she advocated for and made it onto Team Canada.

Additionally, like how Gabriella was advocating for herself to the doctors for having a life full of fun, passion, and risk, which should be the right of all humans. Her doctors were also advocating for what was safest, which is an example of a moral laboratory (Mattingly, 2014).

Recognizing and supporting these advocacy efforts is crucial for fostering resilience and empowerment. Empowerment extends beyond healthcare decision-making and encompasses broader aspects of advocacy and self-advocacy. The self-advocacy movement empowers people to "stand up and speak up for themselves and others" (Pennell, 2001, p.223; Plackowski and Bogart, 2023, p. 64). Bosi et al. (2019) explored patient empowerment; they found that "the notion of empowerment is linked to patients' everyday life and is the base allowing for the patient engagement through which individuals and communities can express their needs, are involved in decision-making, take action to meet those needs" (p.283). In the context of rare diseases, there is a greater value for empowerment strategies due to the lack of expertise, poor quality of life, and low prevalence (Bosi et al., 2019). As Block et al. (2011) have explained in their article, "empowerment has been described as a method by which individuals and communities exert influence and control over resources, events, and outcomes" (p. 91). Influence and control are considered "key characteristics" of empowerment along with self-efficacy, awareness of rights (the individual and community), personal strengths, etc. (Block et al., 2011, p. 91). Self-efficacy is part of individual empowerment because people recognize their "ability to manage challenging situations and accomplish goals, which influences self-perception, feelings, and motivation" (Block et al., 2011, p. 91-92). Each participant I have discussed in this chapter has shown self-efficacy through all their different types of self-advocacy work. At the end of the day, "if you even reach one person, then you change one person, and give them perspective" (Plackowski and Bogart, 2023, p.73), which is truly needed when it comes to rare diseases.

## 5.2 Network Advocacy

*"There is no more powerful advocate than a parent armed with information and options."— Rod Paige*[10]

[10] (Quotefancy, 2024)

Taking care of a child while they are ill is always a challenging process for a parent, but if their child has a rare disease, it is more complex. Once a child is diagnosed or undiagnosed with a rare disease, their lifestyle begins to change, meaning the whole family's as well. Also, it means the child will need a strong social support network of people around them while undergoing appointments, treatments, and regularly. Parents are the primary social support children have throughout their medical journey, especially when the child has a rare disease that involves additional time, care, effort, and support from the parents.

Parents become advocates for their children to ensure their needs are met in various areas, such as ensuring they receive suitable medical care (e.g. treatments, surgeries), accommodations and support in schools, seeking resources, etc. Parent advocates develop knowledge about their child's rare disease, the healthcare system, policies, etc. (Rossetti et al., 2021). They connect with other families in similar situations, raise awareness and educate, work with advocacy organizations, etc. The parents I interviewed have done various advocacy work. They stand up to doctors for their children, advocate for surgeries, medication, referrals, and doctors, join advocacy groups, create programs, and teach their children how to self-advocate.

In this section of the chapter, I will analyze the advocacy work that the parents have done using conceptual frameworks of systemic and parental advocacy (Carey, Block, and Scotch, 2020; Green et al., 2011; MacLeod et al., 2017; Nori et al., 2022; Plackowski and Bogart, 2023; Rossetti et al., 2021; Silva et al., 2017). Also, I will be exploring the connection between advocating and care (Mattingly, 2014; Kittay, 2019).

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As mentioned previously, rare disease awareness can influence access to helpful diagnostic outcomes, treatments, and social support from everyone in a positive way (Plackowski and Bogart, 2023). It can also help improve educational access and outcomes for students with disabilities (Rossetti et al., 2021). Parents advocate for their children in schools by creating an individual education plan (IEP), being involved in the school and building relationships (Rossetti et al., 2021; Carey, Block, and Scotch, 2020).

Barriers do occur with parental advocacy in school systems, such as the lack of knowledge a parent might have about the policies, the terminology used, being overwhelmed by the education system, and the lack of time parents might have to advocate for IEP (Rossetti et al., 2021). From all the parents I have interviewed, some are educators working in schools, so they know how things work in the school system and the IEP process, but those who were not also had a great amount of knowledge about how the school system works. In addition to parental advocacy (or, as Rossetti et al. (2021) name it, 'Individual advocacy), there is systemic advocacy, which is less known (Rossetti et al., 2021). Systemic advocacy includes sharing the child's story on social media to educate the public, starting an advocacy organization, and creating relationships with legislators (Rossetti et al., 2021; Wright and Taylor, 2014). Also, systemic advocacy includes advocating for teacher training and educating other parents about the issues related to the school board (Rossetti et al., 2021). Barriers to systemic advocacy include a lack of knowledge of the education system, time, access to resources, and personal connections (Rossetti et al., 2021).

Julia, one of the parents who is not an educator but is very knowledgeable, is a strong advocated a lot for her daughter before she went to school and continued when she began school. Julia started advocating (specifically systemic advocacy) when she was trying to find a childcare program, that her daughter qualifies for. Unfortunately, Julia could not find any programs, so she and Ivey's home care nurse developed a training model program, which she then used and trained the people in the daycare, which took months, and when new staff came in, she re-trained them. Also, they had a tracking system for medical supplies, symptoms, etc. This was a very successful program, in that they extended the model to other children, so the daycare eventually accepted other children with complex needs based on the model. Fast forward to when Ivey entered school, the home nurse began doing the training there and helped Ivey. So, Julia did the training at the daycare and the home nurse did the training at school due to the jurisdiction that home care nurses cannot go into childcare but can go into schools. Even though the home nurse did the training at school, Julia also advocated at school with her husband. They worked on the IEP for Ivey because she has a three-hour medical management routine and must miss school due to surgeries and appointments.

Furthermore, parental advocacy involves advocating for medical treatment options, medication, and medical interventions and helping other parents in similar situations (Carey, Block, and Scotch, 2020; Nori et al., 2022). Harper, Peyton, Tori, and Sebastian advocate(d) strongly for mediation changes, doctor appointments, surgeries, and changes in the hospital that their child needs to go to and help others along the way.

To begin, Harper feels like she and her husband are constantly advocating for Theo. She advocates for doctors to see him when she brings him in through the emergency room, for Theo to be admitted when doctors want to send him home when he is admitted as an in-patient, and for medication changes. Harper shares that when Theo was in end-stage kidney failure, he was placed on five blood pressure medications, one being an emergency blood pressure medication to be taken when it is too high and would have immediate effect. However, it was not working, and the doctor wanted to send Theo home, so Harper and her husband had to advocate; they ended up in the pediatric critical care unit because the only medication that would help Theo was through an IV.

Peyton and Tori have advocated for their children in similar ways. Both advocated for what they know is better for their child when it came down to looking for doctors and knowing what hospital was better to go to than the one, they were referred to. Peyton shared when their local hospital wanted to send them to the hospital in London, which is their protocol since they are teamed with them. However, Peyton did not agree because a doctor from the National Institute of Health recommended, they see a doctor in Toronto, so Peyton advocated for them to refer her to the Toronto hospital. She expressed to Rose's doctors,

> "I understand that you that your protocol is for us to go to London. I said, however, I need to know that if we decide as a family that she should be at the Toronto Center for care, that your team will support us in that is it because obviously I cannot go if I do not have a referral and if I do not have a referral then they're not going to have access to all of the information you got from here".

Like Peyton, Tori advocated for Lucas's doctors to refer them to another hospital. Tori has grown to understand the system enough to know they did not have to stay in London

to receive treatments. So, Tori asked for referrals to another hospital and met with other doctors. Tori and Sebastian were very careful with the doctors they selected, the procedures, the hearing aid service, plastic surgery, etc.

Additionally, Green et al. (2011) expresses how there is evidence that children can "serve as connecting links to the community for their parents" (p.137). This is true because parents with children living with a rare disease or any medical condition connect because they are experiencing similar situations, and they tend to meet each other at houses, clinics, organizations, events, etc. Sebastian spoke about the networking and connections Tori would make when Lucas has appointments out of town. They would meet other families going through similar situations that they have already experienced, so Sebastian and Tori were able to educate and teach the parents they met on how to handle these specific situations, different strategies to use, etc. I think this is such a great thing parents do for each other because rare diseases and the experiences people go through are not talked about much and there is a limited amount of research there is about rare diseases, it is very beneficial to see not only the parents helping other parents out but also the PLWRD helping each other out too. No one will know more than those who have lived experiences, so it is great to see this type of support and care between everyone in the rare disease world.

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When listening to my participants and reading about the type of advocacy work parents do, there is a clear connection between advocacy and care. The amount of care that parents have for their children leads them to do a range of advocacy work for their children. In Eva Kittay and Ellen Feder's book, *The Subject of Care Feminist Perspectives on Dependency*, Kittay discusses becoming an advocate for her daughter, "To be heard, to be recognized, to have her needs and wants reckoned along with those of others, the mentally retarded individual requires an advocate- a role that has voice at its center. It is in the role of an advocate for my daughter, Sesha, that I enter into the field of "disability criticism". I have wondered not only how and where I fit into this critical discourse, into any public discourse. To do so, I must first tell the reader about her, for

she cannot speak for herself" (p. 259). Kittay learned about disability from her daughter, but also found "appreciation of care as a practice and an ethic" from her daughter and her caregivers (Kittay, 2011, p. 52). Kittay (2002) recognizes that, "foremost among her [daughter's] needs is the need for care. If she is to flourish, she needs good caring care- and lots of it. *Care* is a multifaceted term. It is a labor, an attitude, and a virtue" (p. 259). From all my participants, their advocacy work steams from their care for their child's needs. Their goal is for their child to have a good and healthy life, and to do so, they must advocate. Also, Kittay entering the field of "disability criticism" once she became an advocate for her daughter, is similar to how all my participants (the parents) put themselves in the healthcare field by becoming knowledgeable about the healthcare system, policies, and about the rare disease their child has. Some even changed their jobs to be in the healthcare field, and others used their knowledge to advocate.

Furthermore, my research builds on the work of Mattingly's moral laboratories about African American families care for children with disabilities and illness, which is delivered through narratives, so as a reader, you can understand the moral dilemmas they faced. My thesis follows Mattingly's lead by centering the narratives of my participants because I want the reader to understand their experiences. The difference between my work and Mattingly's, is that her work is on African American families, whereas my work is on families from different ethnicities and races (Whites, Arabs, and Eurasians).

Mattingly's moral laboratories play a role in the care of PLWRD because everyone in their social support network wants all of them (the PLWRD and their network) to have a "morally good life". Moral life is about the parents and doctors fighting for what they think is best for the PLWRD. It is understandable when the parents become stressed and begin to take charge of their child's needs because they want to create a morally good life for their child. It may lead to them losing their parental role since they start to treat their child as the patient or even the disease itself. This is not right. Yes, parents want to create a morally good life for their children, but it should not make them turn into whole different people. Chapter five of Mattingly's book is an example of this position because it is about a mother working hard to advocate for her daughter's health but at the same time losing herself as a parent along the way. Dotty (the mother) is very involved in her

daughter's (Betsy) care to the point where she advocates for her daughter to go through a high-risk and experimental bone marrow transplant, which the clinicians are against. Throughout her daughter's medical journey, Dotty worked hard, and she ended up referring to herself as "Rambo Mom" meaning a tough mom (Mattingly, 2014, p. 100). Dotty was not tough just for the fun of it; she was tough because she wanted her daughter to get the best treatments for her sickle cell anemia, she was very well informed, and she was not afraid to challenge the doctors.

How Dotty ended up being referred to as a "Rambo Mom" is similar to how Tori predicts that there is a star beside her name with the doctors because she would regularly push for things, such as early treatments and scheduling things so Lucas does not miss school. Tori is also like Dotty in that she wants the best treatments for Lucas and is not afraid to use her knowledge with doctors. However, Tori never advocated for things that would put Lucas at risk. Yes, she was an advocate, but she also kept her role as a mother. Tori expressed how the biggest challenges for her as a parent were deciding on elective surgeries and making decisions that would affect the rest of Lucas' life that she and Sebastian were making for him on his behalf so that maybe one day Lucas would look back and question why they decided to take one route instead of another.

Overall, when parents become advocates for their children, they also take on additional roles since advocacy "is a multifaceted role with various embodiments within the rare disease community" (Nori et al., 2022, p. 2). They serve as advocates by educating and motivating others about rare diseases (Nori et al., 2022) and advocating for their children's needs.

## 5.3 Advice from the people that need their voices heard

*"The people with the best advice are usually the ones who have been through the most"*

*- Unknown*[11]

[11] (Quotes 'n Thoughts, 2023)

At the end of each interview, I asked each participant what advice they would give another PLWRD or parent going through what they went through or going through. I asked for their advice because I wanted to end the interview with their thoughts and opinions on things they have already experienced so they can inform PLWRD and parents on what to do, not to do, look out for, etc. This is important for this research because the advice they told me are lived, personal experiences that raise awareness and are essentially advocacy, which not only will help and educate those going through similar experiences but also educate the healthcare professionals and staff, service workers, policymakers, and in general, **the public**! Even just listening to PLWRD and their social support network's stories, which often get overlooked, can encourage others to explore what rare diseases are, the challenges that come with them, the support available, etc.

It is time for a change in how the healthcare system addresses such health challenges and issues, especially when it comes to rare diseases because even though they affect a small population of people, new rare diseases continue to be discovered each year (Rare Diseases Working Group Report, 2017).

### 5.3.1 The PLWRD Advice

As mentioned previously, PLWRD follows in their parent's footsteps and become self-advocates, not just for themselves, but for others too. I asked them: *'What advice would you give a PLWRD who is just beginning their medical journey and those who are about to transition?'* Their answers proved just that because they gave caring and supportive advice to those going through what they went through.

The following are a couple of the PLWRD advice from the interviews:

~ "My main advice while you're still in peds is, get the pediatric team to start creating and getting contacts in the adult realm early… Definitely start working to build your own team, because that team would not be sitting there waiting for you".

~ “Do not like doubt yourself, do not think that you cannot do like things that anyone else can do. There’s supports, there's people that can help, help you along the way into kind of give you the resources to allow you to do well. I would also say, try to learn about yourself. Do not be self-conscious, kind of like embrace what's going on around you, and yeah, I just kind of keep working, keep pushing yourself to be as like as good as you can be with whatever you’ve dealt with”.

~ “So, even if you do not have friends, or you do not have a good social support, make one. Just do not be alone I'd say. Try to have people with you always just being there, always having someone to talk to”.

### 5.3.2 The Social Support Network Member Advice

Parents have done a great amount of advocacy work for their children throughout their whole medical journey, and when I asked them: *‘What advice would you give parents who are just beginning the medical journey with their child and those whose children are about the transition?’* They all were ready with advice regarding how to advocate for children, but some also advised about remembering to care for themselves along the way.

The following are a couple of the parent’s advice from the interviews:

~ “Do your own homework.”.

~ “I think for the most part, do you research”.

~ “Do not think you're alone. Even if it's on the other side of the world, you will find somebody that's been through it, that might have answers to your questions. Take your list to the doctors and do not stop ‘till you find the doctor that is willing to answer the questions. That's my advice”.

~ “Do not be afraid to ask questions. Do not be afraid to tell the medical team what you're worried about, and do not let them limit your visit to one thing. Especially if you're dealing with somebody with rare disorders, things that would not normally be connected might be and so everything is a clue”.

~ “I think there's so much you can learn from others. There's always something to be learned from, even if it's one tiny little nugget of knowledge. It can just help you understand your own journey, or maybe the next step. So, I think isolating yourself can make you go down a slippery slope. So, if you're able to and you have the energy to just reach out, talk to people, go on to Facebook. I think it is important to just keep connected”.

## 5.4 Conclusion

This chapter has examined the importance of advocacy in two parts: self-advocacy and parental and systemic advocacy. I used my participant's lived experiences as advocates to showcase the importance of each. PLWRD become their advocate as they grow up because they begin advocating for their rights and needs. Also, many PLWRD follow in their parent’s footsteps, who have advocated for them all their life. Parents become advocates to guarantee that their children’s needs are met. They become knowledgeable about their children's condition, how the healthcare system works, how the school system works, the different policies, etc. Also, many parents participate in systemic advocacy, which is when they educate the public and work toward making long-term changes. To examine the importance of advocacy, I used Mattingly’s moral laboratory of care and Eva Kittay’s care theory to show the connection between advocating and care In the final section of this chapter, I included advice from each of my participant's that they would give to other PLWRD and parents going through similar experiences. I believe their advice is significant in raising awareness and advocacy because we are hearing directly from people with lived experiences who that often get overlooked or ignored. They in turn help other individuals going through similar situations and they educate the public as well. Overall, “advocates are fighters. They fight against frustration, ignorance, and exhaustion. They fight for awareness, research, equity, and accessibility” (Plackowski and Bogart, 2023, p. 75).

# Chapter 6

# 6 Conclusion

Rare diseases are consistently globally emerging. Individuals can start having symptoms before birth while they are still a fetus, once they are born, in early childhood, or late childhood. My participants were at different stages of their lives when they began showing symptoms of their rare disease. I was able to create a safe space for each of my participants to voice their thoughts and opinions about their experiences and medical journey. My overall goal for this research was to showcase the voices that are not always heard. I had this goal since I was an undergrad doing a smaller version of this research because the problem with this type of work when talking about children's experiences is that we never hear from them. When speaking about children managing or suffering from rare diseases, it's easy to forget they have perspectives and views on it. The experiences PLWRD go through during their childhood and their support network cannot be fully understood without their perspective and contribution. These are challenging times because some of these diseases are not curable, and it is overall a stressful time. No one will truly understand how it is growing up in these conditions than PLWRD and their support network. Their perspective in this type of work is important because they provide meaningful perspectives of medical experiences and interactions. Therefore, my research focused on the voices that need to be heard who experienced and know what goes on during doctor visits, finding the right doctors, the environment of the hospitals/ clinics, finding/ accessing treatments, and the encounters with medical professionals. But, most importantly, who knows and experiences the various feelings, pain, and suffering that come with it all?

My research is important because it focuses on an overlooked population and documents the lived experiences of PLWRD from all ages. It highlights the experiences of the transition from pediatric care to adult care, which may not seem like a big deal because everyone goes through the transition in healthcare. However, my research raises awareness and identifies the unique challenges and needs of PLWRD. It recognizes the importance of a strong social support network that helps ensure the PLWRD is receiving

everything they need, staying by their side, doing extended research, educating themselves about the rare disease and healthcare system, and battling each challenge that gets in their way. My findings can help create improvements in the healthcare system and better the experiences of PLWRD and their social support networks in hospitals. The findings can help improve current policies and practices in hospitals, possibly create new ones, assist the building of transition programs in hospitals, and support all the current awareness about rare diseases.

I conducted my research remotely, whereas others who have conducted similar research have done it in person (Esquivel-Sada and Nguyen, 2018; Mazzucato et al., 2018; Sandquist et al., 2022; Silva et al., 2017). I interviewed PLWRD and their social support network members via Zoom, but I made sure to create a space that made all my participants feel safe and comfortable. I also did not conduct the interviews formally. I interviewed as if we were having a conversation together and allowing the participants to take the lead. I wanted their voices to be heard and let them share any of their experiences and stories that they feel comfortable doing. Furthermore, what makes my research stand out even more is that it is interdisciplinary- anthropology and disability studies combined- and adds to the emerging field of disability anthropology. Overall, I truly want my research to help fix the gaps in the healthcare system and improve the lived-experiences of PLWRD. They deserve to be heard and seen and have an army of people fighting, advocating, and raising awareness for them!

## 6.1 Key Findings

Using the eleven semi-structured ethnographic interviews conducted over Zoom, I explored the experiences of Canadian PLWRD and their support network members. Their experiences uncover several findings in this thesis. I used the theoretical and conceptual lenses of theories of care, theories from medical anthropology and disability anthropology, and theories of support and advocacy to analyze them.

First, in chapter two, I described the three stages of transition (Before, In-Between, and After) through three different experiences, which I analyzed using theories of care and disability anthropology. The experiences identified how the PLWRD social support

networks change once the transition occurs; some PLWRD do not receive any preparation for the transition, and some do, and the level of care, support, and respect from doctors given in pediatric care was not present in the adult care for many. Next, in chapter three, I explored the different support dynamics and roles of the parents, siblings, and friends using the theoretical and conceptual lenses of care, medical anthropology, and disability theories. Mothers and fathers have specific roles when supporting their children living with a rare disease. Mothers are well known throughout literature as the primary caregivers who do everything, and fathers tend to be out of the conversation. I showed how fathers are involved in their children's lives, showing them care and support, making changes to their work schedule to make it to appointments, and just being there for the family. Siblings are another important part of a PLWRD network, and their roles vary depending on the family if they want the other children involved in the PLWRD medical journey from the beginning or just when they get older. When it comes to friends, they also are a part of the PLWRD network because they provide care and support but depends on which type of friend. School friends are either supportive or non-supportive, whereas hospital friends and parent's friends are always supportive and there. Also, I explored changes in power and control dynamics. Once the PLWRD transitions to adult care, they become ready to take their health care into their own hands, meaning the parents must step down and allow them to take charge.

Then, in chapter four, I discuss the supports and challenges PLWRD, and their social support networks face in pedantic care, continue to face in adult care, and new ones that form in adult care through my participant's experiences. I analyzed the experiences using theoretical and conceptual frameworks of care, medical anthropology and disability theories, and advocacy support. Almost all the challenges that PLWRD and their social support network faced in pediatric care continued to be faced in adult care, along with a few new challenges (Table 6). The same goes for the supports. The ones they face in pediatric care continue to face in adult care and the new supports that form are just modified (Table 7). Finally, in the fifth chapter, I focus on the importance of advocacy for PLWRD, and their social support network using conceptual frameworks of self-advocacy and systemic and parental advocacy to understand the importance and impact of advocacy and using Mattingly's moral laboratory of care and Kittay's care theory to

show the connection between advocacy and care. Parents do their research and gain knowledge of rare diseases, how the health system works, the policies, find the best doctors, etc. They become advocates for their children to guarantee their needs are being met. Then the PLWRD grows up, they follow in their parent's footsteps and become their own advocates for themselves and others.

The key findings from my research emphasize the importance of rare diseases, share new insights into how to increase awareness, provide a deeper understanding of the lives of PLWRD as they transition into adult care, and the lived experiences will contribute to disability anthropology and medical anthropology.

## 6.2 Limitations

When I first started planning out this research, I had big plans on how I would recruit participants and who I would talk to. When I began, I realized some limitations with the recruitment process. I planned to use social media platforms to recruit, such as community groups, including online communities on Facebook and X (formerly known as Twitter). When it came to online groups, I knew that I was only able to join public ones, due to ethics, which was a big limitation. The active Facebook groups where I believe I could have gained participants were all private groups, which I could not join. The private groups were the most active in comparison to the public ones. The public Facebook groups were nonactive, had barely any members, did not use the space properly, and were overall, not effective.

Another limitation was not being able to speak with PLWRD under the age of 18 and who are not living in Canada. The eligibility criteria for participating in my research were that you must be in Canada, over the age of 18, have a rare medical condition, be a part of a social support network for a person with a rare medical condition, and must speak English. When I had parents emailing me interested in participating with their children, I had to make sure to check that their child was 18 or older, and if not, they would have to participate solo if they were still interested in participating. This upset me because my goal for this research is to showcase the voices that are not always heard, so having this limitation blocked me from doing so. However, since I will be continuing this project in

my Ph.D., I do intend to include everyone. Additionally, I did have to turn away potential participants because they were not from Canada. As mentioned in the methodology section, I posted my recruitment information on “RareConnect” a website of several rare-condition communities. I hoped to recruit individuals from Canada; however, I did not receive any interest from anyone in Canada, but instead, people from Brazil and Algeria.

## 6.3 To Be Continued

*I have reached the end of my master’s research; however, this is not the end of my time researching PLWRD and its transition from pediatric care to adult care. I will be taking this research further in my Ph.D. because it is very important, and I want to be a part of the growth of awareness and resources around it. But especially continue showcasing PLWRD and their support network members'* ***voices.***

“When you have a rare disease, you face two battles: one, the illness itself, and the other, living in a world where so few people understand what you’re up against”- Unknown.

*I want to continue meeting those living with a rare disease and those who take care of and support them. I conducted my MA research in Canada, I met many great people and had the best conversations with them. I am grateful that they trusted me and shared their stories and experiences that I was able to use in this thesis. I learned from them about what kind of supports and treatments were and were not offered, how the healthcare systems work, etc., in the Canadian context. For my Ph.D., I will be taking my research to another country because there are more people I would like to meet, speak, and learn from. So, that is why this research will be continued in the next chapter of my academic journey.*

# References


Alexion AstraZeneca Rare Disease. (2022). The impact of RARE disease on sibling experience Impact report. www.rarerevolutionmagazine

Biehl, J. (2013). *Vita: Life in a zone of social abandonment*. Univ of California Press.

Black, S. P. (2018). The ethics and aesthetics of care. *Annual Review of Anthropology*, *47*, 79-95.

Block, P., Kasnitz, D., Nishida, A., & Pollard, N. (Eds.). (2015). *Occupying disability: Critical approaches to community, justice, and decolonizing disability*. Springer Netherlands.

Bosi, G., De Santis, M., Hervás, C., Weinman, A., & Bottarelli, V. (2019). Patient empowerment of people living with rare diseases. Its contribution to sustainable and resilient healthcare systems. *Annali dell'Istituto superiore di sanita*.

Brentano, E. (2024, May 16). *24 things people with rare diseases wish others understood*. The Mighty. https://themighty.com/topic/rare-disease/living-with-rare-disease-what-to-know/

Brolin, R., Hanson, E., Magnusson, L., Lewis, F., Parkhouse, T., Hlebec, V., ... & Becker, S. (2024). Adolescent young carers who provide care to siblings. In *Healthcare* (Vol. 12, No. 3, p. 316). MDPI.

Carey, A. C., Block, P., & Scotch, R. K. (2020). *Allies and obstacles : disability activism and parents of children with disabilities*. Temple University Press.

Casey, B. (2019). *Canadians affected by rare diseases and disorders : improving access to treatment /*. House of Commons, Canada = Chambre des communes, Canada,.

Casey, B., & Murray, M. (2022). Arts-based Research in the Social Sciences. In *The SAGE Handbook of Qualitative Research Design* (Vol. 2, p. 517–). SAGE Publications Ltd. https://doi.org/10.4135/9781529770278.n32

Corkins, K.G., Miller, M. A., Whitworth, J. R., & McGinnis, C. (2018). Graduation day: healthcare transition from pediatric to adult. *Nutrition in Clinical Practice*, *33*(1), 81-89.

Currie, G., & Szabo, J. (2019). “It is like a jungle gym, and everything is under construction”: The parent's perspective of caring for a child with a rare disease. *Child: Care, Health and Development*, *45*(1), 96-103. https://doi.org/10.1111/hex.12958

Currie, G., & Szabo, J. (2019). “It would be much easier if we were just quiet and disappeared”: Parents silenced in the experience of caring for children with rare diseases. *Health Expectations: an International Journal of Public Participation in Health Care and Health Policy*, 22(6), 1251–1259. https://doi.org/10.1111/hex.12958

De Morais, R. D. C. M., de Souza, T. V., dos Santos Oliveira, I. C., & de Moraes, J. R. M. M. (2018). Structure of the social network of mothers/caregivers of

hospitalized children. *Cogitare Enferm*, *23*(1), 1-9. https://doi.org/10.5380/ce.v23i1.50456

Esquivel-Sada, D., & Nguyen, M. T. (2018). Diagnosis of rare diseases under focus: impacts for Canadian patients. *Journal of community genetics*, *9*(1), 37-50.

Ginsburg, F., & Rapp, R. (2013). Disability worlds. *Annual Review of Anthropology*, *42*(1), 53-68.

Ginsburg, F., & Rapp, R. (2024). *Disability Worlds*. Duke University Press.

Green, S. E., Barnhill, J., Green, S., Hawken, D. T., Humphrey, L. S., & Sanderson, S. (2011). Creating a village to raise a child: Constructing community in families of children with disabilities. In *Disability and community*. Emerald Group Publishing Limited.

Hartblay, C. (2020). Disability expertise: claiming disability anthropology. *Current Anthropology*, *61*(S21), S26-S36.

Iacobucci, M., Smoliak, O., & Breen, A. (2022). The Experiences of Young Sibling Carers: A Qualitative Study. *Canadian Journal of Family and Youth/Le Journal Canadien de Famille et de la Jeunesse*, *14*(1), 235-255.

Kittay, E. F., & Feder, E. K. (2002). *The subject of care : feminist perspectives on dependency*. Rowman & Littlefield Publishers.

Kittay, E.F. (2011). The Ethics of Care, Dependence, and Disability. An International Journal of Jurisprudence and Philosophy of Law. 24 (1), 49-58

Kittay, E. F. (2019). *Learning from my daughter : the value and care of disabled minds*. Oxford University Press.

Kittay, Eva (2020). *Learning from My Daughter. The Value and Care of Disabled Minds*. Oxford New York: Oxford University Press.

Larotonda, A. (2016). 2.2 Experiencing Rare Diseases. In L. Manderson (Ed.), E. Cartwright (Ed.), & A. Hardon (Ed.), *The Routledge Handbook of Medical Anthropology* (pp. 26-31). Milton Park, Abingdon, Oxon; New York, NY: Routledge.

Leiter, V. (2011). Bowling together: Foundations of Community among Youth with Disabilities. In *Disability and Community* (Vol. 6, pp. 3–25). Emerald Group Publishing Limited. https://doi.org/10.1108/S1479-3547(2011)0000006004

Leiter, V. (2012). *Their time has come : youth with disabilities on the cusp of adulthood* (1st ed.). Rutgers University Press. https://doi.org/10.36019/9780813553306

MacLeod, H., Bastin, G., Liu, L. S., Siek, K., & Connelly, K. (2017, May). " Be Grateful You Do not Have a Real Disease" Understanding Rare Disease Relationships. In *Proceedings of the 2017 CHI Conference on Human Factors in Computing Systems* (pp. 1660-1673).

Manderson, L., Cartwright, E., & Hardon, A. (2016). *The Routledge Handbook of medical anthropology*. Routledge.

Mattingly, C. (2013). Moral selves and moral scenes: narrative experiments in everyday life. *Ethnos*, *78*(3), 301-327.

Mattingly, C. (2014). *Moral laboratories: Family peril and the struggle for a good life*. Univ of California Press.

McKearney, P., & Amrith, M. (2021). Care. *Cambridge Encyclopedia of Anthropology. CEA*.

Mazzucato, M., Visonà Dalla Pozza, L., Minichiello, C., Manea, S., Barbieri, S., Toto, E., ... & Facchin, P. (2018). The epidemiology of transition into adulthood of rare diseases patients: results from a population-based registry. *International Journal of Environmental Research and Public Health*, *15*(10), 2212.

McGarry, K. A., & Mannik, L. (2017). *Practicing ethnography: a student guide to method and methodology*. University of Toronto [Ontario] Press.

Meltzer, A. (2019). Comparative life experiences: Young adult siblings with and without disabilities' different understandings of their respective life experiences during young adulthood. *Families, Relationships and Societies*, *8*(1), 89-104.

Meltzer, A., & Kramer, J. (2016). Siblinghood through disability studies perspectives: Diversifying discourse and knowledge about siblings with and without disabilities. *Disability & Society*, *31*(1), 17-32.

*NCAN's Inspiration Corner*. (n.d.). *Welcome to Inspiration Corner*. Neuroendocrine Cancer Awareness Network. https://netcancerawareness.org/inspiration/inspiration-corner-313/

Nishida, A. (2022). *Just care : Messy entanglements of disability, dependency, and desire*. Temple University Press.

Nori, M., Fisher-Vance, D., Wuerth, L., Colenso, R., & Donovan, D. J. (2023). The Global Role of Patients, Advocates and Caregivers in Rare Diseases. *Future Rare Diseases*, *2*(2). https://doi.org/10.2217/frd-2022-0003

Parker, C., Scott, S., & Geddes, A. (2019). Snowball sampling. *SAGE research methods foundations*.

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

Picci, R. L., Oliva, F., Trivelli, F., Carezana, C., Zuffranieri, M., Ostacoli, L., ... & Lala, R. (2015). Emotional burden and coping strategies of parents of children with rare diseases. *Journal of Child and Family Studies*, *24*(2), 514-522.

Plackowski, E. F., & Bogart, K. R. (2023). "If not me, who?": Awareness- and Self-Advocacy-Related Experiences of Adults With Diverse Rare Disorders. *Qualitative Health Research*, *33*(1–2), 63–80. https://doi.org/10.1177/10497323221135974

Quotefancy. (2024). *Top 5 Rod Paige Quotes (2024 update)*. quotefancy. https://quotefancy.com/rod-paige-quotes

Quotes ’n Thoughts. (2023, April 29). *The people with the best advice are usually the ones who have been through the most.* Pinterest. https://www.pinterest.com/pin/the-people-with-the-best-advice-are-usually-the-ones-who-have-been-through-the-most--291819250867364360/

Rare Diseases Working Group Report. (2017). Critical Care Services Ontario.

Rau, A., & Coetzee, J.K. (2022). Designing for Narratives and Stories. In *The SAGE Handbook of Qualitative Research Design* (Vol. 2, p. 700–). SAGE Publications Ltd. https://doi.org/10.4135/9781529770278.n43

Reiss, J., & Gibson, R. (2002). Health care transition: destinations unknown. *Pediatrics (Evanston), 110*(6 Pt 2), 1307–1314.

Ries, H., Block, P., & Kassem, D. (2024, September 17-19). *The Role of Siblings in Advancing the Inclusion and Wellbeing of People with Disabilities* [Conference presentation]. Community Living Ontario Conference 2024, Toronto, ON, Canada.

Rossetti, Z., Burke, M. M., Rios, K., Tovar, J. A., Schraml-Block, K., Rivera, J. I., Cruz, J., & Lee, J. D. (2021). From Individual to Systemic Advocacy: Parents as Change Agents. *Exceptionality : The Official Journal of the Division for Research of the Council for Exceptional Children*, *29*(3), 232–247. https://doi.org/10.1080/09362835.2020.1850456

Rutherford, D. (2020). Disability Worlds: Wenner-Gren Symposium Supplement 21. *Current Anthropology, 61(S21),* 1-146. https://doi.org/10.1086/706371

Salvador, M. D. S., Gomes, G. C., Oliveira, P. K. D., Gomes, V. L. D. O., Busanello, J., & Xavier, D. M. (2015). Strategies of families in the care of children with chronic diseases. *Texto & Contexto-Enfermagem*, *24*(3), 662-669.

Sandquist, M., Davenport, T., Monaco, J., & Lyon, M. E. (2022). The Transition to Adulthood for Youth Living with Rare Diseases. *Children*, *9*(5), 710.

Silva, M.A., Moura, F. M., Albuquerque, T.M., Silva Reichert, A. P., & Collet, N. (2017). Network And Social Support In Children With Chronic Diseases: Understanding The Child's Perception. *Texto & Contexto - Enfermagem*, 26, pp. 1-10. doi:10.1590/0104-07072017006980015.

Statistics Canada. (2023, September 13). *Access to health care.* Statistics Canada. https://www150.statcan.gc.ca/n1/pub/82-570-x/2023001/section3-eng.htm

Tasker, J. P. (2024, January 22). *Canada’s population is booming- access to family doctors has not kept pace.* CBC News. https://www.cbc.ca/news/politics/canada-popuation-booming-family-doctor-access-1.7087794

Talarico, R., Marinello, D., Cannizzo, S., Gaglioti, A., Ticciati, S., Carta, C., Kodra, Y., Azadegan, M., Taruscio, D., Mosca, M., & Turchetti, G. (2020). Shaping the Future of Rare Diseases after a Global Health Emergency: Organisational Points to Consider. *International Journal of Environmental Research and Public Health*, *17*(22), 8694-. https://doi.org/10.3390/ijerph17228694

Titgemeyer, S. C., & Schaaf, C. P. (2020). Facebook support groups for rare pediatric diseases: quantitative analysis. *JMIR pediatrics and parenting*, *3*(2), 1-10. https://doi.org/10.2196/21694

Titgemeyer, S. C., & Schaaf, C. P. (2022). Facebook Support Groups for Pediatric Rare Diseases: Cross-Sectional Study to Investigate Opportunities, Limitations, and Privacy Concerns. *JMIR Pediatrics and Parenting*, *5*(1), 1-10. https://doi.org/10.2196/31411

Toulany, A., Willem Gorter, J., & Harrison, M. (2022). A call for action: Recommendations to improve transition to adult care for youth with complex health care needs. *Paediatrics & Child Health*, *27*(5), 297–302. https://doi.org/10.1093/pch/pxac047

U.S. National Library of Medicine. (2004, July). *Paediatricians in Canada: Frequently asked questions*. Paediatrics & child health. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2721161/

Watkins, D. (2021, February 28). *Pope Francis prays for people suffering from rare diseases*. Vatican News. https://www.vaticannews.va/en/pope/news/2021-02/pope-francis-cardinal-turkson-message-world-rare-diseases-day.html

Wright, A. C., & Taylor, S. (2014). Advocacy by parents of young children with special needs: Activities, processes, and perceived effectiveness. Journal of Social Service Research, 40(5), 591–605. https://doi.org/10.1080/01488376.2014.896850

Wong-Rieger, D. (2020, April 28). *Covid-19 impact on Rare disease patients: Letter to ministers of health & responses*. CORD Canadian Organization for Rare Disorders. https://www.raredisorders.ca/covid-19-impact-on-rare-disease-patients/

Zurynski, Y., Frith, K., Leonard, H., & Elliott, E. (2008). Rare childhood diseases: how should we respond? *Archives of disease in childhood*, *93*(12), 1071-1074.

# Appendices

## Appendix A: Ethics Approval Letter

Western Research

**Date:** 27 April 2023

**To:** Dr Pamela Block

**Project ID:** 114824

**Study Title:** Aging out of Children's Hospitals - Exploring the Transition Out of Pediatric Care for Youth with Rare Medical Conditions and their Social Support Networks

**Application Type:** NMREB Amendment Form

**Review Type:** Delegated

**Full Board Reporting Date:** 05/May/2023

**Date Approval Issued:** 27/Apr/2023 10:55

**REB Approval Expiry Date:** 06/Dec/2023

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Dear Dr Pamela Block,

The Western University Non-Medical Research Ethics Board (NMREB) has reviewed and approved the WREM application form for the amendment, as of the date noted above.

# Curriculum Vitae

| | |
|---|---|
| **Name:** | Dima Kassem |
| **Post-secondary Education and Degrees:** | Western University<br>Bachelor of Arts Honours Specialization in Anthropology<br>2018-2022 B.A.<br><br>Western University<br>Master of Arts in Socio-cultural Anthropology<br>2022-2024 M.A. |
| **Honours and Awards:** | Deans Honour List<br>Western University 2020-2021<br>Public Anthropology's Community Action Project Award<br>Western University2019<br>The Western Scholarship of Distinction<br>Western University2018 |
| **Related Work Experience:** | Graduate Student Research Assistant at Western University:<br>Care Relationships of Adult Disabled and Nondisabled Siblings<br>August 2022 – Present<br><br>Graduate Teaching Assistant at Western University:<br>Cultures of Health, Illness, Disability and Healing<br>Jan 2024 – April 2024<br>Jan 2023 - April 2023<br><br>Volunteer Research Assistant at Western University:<br>Heart and Mind Study & Hockey FIT Study<br>2018-2019 |
| **Academic Conferences:** | Community Living Ontario<br>Inspiring Possibilities AGM & Conference 2024<br>Richmond Hill, ON, Canada 2024<br><br>Sedimented Histories, Vital Trajectories<br>50th Annual Canadian Anthropology Society (CASCA) Conference<br>Kelowna, BC, UBC Okanagan on Syilx territory 2024<br><br>Dr. Benjamin Goldberg Developmental Disabilities Provincial Research Day, 8th Annual Conference<br>London, Ontario, Western University 2023 |