CHARACTERIZING THE EXPERIENCE OF ACCESSING MEDICAL CARE FOR CHILDREN WITH RARE OR UNDIAGNOSED COMPLEX CONDITIONS

by

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ABSTRACT

There is increasing awareness of rare disease as a distinct entity. Advances in diagnostic testing have identified over 7000 individual rare diseases affecting millions of patients worldwide, many of them children due to the frequent genetic etiology of these conditions. A medical system that is often based on disease-specific expertise and treatment must adapt to meet the needs of those with rare conditions. However, there is little known about the unique experiences of parents of children with very rare or undiagnosed diseases in accessing medical care. An in-depth look into their circumstances will identify areas for improvement and improve the family experience of care.

The objectives of this study were to analyze this parent experience particularly regarding their relationships and communication with physicians. In addition, the parent perspectives regarding the connections between themselves and health care providers, as well as between health care providers, will be explored. Participants were recruited through the TIDE-BC (Treatable Intellectual Disability Endeavor of British Columbia) Complex Diagnostic Clinic, as parents of children with: (1) “Complex intellectual disability,” defined as undiagnosed intellectual disability with other prominent features; and (2) Ongoing care from at least three health professionals. Semi-structured interviews with parents explored a number of topics including: their experience of their child’s illness and its impact on the family, accessing necessary medical care for their child, developing their own expertise and relating to physicians. Interviews were transcribed and analyzed using discourse analysis methodology.

Themes identified included the pervasive and multi-faceted nature of uncertainty and its contribution to changes in roles for parents, physicians and peers. A key gap identified was a lack of coordination of care and lost opportunity for collaboration amongst professionals and with the family. Addressing these unique experiences and moving towards non-categorical design of health care service models will better serve the needs of children with rare and undiagnosed conditions.

PREFACE

This dissertation is an original, unpublished work by T. Dewan, who was responsible for the study design, conduct of interviews and analysis of results.

This study was approved by the Children's and Women's Research Ethics Board of the University of British Columbia (certificate number H12-00034).

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LIST OF ABBREVIATIONS

ASD – Autism Spectrum Disorder

CDC- Complex Diagnostic Clinic

CFRI- Child Family Research Institute

CoC- Coordination of Care

CT- Computed Tomography

DA- Discourse Analysis

EEG- Electroencephalogram scan

TIDE BC- Treatable Intellectual Disability Endeavor of British Columbia

UBC- University of British Columbia

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CHAPTER ONE: INTRODUCTION

The following chapter will introduce this thesis through a description of the existing state of knowledge in the field. The rationale underlying the current study will be described, as will the context in which the study took place.

1.1 Background

The recognition of rare disease as a distinct entity is a relatively recent phenomenon, emerging within the past two decades. The term “rare disease” calls to mind a handful of conditions involving a sparse number of individuals scattered over the globe. In fact, there are over 7000 known rare diseases, collectively impacting about three million Canadians. The exact definition of a rare disease can vary, but typically has a prevalence of less than 1 in 2000. [1] These diseases are often serious, chronic and degenerative. By virtue of their rarity and heterogeneity, they are challenging to study with randomized controlled trials and precise statistical methods, resulting in difficult clinical decision making and a lack of predictive capability for providers and patients.[2]

This further translates into health inequities and disparities that can lead to poor health outcomes for those living with rare diseases.[3,4] Barriers to optimal health can include inequities in accessing diagnostic tests, lack of available treatments and undue social consequences as a result of the disease and the failure of society to acknowledge it.[4,5] The need to re-design health care systems and tailor research and policy agendas to meet the needs of those with rare diseases is becoming a high priority among medical, government and patient organizations.[4,6] However, our ability to identify and diagnose rare diseases continue to outpace our knowledge of the patient experience. An

important component of these redesigns must include attention to the distinct needs and perspectives of individuals who are living with rare diseases.

These distinctive experiences have broad-reaching effects on the personal-social worlds of patients and their experience of health care. Garrino et.al. (2015) organized these experiences around five themes: dealing with disease development, living with the disease, everyday living, relating to others and relating to professionals.[7] Certainly, some of these themes are shared with those who suffer from common chronic conditions, such as living with manifestations of the disease.[8,9] Some however are more specifically related to the rarity of the condition, or alternatively, the reality of having one's health problems remain undiagnosed. For instance, the challenge of describing a rare or unknown disease to a peer or family member will be different than the same exercise for a more common condition. There are likely other important and under-recognized differences between the experiences of those with rare and common chronic conditions.

One reality that is common to many chronic conditions is the presence of uncertainty.[10] This begins with the onset of signs and symptoms that are unexplained, transforming the pre-illness predictable world to one containing many unknowns, often reaching a significant transition point at the time of diagnosis.[11] For some patients, the time surrounding diagnosis facilitates acceptance of the disease and the initiation of coping strategies.[12,13,14] However, this process of obtaining a diagnosis is more complex than it might initially appear.

In the current biomedical diagnostic-therapeutic perspective, there is a common expectation that provision of a diagnosis will be accompanied by specific information regarding etiology, management or prognosis. This assumption may be correct for common, well-described disease entities such as type 1 diabetes where the biological basis is known and targeted treatments are available. Consider however the diagnosis of autism spectrum disorder (ASD) for which etiologic information is lacking, diagnosis is based on descriptive criteria and management consists mainly of developmental and behavioural supports. Recent work by Miller and Rosenbaum (2016) addresses the limitations of this disease perspective in healthcare, that which relies on medical-etiological diagnoses and targeted treatments. First of all, many conditions, particularly those related to developmental or mental health conditions are diagnosed based on descriptive criteria. They are seldom, if ever, accompanied by extensive etiologic information or directed treatments. Secondly, even within a diagnostic group there are often varied manifestations among individuals and therefore it can be inappropriate to consider or treat these patients as a single group.[15] Addressing the complexities surrounding the concept of diagnosis is beyond the scope of this paper. However, it is imperative that the limitations of the medical-etiological diagnosis are acknowledged, as this is a focus in much of the existing rare disease literature.

Even though many diagnoses are accompanied by uncertainty, this is undoubtedly a hallmark of the rare disease experience and, as such, has been a focus of previous research. For individuals with rare diseases, uncertainty is not only restricted to the unknown nature of the illness, but is distributed across many aspects such as existential, etiologic, treatment-related, situational, biographical and social.[16] The

uncertainty is particularly intense when the disease is serious and potentially life-limiting.[16,17] The future now invokes stress and expectation of difficulty and loss, so the present become discontinuous from both the familiar past experience and the unknown future.[16] This persistent and multidimensional uncertainty is central to the illness experience in individuals with rare diseases.[10,16,18,19]

Clearly these distinct experiences among those with rare diseases will influence their interactions with the health care system. These diseases also tend to affect multiple organ systems and require multidisciplinary care, both to reach diagnoses and to deliver ongoing services.[20] However, systematic barriers in the health care system result in widespread disenfranchisement of the rare disease community.[3,20,21] Knowledge and expertise in specific rare diseases are often concentrated in centres of excellence, but knowledge dissemination to front line providers is insufficient to provide care and facilitate appropriate testing and referral.[20,22] Geographic, social and health inequities further hinder access to specialized assessments.[20,23,24] In addition, there are often inadequate services in the social and educational systems for patients with rare or undiagnosed diseases, as criteria for support programs are often reliant on a known and well-described diagnosis.[25,26] These factors give rise to the unfortunate situation where a system that should be providing the rare disease community with support, knowledge and advocacy, is often a principle source of frustration and difficulty for patients.[20,27] This stems largely from a medical system built around categorical and diagnostic divides, as opposed to one that is focused on the need of the patient and family, with provision of supports that are required from a functional, practical perspective.

The initial period leading up to and immediately surrounding diagnosis is of key importance and may have a significant impact on the relationship between patients and their health care providers moving forward. Misdiagnosis regarding the etiology of presenting symptoms is common (up to 40%), as is minimization of the patient's concerns or misattributing complaints to psychological illness.[4,27,28] Not surprisingly, this can set the stage for ongoing mistrust in health care providers and an overall lack of confidence in the medical system.[4,25] Many individuals with rare disease were deeply disappointed in their experience of diagnostic disclosure.[17,29] The delivery of a diagnosis, where one can be found, often occurs under less than ideal conditions, with a lack of information and psychological support.[28,30,31] Also, patients and families prioritize issues such as timing and their own personal readiness to deal with the information presented.[11,26] For patients, it can be as important for the diagnosis to be delivered in a sensitive manner as it is for a diagnosis to be disclosed as quickly as possible.[29] Some have suggested that particularly in lesser known or less predictable conditions, patients may absorb physician's initial comments or predictions in a literal fashion. Physicians need to ensure that they are mindful of the potential potency of their words and are communicating clearly.[17]

Patients with rare disease place a high priority on their interactions with physicians throughout the course of their illness, setting the stage for both positive and negative influences.[29] Positive patient-physician interaction and communication are known to correlate with optimal patient health outcomes[32–34] and can facilitate the patient's adjustment to a chronic disease diagnosis.[35] However, the reality of persistent uncertainty and diagnostic dilemma sets the stage for perceptual differences and

communication challenges between patients and physicians. For instance, even the point of diagnosis is experienced differently from each vantage point. The physician may feel a sense of closure and the comfort of certainty, as their diagnostic dilemma draws to a close.[11] For the individual and family, however, the picture is often incomplete and they are left with persistent and new uncertainties.[11,23] Interactions such as these highlight the different perspectives each has from the outset.

Although health care providers are uniquely positioned to provide support to individuals with rare diseases, another reason that positive relationships may be particularly difficult to achieve is related to roles and expectations. The care of patients with rare disease often requires significant deviation from the typical models of care for patient-physician interaction. These include paternalistic, interpretive, informative and shared decision-making models. Each is distinct, but all are based upon the assumption that expertise rests with the physician and can be shared with the patient, forming the basis of decision making.[36] The foundation of physician expertise and practice in the modern era is evidence-based medicine often leading to clinical practice guidelines that support physicians in providing the highest quality care for a particular disease.[37] This accumulation and compilation of knowledge is often impossible in rare diseases due to few patients and high variability. In these circumstances, it is often the patient who knows more about their condition than does the physician. The physician can thus feel challenged in his traditional authority and helpless due to the lack of guidance and treatment he can offer.[7] Patients are no longer able to look to their physician as the ultimate authority on their condition which contributes to a lack of confidence.[20,27]

However, patients may desire other forms of assistance from their physician. They value advice and guidance on non-medical topics, such as counselling on the social aspects of living with a rare disease and how to cope with reactions from others.[27,38] This support may be even more crucial for patients with rare diseases, for whom medicine does not have clear answers or treatments. A non-categorical approach that is not directed to a particular diagnosis and instead addresses the patient's needs regarding function and quality of life could have great benefit.[15,21,39,40] This shift comes with a need for adaptation as physicians move away from disease-based care, but professional roles are very resistant to change.[15,41] An ongoing struggle ensues regarding role definitions and discrepancies between what physicians are providing and what patients need or expect.[42] Through these encounters, health care providers can contribute to the isolation and stigma felt by an individual with a rare disease.[7,43,44]

This realignment of medical expertise in rare diseases ends up falling on the patient and family. The concept of the expert patient was first coined by the Department of Health in the United Kingdom in 2001, in reference to the emergence of patient self-management programmes and the effect these were having on the physician-patient dynamic.[45] However, this concept is perhaps even more applicable to patients with rare disease, who are often in the position of informing their health care providers about their disease, instead of the reverse.[27,42] This patient expertise comes with both positive and negative effects on the person's adaption and his relationships with health care providers.

Some patients adapt quickly to their new role of "expert" and this is particularly true for parents.[46–48] They can accumulate extensive knowledge of their disease through contact with professionals and peers. However, many continue to struggle with the

burden of being the expert and its impact on their relationships with health care providers.[27,48] Even once diagnosed and personal expertise is gained by the patient, many still report an unwillingness on the part of health care providers to *believe* them.[24] Constructive relationships are possible, but only when there is honesty about a lack of knowledge and willingness to partner.[22,42] Due to their new "expert" role, patients and families are often pressured into a state of constant self-advocacy, not only for their own diagnosis and treatment, but also for larger-scale changes in policy, research and health systems.[27,49] This can escalate to the point where individuals and families are feeling burdened by being in charge of their own care.

In many ways, the experiences of parents of children with rare disease mirror those of adults living with rare diseases themselves. The struggle with the uncertainty and unknowns around the disease remains paramount, with crystallization during encounters with health care providers.[23,24] Similar deficiencies in the health care system are noted by all patients and families, regardless if they are adult or pediatric.[50] Both parents and patients themselves evoke coping strategies such as building their own expertise, advocating for themselves and seeking support from peers.

In some ways, parents experience some aspects of the rare disease journey with more intensity. They can thrive in their role as experts, but struggle with the lack of information and connections that would allow them to execute this role effectively.[38,42,50,51] Motivation towards finding a diagnosis is heightened in parents compared to adult patients.[29,50] The reasons for this include the strong desire from parents to create a future vision for their child and wanting to ensure they have "done everything they can" including exhausting any possibility of treatment or cure.[29] Since this future vision often

cannot be created with any accuracy, parents take a “one day at a time” approach and remain firmly rooted in the present.[50] Similar to their children, the needs of parents change over time particularly in regard to their priorities for their child and family and their emotional responses to their child’s condition.[52] Thus, the approach of the health care system to families of children with rare disease must be tailored to parent/child, stage of life, adjustment to illness and many personal factors that must be elucidated.

1.2. Rationale

Individuals with rare disease and their families clearly have unique aspects to their illness experience that impact their access to and satisfaction with health care. There is evidence that they are systemically disadvantaged in the health care system and that established models of care are inappropriate or inadequate in their circumstances. Health care professionals and families are being impelled to adopt new roles with different expectations in terms of responsibilities and communication. These changes are not yet being widely acknowledged or supported, with the result that accessing health care is a fundamental source of stress and struggle for patients with rare diseases. This situation is unacceptable and change is crucial; but first, evidence must be sought that will further delineate the current experience of patients and families. This investigation must not be limited to those with a specific diagnosis, but should be inclusive of those who remain undiagnosed, for they are at potentially even greater risk of these challenges and deficiencies.

This study will examine the experiences and perceptions of parents of children with very rare or undiagnosed conditions with respect to medical care. All of these parents will

have waited years for a diagnosis for their child, and for some, this was never obtained. Important topics will be explored, such as roles and relationships, communication between families and health care providers and information sharing. This study will include a specific focus on the relationships between patients/families and physicians, since previous work has identified challenges in this area[20,27,42,53,54]. In addition, important patient health benefits are correlated to improving relationships and communication with physicians.[32,55] This study will form part of the greater evidence base that is needed to develop new health service delivery models that will better serve the needs of patients and families living with rare and undiagnosed diseases.

1.3. Context

There are two important contextual considerations that are necessary for true understanding and critique of this study – related to the professional identity of the researcher and the larger research program.

1.3.1. Context of the Study

This study was part of a larger research program, the Treatable Intellectual Disability Endeavour of British Columbia (TIDE-BC), aimed at the identification of medical conditions associated with intellectual disability, to enable prevention and treatment. This program was composed of numerous subprojects, some of which were intended to focus on care delivery and patient experience. The research topic of this thesis grew out of one of these subprojects, a proof of concept study investigating the use of an online health communication platform (My Care Web) to aide communication and collaboration in the care of children with complex needs (hereafter known as “My Care Web study”). I

was also the Principal Investigator of this study, as it was originally intended to be the thesis project of my PhD studies.

In the transfer from a PhD to MSc graduate studies program, I pared down the scope of the project to its current form. The interviews that formed the basis for this thesis study were originally embedded within the protocol for the My Care Web study. The preliminary literature review and topic exploration revealed a knowledge gap regarding the experience of care for children with very rare or undiagnosed conditions which subsequently became the topic for my MSc thesis. The original larger study was continued, but this research question was separated and a parallel study design was constructed (hereafter known as "thesis study"). The analysis of the baseline interviews, in particular, was distinctly separated from the remainder of the study.

A qualitative approach became the focus for this thesis study. The intensive experience of these parents and their children in the health care system gave rise to a rich volume of data. Qualitative analysis facilitates the discovery of patterns and relationships among these complex themes. In addition, research questions can be pursued in a flexible manner using an inductive rather than deductive approach.[56] By virtue of their rare conditions, these patients cannot easily be grouped or even identified at a population level or using large datasets.[2] Thus qualitative analysis involving multiple individuals with the shared experience of being "very rare" has the highest likelihood of identifying those common themes, struggles, experiences and perspectives.

1.3.2. Context of the Researcher

An important consideration throughout this study is my personal identity as a practicing physician, with a particular clinical interest in children with rare and complex conditions. None of the children enrolled in the study were known to me in a clinical context, nor did I provide medical care to them at any point. I disclosed my status as a physician to all participants prior to the interview. This may have introduced some bias; for instance, the participants may have been reluctant to disclose negative thoughts or impressions about physicians involved in their child's care. I maintained a high degree of awareness and caution, particularly during conduct and analysis of the interviews, not to allow my medical training and experience to override or unduly influence the data I was obtaining from the participants. I would argue that my clinical background is also a potential benefit to this study, as I can place the parents' comments in a detailed and knowledgeable context. It is also possible that the participants saw me in a position of expertise and trust, more so than if I was not a physician.

CHAPTER TWO: METHODOLOGY

This chapter will outline the methodology of this study, addressing the aim of the research, participant selection, data collection and analysis. It will also address some important ethical considerations in the conduct of the study.

2.1. Aim of the Research

To analyze the experience of parents of children with very rare or undiagnosed conditions (involving neurodevelopmental and physical manifestations), with a focus on communication and relationships:

a) Between families and physicians
b) Among health care providers involved in the child's care

2.2 Participant Selection and Recruitment

Potential participants for the My Care Web study, and thus this thesis study, were identified through the TIDE-BC Complex Diagnostic Clinic (CDC). This clinic was created to enhance diagnostic success in patients with "complex intellectual disability" (as defined below) through multispecialty evaluation. Children and youth referred to the CDC met the following criteria:

a) Global developmental delay / intellectual disability of unknown cause after implementation of the TIDE protocol (a series of recommended genetic and metabolic investigations)
b) Other systemic, neurological, psychiatric-behavioural features and/or biochemical abnormalities

c) Potential benefit from multispecialty evaluation

The researcher approached all parents or guardians (hereafter known as “parents”) whose children were assessed at the CDC from 2011-2013 and who consented to be contacted for future studies. In addition to the criteria for CDC referral mentioned above, the child was also required to have ongoing care from at least three health care providers. Exclusion criteria included lack of access to a computer or the internet and inability to communicate fully in English.

First contact was made by mailed invitation letter sent from a familiar clinician. Subsequent telephone follow-up provided more detailed information on the study and reviewed the consent form. Study participants consented to the full My Care Web study, including the one year online intervention, in addition to the interviews. Since the segmentation of this study was not undertaken until after the consent process was complete, there was not an opportunity to involve participants who may not have wanted to participate in the intervention portion of the My Care Web study.

In total, 29 families were approached to participate in the My Care Web study. Ten consented to participate, twelve declined for a variety of reasons (see below) and seven were unable to be contacted either due to inaccurate contact information (1) or failure to reply after multiple attempts (6).

For those who declined, the reasons given were as follows:

- Time constraints (4).
- No reason given (1).
- Child too unwell (1).

- Language barrier (1).
- Satisfied with current health care, no need for intervention (2).

The children of the families who declined were varied in terms of their age, home location and diagnosed status, with no apparent differences to those who consented to participate.

2.2.1. Participants

Ten participants were enrolled in the study – eight mothers, one father and one mother-father dyad (who participated in the interview together). Seven were from the Lower Mainland, the most highly populated region of British Columbia lying within 150km of Metro Vancouver and the provincial tertiary care hospital. The other three participants were from the same major regional city with a population size of about 100,000 people and a distance of about 400km from Metro Vancouver. This distribution likely reflects the referral pattern to the CDC, as subspecialists who made these referrals also had outreach clinics in the regional city. The affected children ranged in age from three to fourteen years. Two had confirmed diagnosis, four had provisional diagnoses and four had no diagnoses.

Specifics regarding the family location and the exact diagnoses for the children are withheld to protect their confidentiality as this information may be suffice to reveal their identity.

Table 2.1. Study Participants

No.	Age of Child (Years)	Mother or Father	Location	Diagnosis (Confirmed, Provisional, None)	Disease Prevalence (if known)	Clinical Description	Autism (Y/N)
1	14.2	Mother	Lower Mainland	Confirmed	1:100,000	Metabolic disease, intellectual disability	N
2	3.7	Mother	Lower Mainland	Confirmed	1:100,000	Metabolic disease, mild global developmental delay	N
3	7.8	Mother	Lower Mainland	None	N/A	Severe neurodevelopmental delay, epilepsy, cortical visual impairment, gastrostomy tube feeds	N
4	8.0	Mother	Lower Mainland	Provisional	Unknown	Intellectual disability, short stature, chronic vomiting, epilepsy	Y
5	5.7	Both	Lower Mainland	None	N/A	Intellectual disability, motor impairment, arthrogryposis, gastroesophageal reflux disease	N
6	4.8	Mother	Regional city	Provisional	1:22,000	Global developmental delay, hearing impairment, epilepsy, hypertension	Y
7	5.1	Father	Regional city	Provisional	1:12,000	Intellectual disability, hypoplasia of corpus callosum	Y
8	3.7	Mother	Lower Mainland	Provisional	<1:100,000	Global developmental delay, ataxia, developmental regression	Y
9	5.0	Mother	Lower Mainland	None	N/A	Intellectual disability, prematurity, visual impairment	Y
10	6.8	Mother	Regional city	None	N/A	Severe neurodevelopmental delay, epilepsy, cortical visual impairment, gastrostomy tube feeds	N

2.3. Interviews

Data was obtained from interviews with study participants with methodology outlined below and explicit description of ethical considerations.

2.3.1. Ethical considerations

One important consideration in the ethical conduct of this study is the distress that can come from relaying a difficult experience such as the diagnosis of a child with a serious condition. Study participants were cautioned about this risk. However, most literature suggests that parents tend to perceive the benefits of participation in this type of research outweigh the distress.[57,58] During the conduct of the interviews, the interviewer was very cognizant of the potential impact of questions on the participant. The open-ended structure of most questions allowed the participant to reveal only as much information as they felt comfortable. If negative emotions were expressed, the interviewer offered to take a break or defer certain topics in the interview. Participants were given contact information for members of the research team and encouraged to reach out if they needed emotional support after the interview. This did not occur, but if it had, the research team would have connected the participant with psychological support.

2.3.2. Conduct of the Interviews

Semi-structured interviews were conducted at a location of the participant's choosing: four in the home, two at BC Children's Hospital and four by telephone. The intention of the interviews was to elicit information about specific topics related to the study aims, while allowing flexibility for participants to deviate into other areas of their own priority. The last component of the interview also directed participants to comment on their comfort with technology and thoughts about security of online information, related to the My Care Web study. The interviews were completed prior to any detailed information or

training given about the My Care Web intervention. Interviews were chosen as a data collection modality for their ability to generate meaning and theory out of subjective experience.[59] In addition, interviews allow for full and dynamic exploration of the individual experience.[60] See Semi-structured Interview Framework in **Appendix A**.

The researcher began the interviews using a framework outlining the main topics of focus, based on the predetermined research aims. There were four main sections:

a) Introduction and context
b) Experience of living with a chronic disease
    - Focus on the individual narrative and identification of stressors and coping strategies
c) Communication and the health care team
    - Focus on access, communication strategies, relationships and connections
d) Technology profile
    - Focus on current use of technology and perspective on security

The interviews were constructed on open ended questions, allowing for free discourse from the participants and flexibility to direct the interview towards the most relevant topics. Directed questions were used to elicit comments on specific topics that were common to all participants, if those topics did not naturally arise. The researcher used language that was easily understood, but also attempted to match the participants' terminology as these parents had extensive experience in the health care system and significant knowledge around their child's condition.

As part of communication interview component (Section C), participants were coached to diagram the connection of health care providers with their child/family and with each other through a technique known as care mapping. This is a grassroots technique developed and used by parents to describe the systems and providers involved in the care of their child with complex needs. Although not a tested scientific methodology, this approach is useful in guiding parents through the comprehensive health, educational and developmental services that interface with their child and family.[61] Care mapping offers participants a tool to express their thoughts in a way that is meaningful to many parents of children with complex conditions. For these interviews, care mapping assisted with conceptualization of the relationships between the wide variety of individual health care providers and services. The exercise often gave rise to a novel reflection from the participant or a new trajectory of discussion. A sample care map can be viewed in **Appendix B**.

All interview data carry a risk of deviation due to researcher bias. This risk could have potentially been heightened by assumptions and preconceptions from my medical background. In addition, the education and practice of medicine carry a very specific and prescriptive approach to interviewing that is highly distinct from that required for qualitative interviews. Performing interviews as a qualitative researcher was a marked deviation from years of training as a physician. I accomplished this and mitigated bias through mentorship with an experienced qualitative researcher (McKellin) and thoughtful, deliberate conduct of the interviews. This risk of bias must be balanced with the value and appropriateness of the interview methodology, as outlined previously.

2.4. Data collection

Interviews took place from Jan. 2013 – Jan. 2014, in coordination with recruitment for the My Care Web study. The extended period was related to amendments required in the My Care Web study and resulting delays. The first interview was conducted in Jan. 2013 and the other nine took place between Oct. 2013 and Jan. 2014. Throughout the interview period, an inductive process was adopted in the interview framework, to allow for refinement of questions and inclusion of additional avenues of inquiry[59]. Interviews were audio-recorded and transcribed verbatim. These recordings and transcripts formed the basis of the study data, in addition to field notes and observations that were recorded by the interviewer.

2.5 Ethical considerations

Ethics approval was obtained by the UBC Children's and Women's Health Centre of BC Clinical Research Ethics Board. This submission was in conjunction with the larger My Care Web study. Informed consent was collected from all participants as described above. With respect to the interviews, participants were advised that they may refrain from answering any question. Prior to the interview, they were given information about the interview purpose, content and process as well as initial information on the My Care Web study, to which they were also contenting to participate. All identifying data were removed from the interview transcripts, including names of parents, children and health care providers. In many cases, this also included the specific gene or diagnosis that was ascribed or suggested, as with ultra-rare conditions this alone could be enough to identify the child. Any data retaining identifiers, such as original recordings and the link

between study ID number and participant identifiers, were stored electronically in a secure folder on the Child and Family Research Institute (CFRI) server with hard copies stored in a locked cabinet in a CFRI office.

2.6. Data analysis

The qualitative methodology used for this study was discourse analysis (DA). In its simplest definition, DA is the study of language above the level of the sentence. This translates to a broader study of language in context as opposed to phrases examined in isolation. In particular, DA examines the complex and inseparable relationships between language, action, knowledge and situation. DA recognizes that language is action and has widespread influence both on the participants in the discourse and on society as a whole.[62,63] This crucial recognition of the importance of contextualized language makes DA the most appropriate methodology for this study. As an illustration, DA infers that language can be used to enact and sustain social roles. Roles definition and communication are widely acknowledged to be key factors in establishing relationships between patients and health care providers.[55,64,65] In this study, DA provides the ability to analyze the broader meaning, purpose and function of language and the "actions" that accompany – role development, communication, interaction, collaboration.

Implementation of the DA methodology in this analysis was based on an approach described by Gee (2011).[62] Within the interview data, four tools of inquiry were used to analyze seven building tasks. Tools of inquiry included social languages (different styles of language used for different purposes), discourses (the integration of language and non-language elements to enact a particular identity), conversations (how our words

relate to themes or debates in broader society) and intertextuality (how our words relate to the words of others). These were used to analyze building tasks that describe how language is used as action. These building tasks include: significance, practices, identities, relationships, politics, connections and sign systems. For instance, to address the first building task would be to ask the question “How is this piece of language being used to make certain things significant, or not?” An example of this application would be a parent using the social language of medical jargon to enact their identity as an expert parent.

During the course of this examination, codes were created and applied to the interview transcripts inductively, with the intention that these codes arise out of the data itself so that preconceived theories and opinions of the researcher did not have undue effect.[66] Once initial inductive codes were created and after full review of all ten transcripts, these codes were refined to be discrete and mutually exclusive through second and third reviews of all transcripts. These final codes were organized into a codebook containing their definitions, inclusion/exclusion criteria and illustrative examples. Codes were then grouped into themes and the relationships between the themes were constructed, with frequent return to the original transcripts and revision. Other components of data were referenced and reviewed throughout this process to enhance and verify the analysis. In particular, the original audio recordings were reviewed so that elements of paralanguage (voice quality, intonation, tempo, pauses, etc.) could be included in the analysis. Likewise, field notes and care maps were often reviewed in the categorization and identification of themes.

I undertook a number of means to optimize the trustworthiness and rigor of my results. This began with an explicit, thoughtful and ongoing examination of the biases that could be anticipated to influence the results, namely those introduced by my professional identity as a physician and by the sampling selection through which I composed my study population. Throughout examination of my data, I kept a clear audit trail tracking the evolution of my analysis with particular focus on decision points or deviations taking me in one direction versus another. I actively considered, interrogated and recorded alternative themes and theories to explain phenomena through comparison of different perspectives. Finally, I intend to seek feedback and validation from the participants themselves through one-on-one follow-up discussions in the near future.

CHAPTER THREE: RESULTS

This chapter will begin with a brief description of each family to allow for contextualization of each participant's perspectives and to enable deeper analysis of the themes. The key themes that emerged from the data will then be described with emphasis on the variations within the study population and the potential relationships between these variations and individual characteristics related to the family and their diagnostic journey.

Family Summaries

Family #1: Two daughters in this family have a type of organic academia, a metabolic disease that is genetically inherited (prevalence about 1:100,000). The disease presents early in life with characteristic symptoms and laboratory findings. It is associated with neurodevelopmental impairment and can impact many organ systems. Management is based on dietary modifications and nutritional supplementation. There is a high risk of early mortality between 7-12%.[67] The first child in the family was diagnosed at five months of age, shortly after symptoms developed. The second child was diagnosed prenatally. Both daughters developed intellectual disability and cardiomyopathy. Unfortunately, the younger daughter had progressive cardiac disease and died at age 9 years, two years prior to the study. The older daughter was fourteen years old and doing well at the time of the interview. The medical care for both children was centralized in the biochemical diseases clinic at BC Children's Hospital. There were two other younger children in the family who were unaffected – a five year old son and a two year old

daughter. Parents were married and of Filipino descent, but born in Canada. Both parents graduated from high school. They had a large extended family living in the area.

Family #2: The three year old son had a metabolic disease that was genetically inherited (prevalence about 1:100,000).The disease presents early in life with liver enlargement, growth failure, low blood sugar and developmental delay. Management is based on dietary modification and prevention of hypoglycemia. Prognosis is generally good.[68] Concerns arose around eight months of age and the child was diagnosed at age two years. The first specialist referral (to a pediatrician) did not address the concerns. A second opinion was sought that led to additional referrals to two subspecialty services and plans for more invasive testing. The mother eventually identified the correct diagnosis through online research and sought the opinion of a subspecialist in the United States who confirmed the diagnosis. Ongoing management was taking place through both the Canadian and American biochemical diseases subspecialty services, the latter mainly through email communication. The child was doing well at the time of the study, but manifesting mild developmental delays. There were no other children in the family. Parents were married and of European descent, but born in Canada. Both parents had additional diploma training beyond high school and both worked in a health care related field. They had extended family support.

Family #3: The seven year old son was undiagnosed and had severe neurodevelopmental impairment involving all developmental domains, epilepsy, visual impairment and gastrostomy tube feeding. His seizures were difficult to control and had resulted in multiple emergency room visits and hospitalizations. Diagnostic investigations regarding the etiology of his condition took place mainly early in life and

were re-examined on referral to the CDC. There was one younger son (age 4 years) who was well. Parents were married and of Middle Eastern descent. They were first generation immigrants to Canada but had no language barriers. Both parents had a high school education. Father was working full-time and mother was staying home. Their situation was characterized by social isolation from their extended family and community, as a result of the stigma associated with their son's condition.

Family #4: The eight year old son had autism, moderate intellectual disability, history of seizures, unexplained vomiting and short stature. A preliminary diagnosis for his condition had been proposed, but the mother did not know the details. Concerns arose at one month of age, but most of his initial diagnostic work-up took place during a hospital admission at age 6 years, when he presented with seizures. His preliminary diagnosis was reached shortly thereafter but was not associated with any specific treatments. Earlier on, the mother felt that her concerns were not being addressed appropriately by their pediatrician. There were two younger boys in the family, both well, who were age seven years and eight months. The mother mentioned that she was nineteen years old when she had her first child and felt that her age impacted how physicians interacted with her. In particular, they would tend to speak to the maternal grandmother, who was often present at appointments. The mother was currently in a common-law relationship with the father of the youngest child. There was no mention of the older boys' father. The mother had a high school education and was of European descent, but born in Canada. They had extended family support in the area.

Family #5: A five year old daughter was undiagnosed and had moderate intellectual disability and motor impairments, ataxia and unexplained vomiting. Concerns arose

prenatally with an abnormal ultrasound result that led to additional prenatal testing and subspecialty referral. After preliminary testing, parents elected not to proceed with invasive testing or termination. At birth, their daughter had respiratory symptoms, limb contractures, lethargy and feeding difficulties. She was transported to the tertiary neonatal intensive care unit to undergo investigations. Diagnoses were suggested but not confirmed. Over the ensuing years, concerns including developmental delay, abnormal movements and vomiting led to presentation to medical care and ongoing investigations. There was one older son, age eleven years, who was healthy. Both parents participated in the interview. They were married. They were originally from India and both received doctoral degrees in the same health care field. Initially they immigrated to the United States. In Canada, they originally located in a small city (population 35,000) in British Columbia, where their daughter was born. When the daughter was around one year of age, they moved to New Brunswick for the father's work. They subsequently moved back to BC, to Metro Vancouver, about a year later. One of their stated motivations for the final move was to be closer to subspecialty care for their daughter. Both parents were working full-time, but not in their field of training.

Family #6: A four year old daughter had autism, moderate intellectual disability, epilepsy and hearing impairment. She had received a preliminary diagnosis of an intrauterine infection as being the underlying etiology (prevalence 1:22,000).[69] There is no specific treatment for this condition. Concerns began at five months of age with onset of seizures. Diagnostic investigations were conducted through their pediatrician and a subspecialty service. The eventual diagnosis reached was first suggested by the mother more than one year earlier but dismissed by the pediatrician. This diagnosis was again

suggested at the CDC clinic during the child's assessment at age 4 years. Confirmation was pending, awaiting further investigations. There was one younger daughter in the family, aged two years, who was healthy. Parents were married, had high school education and were of European descent (born in Canada). Mother stayed home full-time and father was self-employed in the family business. They had extended family supports in the area.

Family #7: A five year old daughter had autism and moderate intellectual disability. A preliminary diagnosis was assigned – a genetic condition with a prevalence of 1:12,000-1:24,000). Some cases are inherited, but most arise de novo.[70] There is no specific treatment for this condition. Manifestations can include epilepsy, intellectual disability, motor impairments, behavioural difficulties and feeding issues. Concerns first arose at five months of age with eye abnormalities and developmental delay. This family also sought a second opinion after their first pediatrician did not adequately address their concerns. They experienced early difficulty accessing subspecialty care, in her first two years of life. The preliminary diagnosis was proposed at the CDC assessment at age 4 years. Confirmation was pending, awaiting further investigations. There was one younger adopted daughter in the family, aged one year, who was healthy. They decided to adopt their second child due to concerns about the potential recurrence risk of their first daughter's condition. Parents were married. Their second child was not biologically related to them. Both parents had additional education after secondary school. Father worked in information technology at a health care facility. They had extended family supports in the area. Parents were of European descent, born in Canada, and their second daughter was Chinese.

Family #8: A three year old son had ataxia, intellectual disability, autism and developmental regression. A preliminary diagnosis was assigned – a genetic condition with a prevalence of 1:100,000. Most cases are inherited, but some arise de novo. Manifestations are characterized by attacks of ataxia, vertigo, headache and nausea.[71] There is medical therapy thought to decrease risk of recurrent episodes. Concerns first arose at age ten months of age with an episode of altered level of consciousness and developmental regression. Hospitalization occurred and the event was diagnosed as acute cerebellitis, recurrence was not anticipated. However, the event recurred at age seventeen months, leading to another hospitalization and further investigation. The mother initially proposed the diagnosis during that hospitalization and he was initiated on the appropriate therapy. However, the mother reported that both she and the child's neurologist did not feel this diagnosis is accurate, which prompted the referral to the CDC clinic at age three years. There was one older daughter in the family, age twelve years, who was well. The children had different fathers. The mother was married to the boy's father, but they were in the process of separating. Both parents had a high school education and were of European ancestry, born in Canada. They had extended family supports in the area, particularly on the father's side.

Family #9: A five year old son had a history of premature birth, autism, visual impairment and moderate intellectual disability and was not diagnosed. He was born at 32 weeks gestation and was initially hospitalized in the neonatal intensive care unit for two months. He was followed by a pediatrician and developmental supports since hospital discharge. The pediatrician spoke the parents' first language. By one year of age, developmental delays were noted. It was unclear at what stage this was felt

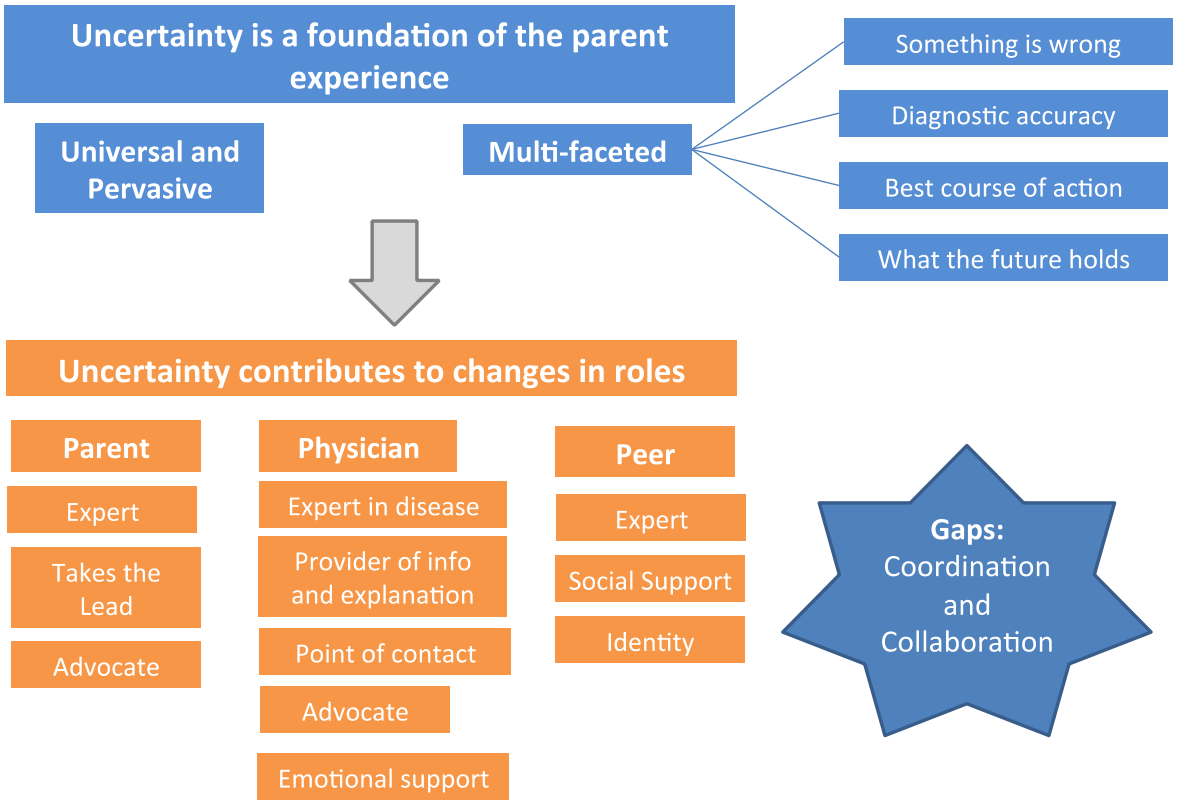
inconsistent with his prematurity. Additional investigations began around age two years with CDC assessment at age three years. There were no other children in the family. Parents were married and immigrated to Canada from China. English was a second language to both parents and language was a barrier to some degree during the interview. Both parents had a high school education; father went on to post-secondary studies as well. Father worked in the information technology field and mother worked part-time at a community child care facility. Extended family all lived in China. They had a limited social network, based mainly on the mother's work relationships.

Family #10: A six year old son had severe neurodevelopmental impairment in all domains affected, epilepsy, visual impairment and gastrostomy tube feeds and was not diagnosed. Concerns arose at nine months of age related to vision and development. Diagnostic investigations were carried out at that time (in the first one to two years of life), mainly organized by one subspecialty service. This resumed on referral to the CDC clinic at age six years, initiated by the same subspecialist. He had presented to the emergency room and been hospitalized multiple times due to seizures. There was one older daughter, age eleven years, who was healthy. Parents were married. Mother stayed home and father worked full-time. They both had a high school education and were of European descent, but born in Canada. They had extended family supports in the area.

Overview of Themes

One key theme that emerged from the data was the pervasive and multi-faceted nature of uncertainty and its impact on the parents' experience of care and coping with their child's illness. This appeared to lead to deviations from the traditional role structures and expectations between parents and physicians with apparent gaps in the distribution of these roles and responsibilities. Although the interview framework and content was designed to be more general, many participants focused on the medical aspects of care and their interactions with physicians. Possible explanations will be reviewed in the discussion section.

Figure 3.1. Overview of Themes



3.1. Theme One: Uncertainty is a foundation of the illness experience for parents of children with rare and undiagnosed conditions

Uncertainty formed a foundation of illness experience for these parents. This was a universal, persistent and complex phenomenon as described by the participants.

3.1.1. Uncertainty is universal and pervasive

Uncertainty was central to the illness experience for parents in this study who frequently reported stress related to a lack of answers and information regarding their child's condition. This held true for children with no diagnoses, provisional diagnoses and confirmed diagnoses. The commonality of this experience among diagnosed and undiagnosed families was likely related to the source population. A diagnosis identified through the CDC would be expected to be at the extreme end of the spectrum of rarity (hereafter dubbed "ultra-rare"). Since information about the manifestations, course and treatment of diseases is often derived from large scale studies involving many affected individuals, the information available on ultra-rare conditions is often scant, making their experience similar to those who are undiagnosed.

Only one parent (#1) did not report significant distress related to uncertainty. Her unique experience may be related to the early timing of diagnosis. This family avoided the diagnostic odyssey experienced by the other families, as their first daughter was diagnosed very shortly after her symptoms developed. Her diagnosis also connected them immediately to a subspecialty service with expertise in the condition. Although unknowns certainly still existed, as for other patients with rare disease, these factors

may have contributed to more information about the condition, more confidence and less distress surrounding uncertainties, diagnostic and otherwise.

Although other parents were concerned about the lack of disease-specific information, they did not expect the assignment of a diagnosis to impact their child meaningfully in terms of service eligibility or treatment options. The reason for this expectation was not explicitly stated, but may have come from communication with physicians or their own independent research. For one mother (#10), this lack of perceived utility of an eventual diagnosis negatively influenced her desire for testing. This family also had already been living without a diagnosis for many years.

> *"I mean the longer we go without a diagnosis the more….I don't think it matters because whatever he's diagnosed with isn't something that's going to change his life…"*
>
> *(Participant 10)*

Even still, the parents of children who were undiagnosed still invariably displayed some motivation to find a diagnosis for their child. Some parents felt that having a name for their child's condition could help them to cope with the reality. One mother in particular (#2) felt that remaining undiagnosed would be intolerable. However, her child was diagnosed relatively early in life and she did not experience the same extended diagnostic journey as many of the participants. For other families, the passage of time may have muted their focus on a diagnosis.

> *"I remember a nurse said to us 'you know you might never know, there's a lot of parents out there that never get an answer or a reason or anything.' I think that would just have been torture not to know, you know what I mean?...I guess that's*

what everyone says right, if they could have an answer for why their knee hurts they feel better to say it's not in their head kind of thing."

(Participant 2)

Parents did not expect a diagnosis to erase all uncertainty. In fact, the stress associated with their unanswered questions was as bothersome to many of the parents of the diagnosed children, as to those who were undiagnosed. However, a diagnosis was thought to help manage the complex and multi-faceted set of unknowns that these parents face.[16]

3.1.2. Uncertainty is multi-faceted

This study uncovered multiple components to uncertainty, many of which are not fully appeased or alleviated by a diagnosis.

"Something is wrong," Uncertainty about disease manifestations – Uncertainty was experienced from the very beginning for these parents. Many described early experiences when they knew something was wrong with their child, but felt that their concerns were not being addressed. This phenomenon related to many factors including the episodic or subjective nature of the presenting symptoms, a lack of physician knowledge about a potential rare condition or difficulty accessing a physician who had the appropriate expertise. Early on, the concerns were often related to developmental differences or episodic symptoms that were difficult to describe. Garnering understanding and support from their front line physicians proved challenging for some parents.

"…She was so upset and crying and I couldn't calm her…she would make these noises and they would go on and on…and when her head turned to one side she barely moved, she just laid there. So I was thinking maybe there was some kind of epilepsy, something going wrong…I went to the doctor many times, took pictures, you know, what is happening…I kept going back to the doctor saying there's something wrong…"

(Participant 5)

"I noticed he was doing some weird things with his hands like he would start shaking them and he would have some weird blank stare in his eyes…That's the first reason I thought something was wrong with him because I took him to the doctor and I told, I basically expressed my concerns to him…I was just told that babies sometimes shake when they get scared so it was kind of brushed off…"

(Participant 4)

These interactions with physicians intensified the parental uncertainty around the presence, nature and cause of the signs and symptoms they were observing. One parent (#2) who was seeking medical care for her child's growth and developmental regression did not feel these concerns were adequately addressed, which in turn made her doubtful of their significance.

"…because he just told us someone had to be small and not everybody can be big…we hit like sort of a roadblock and thought, okay, well I guess our kid is just supposed to be little. And then we went a few more months and just something wasn't right."

(Participant 2)

Not surprisingly, these perspectives were more common when dealing with primary care physicians and community pediatricians than with subspecialists.

There were some exceptions. In particular, for family #1, because their first daughter was diagnosed in the setting of an acute metabolic crisis and their second was diagnosed prenatally, they did not have unaddressed concerns related to diagnosis or early stages of disease. The children in families #3 and #9 had extended stays in the neonatal intensive care unit, leading to early alliance with specialists and subspecialists. In a sense, these parents may have been already expecting their children to deviate from the typical course in terms of their development. Overall, these two families were less focused on the topic of uncertainty, which may have been impacted by these early alliances and shared understanding with professionals, in addition to other factors.

Uncertainty about the accuracy of the diagnosis – Most children had received a provisional or "working" diagnosis at some point in their lives. Even more commonly, a test result had been positive or suggestive of a particular condition, but inconclusive. When provisional diagnoses were ascribed, parents maintained skepticism about their accuracy. This lingering uncertainty resulted in a diagnostic journey that was officially concluded, but not necessarily complete. A lack of clarity in the communication between physicians and parents also contributed to this persistent uncertainty. Understanding complex test results and interpretations were challenging to some of the parents.

> *"…I don't remember what it is that he has, we have so many different, he's not really diagnosed yet. So I don't really know anything about his diagnosis…one of them I know is [diagnosis]…something to do with I guess his DNA."*
>
> *(Participant 4)*

> *"…we find out he's missing half of a chromosome on his DNA…and so they also test me and [father]. [Father] is also missing that same half chromosome, so that's now the diagnosis right?"*
>
> *(Participant 9)*

In some cases, the parents perceptively realized that their child's physicians were also uncertain and demonstrated a sophisticated understanding of the complexities involved in diagnosis. Not surprisingly, parents with higher levels of education and who spoke English fluently were more likely to have a clear understanding of the results of these diagnostic tests and assessments.

> *"…so we attended the TIDE clinic…and we were reviewed by the panel and they came up with the [diagnosis], that she met the clinical presentation for [diagnosis]. And I understand there's some dissention from the people about that, but that seems to be the consensus of opinion."*
>
> *(Participant 7)*

The two parents (#1 and #2) who displayed confidence in their child's diagnosis were also those with treatable metabolic conditions. In addition, both of their children had been diagnosed at a relatively early age (before age two years). Thus, they hadn't received other preliminary diagnoses that had gone on to be disproven.

Uncertainty about the best course of action – Decision-making about treatment and management plans for children with undiagnosed conditions is complex and filled with many unknowns. This situation is similar for children with rare conditions due to their small numbers and lack of rigorous clinical trials. Even parents of children with treatable metabolic diseases expressed significant distress over the lack of clarity around treatment decisions. Family #2 expressed frustration at the lack of guidance regarding

dietary management and ongoing surveillance with respect to her child's condition. This is a good example of a condition that, although diagnosed, doesn't have clear, evidence-based management protocols.

> *"A lot of it is trial and error….we kind of, we kind of make it up. I don't think that's really a great way to go about your kid's health…"*
>
> *(Participant 2)*

For the other family with the metabolic disease (#1), the course of action was clear until the disease became too severe to control, as in the case of their second daughter. This concerned the family as it seemed that at some point there would be no further treatment options.

> *"…I felt like there's a brick wall in front of me and…we can't go any further with this…it's a big frustration for a family to feel like, you know, there's no other option for you."*
>
> *(Participant 1)*

For the other families who had no diagnosis or only a provisional diagnosis, this wasn't a main point of focus. As mentioned, none of them felt that their child's condition, if diagnosed, would be associated with a specific treatment or cure. They had already adopted an approach concentrated on addressing the manifestations or symptoms of their child's condition. They were however unsure of how to navigate some of the more practical or functional issues that will arise such as facilitating transition points and guiding entry into school or adult services.

Uncertainty about what the future holds – All parents exhibited concern regarding their child's unclear prognosis. Parents were unable to create a vision for their child's

future, leading to considerable distress. Father #7 clearly expressed both the desire to know the future and the understanding that this information is not available. They had received a provisional diagnosis for their daughter, but this uncertainty did not appear to be dependent upon its confirmation. He was of the opinion that the prognostic information they desired was, in its essence, unobtainable.

> *"They wisely didn't make a lot of predictions about what her future could be...of course we want to know what her future is going to be but nobody would tell us because, of course, nobody knows. But that is something as parents we desperately need to know. That's an unfilled need, nobody can fill it."*
>
> *(Participant 7)*

Indeed, one of the parents of a child with metabolic disease (#2) expressed a very similar perspective. This was the only point during the interview when this mother became tearful and obviously distressed.

> *"…it used to keep us up at night…just sort of what, what the future is…we just feel like we want to know where, what he future is for him and not just say oh he's going to be the smallest or oh he's going to be able to work as an accountant because he can't physically do the things that other kids and adults can do."*
>
> *(Participant 2)*

Other parents of undiagnosed children wondered about the potential degenerative nature of their child's condition, their developmental potential and even their life expectancy.

" So they're still trying to find out what went wrong or if it has deteriorated or if it is deteriorating…So for now…we don't [know] what is wrong and what does the future hold or what it will be like?"

(Participant 5)

Most parents of the undiagnosed children were hopeful that a diagnosis, even one that is ultra-rare, would give them information about prognosis.

"If you tell me [he has] a certain syndrome at least I can have some examples like, you know, people who have that syndrome before…at least I can know how ten years from now, what it's going to look like for [name] but right now I don't know. That's the most stressful right now."

(Participant 9)

These future concerns of some parents were entangled with worries about transition to adult care or the systems that exist to support adults with developmental disabilities.

[Discussing transition from school to adult services] "…So at this time I'm engaged in, I wouldn't call it a battle, but aggressive research about what's going to happen and how it's going to be handled and who's going to make the decisions and under what criteria…So it's a real, real challenge because I have to decide between uncertainties."

(Participant 7)

The distress related to uncertainty around prognosis was shared by nearly all parents, regardless of their child's age. Three families did not mention this as a concern, namely #1, #4 and #8 (children aged fourteen, eight and three years). There were no clear differences in this subgroup. For family #1, their experience with the death of the other child may have led them to live more "in the moment."

3.2. Theme Two: Uncertainty contributes to changes in roles

The persistent and complex uncertainty surrounding the care of children with ultra-rare or undiagnosed conditions leads to a shift away from the traditional roles occupied by parents and health care providers. Particularly for physicians, their role in the health care encounter hinges on their medical expertise and ability to offer treatment or, at the very least, counselling on the condition and its prognosis. This new role delineation has fundamentally shifted the responsibilities of parents and physicians leading to areas of overlap and encroachment into new territory. Descriptions of these themes will also highlight challenges and barriers when these new roles and responsibilities are enacted.

3.2.1 Parent Roles

The parent as expert - One major change in the roles between parents and physicians is in the distribution of expertise. Most parents in this study went to great lengths to educate themselves about their child's condition, whether or not a diagnosis had been given. Their research was largely aided by the internet and online social networks, but also by connecting independently with expert professionals. Most parents had a high degree of confidence in the knowledge they had acquired, in many cases finding their own expertise superior to that of their child's physicians. One parent (#5) of a child with

an undiagnosed condition was particularly poignant about the best sources of information and the concentration of expertise. This perspective was undoubtedly influenced by the parents' extensive training in the health care field.

> *"[discussing specialists] we understand their limitations, but they're also human right? They have many university degrees, but that doesn't mean they are any more educated necessarily right? But having said that sometimes we think we know more than them…it's our daughter so we explore so much online…There's more knowledge online than what they can tell us."*
>
> *(Participant 5)*

Even parents of the children with metabolic conditions who received expert subspecialty support felt similarly. In particular, mother #1 relied upon herself to stay up-to-date on scientific advances in relation to her daughter's condition. Her rationale, shared by many parents, is that professionals wouldn't be as motivated as parents ("this is my child"). In addition, the attention of professionals is split among a large number of patients and diseases, whereas the parents' singular focus is on their child and his/her condition.

Nearly all participants also emphasized the importance of knowing their child as an individual. This was particularly salient in the setting of an undiagnosed condition where this child-specific knowledge would form the foundation of the parent's expertise due to the lack of disease-specific information. Father #7, whose child had a provisional diagnosis, based his expertise on his child's communication and behaviours.

> *"She's patient and pleasant as a rule and if she's not, then there's something wrong and we pay attention to that…She has other gestures that are not official signs anyway, but I know what they mean, we know what they mean. And she has a couple of very minor vocalizations that she does and we know what they mean."*

(Participant 7)

A primary source of frustration for parents was to have their expertise in their child go unrecognized by health care providers. This was explicitly discussed by all but two participants, most commonly when interacting with physicians or school personnel. One parent (#10) described the challenge of working with unfamiliar physicians in the emergency department when her child presented with a prolonged seizure.

> *"…because they don't know him like I know him. They insist that we stay the night and, you know…I really don't think we need to…they don't know what his normal is…"*

(Participant 10)

Later, when describing the creation of a care plan for the school setting, she reflects:

> *"…you kind of get dismissed because you're not the professional. And yet you're the one that's been, you know, pretty much the expert at it for the last seven years. Sometimes you don't get listened to because you're just the mum…"*

(Participant 10)

This individualized knowledge helps to give parents the confidence to make decisions, often in the face of incomplete or non-existent information.

There were two parents who did not emphasize their expertise. Mother #3 in particular, expressed stress and frustration related to professionals expectations of her knowledge. She felt burdened by her obligation to report her child's symptoms, which she describes as subjective. This is in quite sharp contrast to most of the participants who felt that their expertise was under-recognized by professionals.

> *"They want at the beginning especially as residents and doctors on call and fellows are changing they have no clue so we have to start from the beginning so that's one issue I want to end. Second is they want to know what kind of seizure mum he's having, how is he seizing like they always want to ask the mum or the dad like the parent what is going on right now I want you to tell me based on what you're telling me I'm going to prescribe blah, blah, blah, blah, okay. Unless he has a fever when I take him and they can measure it, everything is almost like hearsay."*
>
> *(Participant 3)*

However, this same mother felt she had expertise in certain areas, for example she confidently describes her views and interventions around seating and mobility.

> *"Because I break a lot of rules [laughing]…and it's worked for me, you know. Certain seat belts I don't put on his arms so he can move and that's why he's very active."*
>
> *(Participant 3)*

The other parent who did not emphasize her expertise was in family #9. Her focus was on accessing written information and reports from the physicians, tapping into their expertise. Some of this distinction may have related to a language barrier limiting her ability to retain verbal information or conduct her own independent research. Cultural and personal factors may also be underlying.

The parent takes the lead - A number of parents took a leading role in the search for their child's diagnosis. In three cases, the accurate diagnosis was actually first suggested by the parent. Mother #2 was instrumental in researching a likely diagnosis

online and connecting with an international expert (based in the United States) for guidance regarding diagnostic testing. Mother #8 also discovered a likely diagnosis online and brought it to her child's neurologist for consideration. Mother #6 actually suggested the correct diagnosis to her child's physician years before it was pursued. These three parents came from a variety of backgrounds, with only one having additional post-secondary education. This level of parental involvement in the diagnostic pursuit was viewed differently by each individual, but seemed to create challenges in the physician-parent relationship. In particular, the division of responsibility for finding the diagnosis was not always clear and this contributed to difficulty in navigating this partnership.

At the extreme end of this involvement, some parents felt that the responsibility for finding a diagnosis for their child rested on their shoulders. Mother #8 expressed resentment towards her child's neurologist, implying that the physician was not working hard enough to find the diagnosis. This relates back to the division of responsibility as mentioned above.

> *"…so I found this disorder that just seemed to fit [child] and so I told the doctor about it, and then she rolled with it." On later reflection: "I thought how, how hard is she working if she took what I had and ran with it? It just bothered me a little bit."*
>
> *(Participant 8)*

Mother #5 describes a negative experience with a community physician, also related to who takes ownership in the search for a diagnosis. Although this family had the highest levels of parental education, as well as self-described motivation and capacity to

contribute to the diagnostic search, this perceived lack of engagement of the physicians was very distressing to them.

> *"…one doctor said to us…if you ever find out what [child] has, let me know and I got so mad, I'm like maybe you try to find out what's wrong with [child]….I have to…try and figure out what needs to be tested."*
>
> *(Participant 5)*

Other parents had more overt disagreement with physicians about the route to take with diagnostic testing. For Mother #2, seeking her child's diagnosis outside of the Canadian healthcare system created awkwardness and potentially conflict with their local subspecialist physician. This mother preferred to work with the American subspecialist, but due to logistical constraints, had to maintain a relationship with the Canadian subspecialist. Participant #6 had two experiences with her child's physician where she proposed a diagnosis, had the physician disagree and then had both diagnoses eventually confirmed (the congenital infection and autism spectrum disorder).

> *"…we just felt like any ideas we had were shot down and, of course, we don't, we're not medical professionals so we don't know anything, but just like we felt like he wasn't open to options. And again when I presented that I thought [child] might have autism he shot me down, he said no."*
>
> *(Participant 6)*

In the other families, this leadership role as it relates to diagnostics was not a point of focus. Mother #1 was very proactive about looking into treatment options, including those that were more experimental and not offered by her daughter's treating medical team. Mothers #3, #4, #9 and #10 were not actively pursuing a diagnosis through their own efforts. The families who had children with the most severe neurodevelopmental

impairment (#3 and #10) seemed more focused on the day-to-day needs of their child and family. Time constraints and other areas of focus could have contributed to their lack of motivation to take a leadership role in their child's diagnostic search. Also, for mothers #4 and #9, although they expressed interest in finding a diagnosis, they took direction from their child's physician about the nature of testing.

The parent as advocate- Many of the tasks and roles in which parents engage, including those described above, are related to advocacy. Parents in this study felt that it was up to them to garner the services, supports and even diagnostic testing that their children required. Mother #2 felt that her constant presence and advocacy was necessary to ensure that adequate care and exhaustive testing took place.

> *[Discussing diagnostic testing] "I guess I'm the sort of person who wasn't going to take no for an answer kind of thing. And I would hate to think that there was other people out there that would have taken we don't know as the answer and where he would be if we, if we hadn't pursued it."*
>
> *(Participant 2)*

This role was universally adopted by the parents in this study, however the focus of their advocacy efforts was variable. Mother #1 was actively pursuing a diagnosis of autism, with the hope of obtaining the associated respite and developmental funding. Mother #4 was advocating for a communication system to be available in the home, to allow better communication between child, parent and siblings. Families #5, 6, 7 and 9 were all approaching, engaging in or reflecting on school transitions. They were advocating across various settings and agencies to ensure that developmental and behavioural supports were in place.

Again, the families who had children with severe neurodevelopmental impairment differed to some degree, in particular neither saw herself as an advocate. Mother #3 reported personal struggles and burnout that may have also influenced the roles she adopted. Mother #10 explicitly denied the advocate role. This seemed to be in comparison to her peers and may have related to the efficacy with which she felt she enacted this role. However she clearly engaged in securing services, supports and funding for her son which may speak to different perceptions of advocacy between the mother and the interviewer.

> *"You know, there's all these worries and so yeah, sometimes you kind of just wish there would be it would be easier to find these answers than having to be I don't know like I mean I know from other mums or one in particular who is very you know she's a real advocate and she just knows everything about everything but that's her, her life is that but not everybody is like that not everybody like I'm not like that, I'm not one to go on line and figure out where everything is and phone and, you know, it's hard for me to kind of figure it out. I just decide, I just think, okay, well I'll just do it myself, you know, instead of trying to fight for things so…"* *(Participant 10)*

3.2.2. Physician roles

With parents adopting roles related to expertise and leadership, what is their perspective on the physician role? Parents identified roles for their child's physicians, outlined below, that they presented in both a positive and negative context. In some cases, relationships broke down when the parents didn't see a role for the physician or when their expectations weren't being met. Many parents were realistic that physicians, particularly front line physicians, lack knowledge and expertise about their child's specific condition. Some parents perceived the role of physicians as broader than simply the medical expert. Some strong physician-parent relationships were rooted in non-expert physician roles.

The physician as expert in disease- Only two participants (both of whom had children with metabolic conditions) emphasized the expertise of their child's main physician, in both cases a subspecialist.

> *"…this disease is so, you don't, they don't know much about it right? So that's why I feel Dr. X (American subspecialist) kind of is the, I would say the go to one and I mean he has over four hundred patients that he's actually physically seen and worked with…and so I sort of feel like he's kind of at the top with the most knowledge…"*
>
> *(Participant 2)*

All parents in the study acknowledged some useful expertise in their child's physicians, particularly subspecialists, but were often realistic about the limitations of their knowledge. Even for the two participants mentioned above, the physician was not

considered the sole, or even main, source of information. Information obtained from independent research and interactions with peers remained highly prioritized.

For the parents whose children were undiagnosed, the physicians' expertise was limited by an unclear etiology for the conditions. Physician education remains largely organized around the investigation and management of specific diseases and disorders.

Physician as provider of information and explanation- Even outside of the context of diagnosis-specific expertise, the ability to transmit, explain and contextualize information was uniformly valued in their child's physicians. Some of the parents desired high levels of disclosure of their child's medical information, particularly families #2 and #9. In family #2, this could be related to the mother's occupational background in health care and her past experience being unaware of abnormal test results.

The language barrier of mother #9 may have made written copies of her child's information more important for reference and sharing purposes. She saw her child's community pediatrician as the one who can receive, compile, translate and explain her son's assessments and test results. However, in many ways she was unsatisfied with this role distribution and expressed a desire for more direct access to this information.

> *"I go see Dr. X, he's my pediatrician. So I guess all the information goes back to him and then he's my main resource. I would just go back to him and he would check his computer…I never see the report because they all go back to Dr. X."*
>
> *(Participant 9)*

In the current system, medical information is delivered primarily through physicians. This creates a situation where physicians can be viewed as either barriers or conduits to information particularly when families desire the “raw data.”

Some families found that the contextual information and explanation provided by physicians to be inadequate. Father #7 desired both more specific details and more explanation from his child’s physicians. He is highly educated, but not in a medical field, and for him more information may have assisted with coping and increased his knowledge of his child’s condition.

> *“…she had an EEG and a CAT scan. And those revealed…some structural abnormalities in her brain. And at the time I did not feel that I was told what those structural abnormalities might have been. So to give you the exact words, they said that her corpus callosum was thin…Now I, I know what those structures are but what I didn’t know and what I wasn’t able to find out for several years was how thin her corpus callosum really was. So, for example, you know, was it thin like saran wrap or…like it’s ten percent thinner than what you might expect? And I had no idea where she was on that scale for a long time. So I was unable to evaluate that piece of information and I found that quite frustrating.”*
>
> *(Participant 7)*

Physician as point of contact- Parents desired a point of contact with their child’s physician. Accessibility and responsiveness were two of the most frequent factors mentioned when describing satisfaction or dissatisfaction with physician relationships. In those with a strong connection to their family physician, families #4, 6 and 7, easy accessibility was a consistent feature of that relationship. Logistical factors, such as difficulty getting an appointment, not having an inquiry addressed in a timely manner or

needing to secure a re-referral before an appointment, were related to statements of dissatisfaction particularly with respect to front line physicians (family physicians and community pediatricians) but also with subspecialists. This negative impression persisted even if physicians offered expertise and a positive interpersonal interaction. In at least five cases (families #2, 4, 6, 7, 8), these concerns led to relationship breakdown with a community physician. For family #6, the mother reported both positive and negative experiences with physicians as it relates to accessibility and responsiveness. This was a foundation of her positive relationship with the family physician, as outlined below.

> *"…family doctor [is] quite easily available and very good in the sense that she'll get back to me very easily…I've gotten calls from her at home even or from the hospital at ten thirty at night because she needed to tell me something that was important…so I feel like there's very good communication there."*
>
> *(Participant 6)*

However, the same family experienced relationship breakdown with their community pediatrician. This was partially related to difficulties getting a response when they had concerns, but had a number of other underlying factors as well.

A similar experience was reported by mother #4. She had a positive relationship with a family physician who was accessible and available to her at short notice. She expressed dissatisfaction with her son's community pediatrician, in part due to the cumbersome process that was needed to get an appointment, as outlined below.

> *[Community pediatrician] "I don't really bother with him anymore because it's such a hassle to get in there…every time they call me and they'll be like we need to see [name]….but you need to get a re-referral…so then I'll go to my doctor and my doctor will send a referral. And then even after that I still have to wait for an appointment…And it's just like, you're the one who wants to see me, why am I going through this whole hassle to come in there?"*
>
> *(Participant 4)*

For one mother (#8) even a strong positive impression of her child's community pediatrician during the visit did not override the logistical challenges of getting an appointment, with the latter resulting in dissolution of the relationship.

> *[Community pediatrician, when drawing the care map]"…she's not on here because I never see her…I mean the one time I saw her, she was amazing, but getting in to see her is just ridiculous."*
>
> *(Participant 8)*

Families #3 and #9 both seemed satisfied with a point of contact through their community pediatricians.

Some families didn't mention this as a specific issue. For family #1, they had longstanding, centralized access to the biochemical diseases subspecialty service, which provided or coordinated all their child's medical care. Although they may have valued the responsiveness of this team, it was not mentioned. For family #10, they only accessed community medical care through the emergency department but were pursuing a relationship with a new community pediatrician. Their point of contact was by phone to the subspecialty service at BC Children's to a nurse and physician.

Physician as advocate-The efforts of physicians and other health care providers in securing support or funding for the child was highly valued by a few parents. At times, these situations were related to the diagnostic process or the rare disease itself, and at other times they were not. For instance, mother #1 describes her child's subspecialty physician advocating for developmental assessments, support for live-in caregivers and government funding (none of which were specific to the child's diagnosis). Physician motivation to find a diagnosis for the child and advocate for testing and diagnostic assessments was viewed positively by many families. Parents appreciated the support of primary care physicians in creating (by means of referral) or brokering relationships with their child's specialists. Mother #4 had a strong relationship with her family physician, who advocated to her son's community pediatrician when concerns were not being addressed.

> *[Regarding family physician] "When I was expressing concerns to her about Dr. X., like she's the one that was like, okay, I'll call him and I'll talk to him and I'll figure out like what's going on and stuff…she'd always basically take that extra step to help me, yeah, so I really, really liked her."*
>
> *(Participant 4)*

Mother #3 valued advocacy in general, but this was more related to the allied health professionals involved, in particular to a community physiotherapist. This was also a mother who was less focused on diagnostics and medical care, which may have de-emphasized the advocacy role for physician.

Physician as emotional support- In some cases, parents described physician relationships, positive and negative, in terms of their ability to provide emotional support

to the family. This was another source of dissatisfaction expressed by mother #6 in relationship to their community pediatrician - that he wasn't providing the empathy and emotional support she desired.

> *"…I just remember one time that he said, you know…she's not dying and there's other kids that are dying and we have to help them. It's like whoa, that's kind of hard and I realize it's true, but I think what we were going through too was a little bit hard…"*
>
> *(Participant 6)*

This emotional support was a foundation of the satisfaction that father #7 expressed with respect to both his community pediatrician and family physician, both of whom treated them "as a family," as opposed to focusing only on the child.

> *"Our new pediatrician is I would say much more empathetic to sort of the parental needs and he was, he's very sensitive to our situation and he's very patient with us."*
>
> *(Participant 7)*

This theme was not a focus for all participants, many of whom received emotional support from family members (#1 and #5) or peer groups (#3 and #10). Family #9 did not discuss the source of their social-emotional support, which may have had cultural underpinnings but could also have been due to personal factors.

3.2.3 Peers play key roles

Nearly all the participants emphasized the key role played by peers. Some of the participants were connected to disease-specific individual peers or support groups, particularly the two individuals with metabolic diseases. Commonly, peers were

alternatively identified in relation to proximity or the child's disability. For mother #3 peers were identified by virtue of also having children with ultra-rare or undiagnosed conditions.

Peers as experts - In some cases, peers were considered experts in a particular disease. This was the case for mothers #1 and #2, with connections formed mainly online. For the other families, peer expertise was rooted in their ability to navigate health and social systems. One parent (#8) identified her daycare provider, also the mother of a child with special needs, as playing a key expert role. Both her child and the peer's children were diagnosed with autism – a diagnosis that comes with particular funding streams and developmental supports, which would have made the peers' expertise even more valuable.

> *[Describing daycare provider] "She is probably my number one support, she's amazing. She knows everything, the system, everything. With six kids and five that are [diagnosis], she has been a wealth of knowledge for me…."*
>
> *(Participant 8)*

Some participants (#2, 7 and 10) placed particular value on relationships with peers who had older children. These peers offered them a glimpse into the future and detailed advice on navigating challenging times, such as transitions. These peers are described as "mentors" by some of the parents.

> *[Describing a peer] "X is a parent, he is a parent of a man in his thirties, who was one of the first people in Canada diagnosed with [diagnosis]. So in many ways X is me in twenty five years. He's made several attempts to connect with us and to give us advice, so he's been a big help and he probably will continue to be for some time."*

(Participant 7)

> *[Describing a peer support group] "they've been able to tell me, this is what you do, you know, just do this and that and this…because they've all done it so it's kind of worked out really well for me because [child] is a year behind them. So that's been helpful, you know, just with everything. How to get, how to get our van, how to go and get the conversion done and who to go with and, you know, just all kinds of information."*

(Participant 10)

Peers as social supports - A number of parents described the benefit of connecting with peers as a way to normalize their child's condition and their own experience. This was particularly poignant for the parents of the two children with metabolic conditions, but also held true for a few of the undiagnosed parents.

> *"…there was a conference on, on this disease and I went to, I attended one of them just to meet other people. It was kind of pretty cool too, you know, sometimes I saw it's like walking around with the only kid that's got pink spots and green stripes and to walk into a whole roomful of people with pink spots and green stripes was pretty cool. So just to feel like you're not alone kind of thing."*

(Participant 2)

> *[Describing a peer relationship] "It is huge, it's totally invaluable right, you know…we'd want the kids to meet up. I mean having another kid with a rare disorder meet up with another kid with the same rare disorder makes them normal right?"*

(Participant 1)

One parent (#3) connected strongly with a rare disease organization that gave her a sense of belonging and community.

> *"You don't want to alarm parents, but I wish [Foundation] was right up there. Because when [child] was having seizures they sent somebody from epilepsy, BC Epilepsy Society, with a big book about seizures and well there was no such thing as rare disease. So I would have wanted now, if I had a child with special needs, I would want [Foundation] to be one of those people visiting me and talking to me about stuff." Later: "…and if you start with [Foundation], for me that was the blessing. Some have older kids, younger kids, but at the end of the day we all have unknown in common."*
>
> *(Participant 3)*

Peers and identity- In a number of cases, relationships with peers are intertwined with parents' descriptions of their identity as the parent of a child with special needs.

One mother (#10), who eventually formed a strong bond with a peer group, was initially very reluctant to engage as it didn't fit with her perceptions of herself and her life. She framed this transition as related to her adjustment to being the parent of a child with a disability.

> *"…at the beginning, they had suggested a support group, asked me if I wanted to go and I didn't, I wasn't open to that…I didn't want to because I didn't want to be part of that group, you know what I mean, I didn't want to be one of those mums with the disabled kid. I was just gonna go on with my life the way it normally is."*
>
> *(Participant 10)*

Another mother (#1) was in the position of providing expert peer guidance to another family, whose children had a related condition. She describes their relationship as reciprocal. However, it is clearly mother #1 who is able to offer more support and

assistance particularly with regard to organization and logistics. She seems to solidify her identity by comparison with this peer.

> *"I felt like how I coped with my two girls, versus how she's coped with her two kids with [diagnosis], I was coping a lot better…she was always like: oh my god, I'm so sorry I really need another case of [formula] because I, I just…she didn't have the time or the organization to order it in time…so I'd be like, okay, yeah, yeah come on and pick it up."*
>
> *(Participant 1)*

Another parent (#3) expressed the willingness and desire to help another family through shared experience. This was in the context of attributing meaning to her child's life and their struggles.

> *"I just, I don't want his life to be in vain and if I could contribute to another family. Like, it's going to be very hard, it's going to be super hard, but if we can all kind of come together. Like if you can take me and introduce me to another family and say this is going to be ok, it's going to take time, it will happen…"*
>
> *(Participant 3)*

A few parents did not emphasize the role of peers, namely participants #4, 6 and 9. These parents instead relied heavily upon allied health and school personnel to provide them with advice. Family #6 had strong social-emotional support from extended family and from a home-based therapy team (behavioural consultants and interventionists). Families #4 and 9 mentioned expertise and support provided by allied health personnel. Family #9 didn't specifically mention topics related to social support and identity.

3.3. Theme Three: Gaps exist in Coordination and Collaboration

This theme relates to a gap in the division of roles and responsibilities between parents and health care providers as described above. Invariably, the children involved in this study required the care of multiple physicians, in addition to community and allied health providers. Parents were often concerned about the disconnected and fragmented nature of this group of professionals.

In many cases, they felt that they were the only functional connection among health care providers involved in their child's care. This was a source of frustration for some families.

> *"I think they all should be communicating, but that's it, you know, like they communicate with me, they all communicate with me, but they should be communicating together for sure."*
>
> *(Participant 8)*

Many families feel the sharpest divides are between tertiary care and community based providers and between physicians and non-physicians, which they described in the exercise creating their child's care map.

> *"…these are completely separate entities right – this is the hospital and this is, you know, the community right? And to facilitate something to connect all of that for my child's care is really dreaming, it is."*
>
> *(Participant 1)*

"And the doctors are all in their own group and child development is all in their own group, I think they've been placed just in their own little world."

(Participant 10)

The connections that did exist tended to be formed through the transmission of paper records, felt by parents to be an imperfect process. One parent addressed the unpredictability of these communications by taking full responsibility for transfer of information in the form of written reports by physically bringing them to each appointment.

"That's important because I need to bring his records everywhere I go. Like I don't know, if I see this doctor, what kind of report he needs?"

(Participant 9)

A few parents resented this role of passing information from one physician to another, feeling that paper records were either inadequate or under-utilized.

"…like when you're admitted, they'll have a whole bunch of, you know, doctors asking the same questions and I'm like well can you look at her chart or something. So, it gets a little frustrating…like when I go to biochemical diseases clinic and they ask me a lot of questions. I mean the information is there for them as well right?...there's a report from cardiology about the last meeting we had. But they rely on my information. So they're sort of like, okay, well, what happened in that cardiology appointment?"

(Participant 1)

A few of the parents were also frustrated by the need to verbally tell their child's story during encounters with physicians (giving the medical history). Some families (#1, #4, #8) disliked the repetitive nature of this exercise and felt the information could be easily

obtained from the child's paper records. For mother #3, her reluctance to relay her son's medical history may have also related to ongoing guilt regarding the etiology of her son's condition.

> *"They want it [from the beginning], especially as residents and doctors on call and fellows…they have no clue, so we have to start from the beginning and that's one issue I want to end."*
>
> *(Participant 3)*

Many felt that the lack of coordination contributed to delays or inadequate care delivery for their children, feeling that no one was taking responsibility. One parent (#2) likened the experience at the beginning of their diagnostic journey to physicians passing her child around like a "hot potato." To some degree, this was related to the nature of her son's illness – a metabolic condition that initially presented with liver manifestations, leading to uncertainty regarding which subspecialty service should investigate.

> *"…there was just this back and forth and I feel like we kind of got lost in there for a few weeks between trying to find out who we're seeing…so there was just that sort of back and forth. And it just took some time, where you just feel like could you just not, let's just get some of it rolling rather than whose taking responsibility for it…Yeah, it was just, you know, well, the hot potato."*
>
> *(Participant 2)*

Most families did not perceive a central point of contact or a professional who was overseeing their child's care, particularly the families of eight children who were not definitively diagnosed. They were left relatively unsupported to manage the confusing systems and processes involved in managing their child's medical care. Often, parents desired assistance with organizing and navigating not only medical services, but

developmental and social services as well. All parents expressed this unmet need on some level.

> *"That would be so ideal and that one person would have the wealth of knowledge of everything you know: which department I need to talk to for [child] to get his benefits and what he, what we can get for him. Know my appointments, know my, I need another me, you know, and I think, I think there's a fundamental problem with the system is that nothing is streamlined, everything is complicated and lengthy…"*
>
> *(Participant 8)*

This lack of centralization and coordination left some parents unsure about the plan or direction they were going. Parents wanted the sense of this overall plan, even if they knew a diagnosis was not forthcoming. The desire was greater for families who were more focused on finding a diagnosis, presumably as they wanted to consider future diagnostic options.

> *"…there was just sort of that lack of, I don't know, communication or what the next steps were. I was just sort of like, okay, well what are we going to do now? What is going to happen now? Okay we don't have any answers but we're, we're still here sort of thing."*
>
> *(Participant 2)*

Family #5 harboured serious concerns about the potential impact this lack of coordination could have on their child's well-being. These parents had post-graduate training in a health care field and held a dual perspective on the need for collaboration – not only from the parental perspective, but also from the professional perspective.

> *"And so it was things like that which really scares me. That because there's a disconnect between all these healthcare professionals. Like what may be good for,*

for example, for some people it may be good for diabetes, may not be good for low blood pressure…So that person may have these complicated healthcare conditions but one doctor may be a specialist trying to cure it but the same treatment might cause an issue. So it's, I find that very scary in our healthcare system, because yes we have ten specialists seeing [child] but have they ever sat down and really discussed [child] or have they really talked about her?"

(Participant 5)

This same family emphasized the benefit of shared goals amongst health care providers and the family which could also have been a component of their previous professional training or practice.

"…being able to share with other people, specialists or doctors or therapists, and for all of them to kind of have the same vision or goals to find a cure or find an answer is really, really comforting."

(Participant 5)

This need for collaborative goal setting was also mentioned by mother #6, but not by the other families.

"…they all kind of connect really but I don't know if that's what you mean? I think that they all connect because each one of these people doing their thing…their ultimate goal is helping [child] and helping her succeed."

(Participant 6)

CHAPTER FOUR: DISCUSSION AND CONCLUSION

The intention of this study was to examine parents' experiences when accessing health care for children who have ultra-rare or undiagnosed conditions. Information was elicited from parents to reflect their conceptualization of how these providers functioned as a group, shared information and related to each other. The results conclude that uncertainty and the many unknowns surrounding the child's condition is an important component of the parents' experience of their child's illness, and their interactions with health care providers. This fundamental uncertainty leads to different roles and responsibilities being adopted by parents and physicians than what would traditionally be expected. The role of peers, that is parents of children with similar conditions, was also a prominent focus. Finally, parents perceived a general lack of coordination and collaboration amongst the health care providers involved with their child.

4.1. Findings related to uncertainty in context

Uncertainty was identified as a universal phenomenon for the study participants. This is not a surprising result considering that uncertainty is a pervasive component of all chronic illness experience, as individuals struggle to align their expectations and assumptions with their new reality.[11,18] As Mishel (1988) describes in the uncertainty theory, when illness-related symptoms or events are experienced, patients attempt to create cognitive schemata to attribute meaning. In many situations, information can be gleaned from physicians and other sources that assist in the construction of these schemata, thereby minimizing uncertainty.[10,72] Since expertise in rare conditions is limited, parents struggle to find meaning for their child's conditions. Other studies have

endorsed the complex nature of parental uncertainty when dealing with a child's rare disease, distinct from the experience of adult patients, and remarked upon a variety of themes, such as normalization uncertainty ("how will this impact my child's life") and parent-as-proxy decision making uncertainty ("is my child in pain").[19] The uncertainty that parents in this study experienced was also multidimensional and, due to the research question at hand, was strongly related to interactions with the health care system.

One particularly formative experience for many parents was the initial impression that something was wrong with their child. This sense could be related to features, symptoms or episodes, but was often difficult to fully appreciate or describe. Relaying these concerns to physicians, particularly front line physicians, was challenging. Often parents were frustrated by these interactions and struggled to have their concerns appreciated and validated.

This uncertainty that "something was wrong" seemed to be mediated by certain factors. For instance, those who had received early care through NICU (#3, 5 and #9) may have benefitted from an early shared understanding with physicians, recognizing that a medical condition was present that required ongoing care and attention. However, family #5 went on to struggle with the "something is wrong" uncertainty when their child manifested episodic symptoms of an unclear etiology. These subtle, episodic and non-specific signs and symptoms are likely more prone to this type of uncertainty, particularly in comparison to a classic or acute presentation of disease as occurred in family #1.

These early difficulties are commonly reported by patients of all ages with rare diseases, many of whom initially feel brushed off or misunderstood by their physicians.[27,29] The lack of physician knowledge about rare conditions and their manifestations may make them less likely to respond appropriately and is an important contributor to these negative experiences.[29] Additional contributors could include the difficulty recognizing and appreciating episodic or unusual disease manifestations. Working through these early interactions is challenging and can contribute to uncertainty on both sides – is something actually wrong with the child, and if so, what? Disagreement on this point could set the stage for ongoing miscommunication and distrust.[27,73]

Another aspect of uncertainty, one that is not as commonly reported in the literature, is that which remains even after a diagnosis has been assigned. Most children in this study had a positive test result or presumptive diagnosis at some stage that was later disproved or discarded. This often gave parents less confidence in future diagnoses that were suggested. Even the parents of children with current provisional diagnoses continued to express doubt about the validity of the suggested disease and concern about the possibility of misdiagnosis. These parents were likely particularly prone to this uncertainty due to their extended diagnostic journeys leading to an increased likelihood of misdiagnoses or “red herring” results.

The families who did not experience uncertainty regarding the accuracy of the diagnosis were the two with metabolic conditions (#1 and #2). Both diseases had associated treatment, based on dietary modification and supplementation. Perceived response to these treatments could have provided proof to these families that the diagnosis ascribed was the correct one. Metabolic diseases also receive care through a centralized

multidisciplinary subspecialty service which could have further reinforced the accuracy of the diagnosis, since it was accompanied by a distinct change in service delivery.

The frequency of misdiagnosis in rare diseases overall is high, in both adult and pediatric patients; however, other studies have not found this to be a major concern to patients and caregivers.[19,74] These other populations sampled seemed to have more confidence in the accuracy of their diagnosis. An additional factor potentially contributing to a higher frequency of diagnostic uncertainty is the recent expansion of available testing, including whole genome sequencing, that raises greater possibility of unclear, uninterpretable or false positive results.[75,76]

Some parents in this study expressed a lack of clarity and confidence regarding the best course of action when managing their child's disease and manifestations. This was more of a concern for the parents of children who had treatable metabolic conditions, where there was a greater expectation for guidance and intervention. However, due to the rarity of these conditions, treatment plans are often not rigorously studied or evidence based.[6] In many circumstances, even the response by individual patients can be unique and varied. Dealing with the unknowns regarding treatment options is a major source of distress for patients with rare disease and particularly for parents. Even if treatment options exist, they are often not universally available or effective.[19,29]

For the other families, this uncertainty about the best plan of action related to issues both related and unrelated to diagnosis. Some families wanted advice regarding the day-to-day management of their child's symptoms or behaviours. Others were looking for a "big picture plan" regarding ongoing diagnostic work-up.

Finally, the desire for prognostic information was universal and often intense among participants. This was one of the greatest motivators behind many of the parents' goals, such as the ongoing search for a diagnosis and the construction of a peer support network, with the latter providing a glimpse into the future. Parents of children with rare disease, chronic disease and neurodevelopmental disability are all concerned about prognosis and their child's future.[12,38] This arises out of a struggle to align the reality of their child's illness with the future vision they had previously created.[77] Repercussions of this uncertainty include the inability to plan, not only for their child's future, but also for themselves and their other children who may be impacted.[77] To some degree, this concern related also to the perceived lack of support for adults with chronic diseases and disabilities, as well as reticence about transitioning out of pediatric health and social systems. Transition points are known to be a key source of stress for parents of children with chronic conditions, particularly the transition to adult services.[50] In this study, there was not a clear relationship between age of the child and degree of concern regarding prognosis.

To a variable degree, parents in this study were aware of the uncertainties that would persist both before and after a diagnosis. Although these participants were not followed longitudinally, other studies have shown that the desire for a diagnosis is most intense when problems are first recognized, likely related to a hope for treatment or cure.[12,25,26] The natural history of this interest is to decline over time as parents realize that a diagnosis is unlikely to change their child's prognosis or even service eligibility.[25]

4.2. Findings related to roles and relationships in context

The many unknowns surrounding the etiology, diagnosis, management and prognosis of rare diseases had significant implications on the parent-physician relationship. One important change is a shift in roles and responsibilities, in part related to a lack of concentrated expertise on the part of the physician. Many parents in this study felt they held much of the expertise related to the child's disease or condition compared to health care providers. Another notable component of their expertise was related to knowing their child individually, even more crucial in the setting of a rare disease with significant individual heterogeneity in its manifestations. This phenomenon of the expert patient (or in this case, parent) is well described in the current literature and in popular culture.[27,46,50]

There were a few interesting deviations from this anticipated phenomenon. For the mother in family #3, the burden associated with being placed in an expert role was a source of distress for her. Although she was confident in some areas of her child's care, reporting her observations and thoughts to the medical team, particularly regarding her child's seizures, was seen as overly subjective. This could be contrasted with family #10, who also had a child with difficult-to-control seizures and severe neurodevelopmental impairment. This mother was very confident in the reporting and management of her child's epilepsy. There are many factors that could contribute to these differences, including personal and cultural. However, the lack of universality of this phenomenon is important to recognize and may come with a shift in approach with families who do not perceive themselves in the expert role.

The phenomenon of the expert parent in the setting of a child with an undiagnosed condition or when the foundation of the expertise lies in knowing the individual child is an under-explored area. This has the potential for broad applicability across pediatric complex, chronic conditions. This type of expertise also resonates with the current attention to personalized medicine with its focus on assessment and tailored therapy to the conditions, genomes, goals and preferences of the individual.[78] Physicians, particularly generalists, have difficulty keeping up with the pace of research and rapidly expanding pool of rare diseases.[30,54,79] Parents in this study often found that information that came from online sources or from peers more helpful than that gained from physicians. To some degree, this is endorsed by prior studies, although these tended to rank information obtained from physicians at a higher value.[80,81]

Due to their expertise and motivation to find answers, many parents saw themselves in a directive and leading role in the pursuit of further diagnostic testing and in their child's ongoing care. Parents strongly identified with the role of advocate; in their words, frequently needing to "push," "hassle" and "fight" for services and supports for their child. Parents of children with a variety of neurodevelopmental conditions and chronic diseases also commonly see themselves in an advocacy role. A common theme for many parents, in this and other studies, was the need for advocacy and vigilance when relating to developmental services and school supports.[82] There was significant variability in the parents' satisfaction with taking on these roles, with some expressing more desire for a professional to take the lead and feeling overwhelmed by this responsibility. Other studies have suggested that parents thrive in the expert role, but most would still appreciate greater support.[12,42,46]

This rise of the parent to take on roles of leadership and expertise created difficulty at times when interacting with physicians, who would traditionally have filled these roles. Parents often felt that their expertise was not recognized, since they are not "professionals". Some parents encountered significant barriers when trying to provide input into the diagnostic process, when physicians were not always open to considering their ideas and suggestions. The conflict created by the lack of professional recognition of parent expertise and the central role they play in their child's care is well described in the literature, in reference to many types of childhood chronic disease and disability.[12,46] There may be an opportunity for greater partnership, particularly in the diagnostic pursuit, but both physicians and parents may need more support and guidance about how to navigate these roles.

Even though challenges exist in the parent-physician relationship, many parents managed to develop and maintain highly satisfying relationships with physicians, both generalists and subspecialists. The physicians involved in these relationships were sometimes valued for their medical expertise, particularly for the families of children with metabolic conditions. For others, their value was sometimes related to fulfilling other needs of the family. These parent-physician relationships often supported and facilitated the parents in acting out their roles of expert, leader and advocate. Parents in the study also described current or outstanding roles and responsibilities that they would like their child's physicians to take on – that of provider of information and explanation, point of contact, advocate and emotional support.

Even with the high level of parental expertise, physicians were still viewed as an important conduit for information. Providing access to their child's information and giving

meaning and context to these facts were valued roles that physicians played in the child's health care. However some families, particularly #2 and #9, also desired direct access to the "raw data" of their child's medical information in which case physicians could be viewed as a barrier to this information. Their motivations are likely quite different with mother #2 working in the health care system and taking a strong leading role in her child's diagnosis, whereas for mother #9 the desire may have reflected her difficulty communicating in English.

Research into patient-physician communication in rare diseases reinforce the importance of professionals providing guidance and context to medical information.[29,46] However, communication is challenging, particularly as it relates to childhood disability and uncertainty around diagnosis.[83] A combination of transparency and attention to physician-family communication in the setting of uncertainty would likely be most effective to support the information needs of the majority of families.

In addition to their need for information, some parents desired a physician point of contact who is accessible and responsive. In this context, access included availability of the service (for example, having an identified primary care physician), lack of barriers (for example, a streamlined experience making an appointment) and acceptable response (for example, timely).[84] Not surprisingly, this is a priority for many parents of children with complex illnesses.[46] Ready access to one's physician, particularly in the primary care realm, is highly correlated with patient satisfaction.[85] Perhaps even more importantly, positive relationships including responsiveness and strong communication with physicians correlate with better health outcomes for all types of patients with chronic diseases.[32]

Some parents in this study sought out their child's physicians for social-emotional support, instead of or in addition to medical expertise. These parents wanted their physicians to adopt a sensitive and understanding position and to appreciate their struggles as a family. These types of interpersonal skills are highly valued in other studies also.[12,30,54] Particularly for patients with rare disease, the physician can provide important guidance on the social aspects of living with a rare condition or communicating to others about their diagnosis.[29] The physician role was also viewed positively when it was used to advocate and support the family in accessing desired services, another theme commonly reflected in the literature.[46] However, for the parents in this study, the ability or lack of ability for their child's physicians to provide them with information and guidance is a major theme running throughout their discourse regarding roles and relationships.

Finally, the important role that peers play for parents of children with ultra-rare and undiagnosed diseases was a key theme. This is demonstrated in many studies spanning all ages and types of chronic conditions.[86] In this study, peers were felt to contribute expertise, social support and to assist with formation of the parent's identity. The expertise that exists among peers with the same condition (or parents of children with the same condition) is widely recognized in the literature.[81,87] In many cases, as in this study, peers are one of the most important sources of guidance and advice. They can place information in context in a way that health care providers cannot, with expertise that extends to the day-to-day management and challenges that arise in the condition.[87]

Peers are an important source of social support and community – a highly prioritized role for nearly all parents in this study. This is one of the major motivations underlying the search for a diagnosis in the first place. Most parents aligned with a peer social network that was based on proximity, the nature of the child's disability or the nature of the support they were seeking, as opposed to being disease specific. Whether or not they were diagnosis-based, these networks gave the parents a sense of normalization of their own experience and their child's condition. The value of peers who share a similar moral or practical experience is reported as meaningful to many individuals with rare disease.[29] Although the value of disease-specific peer networks is well documented, the value of peer groups formed on the basis of disease manifestations, disability or proximity have not been fully explored in the literature.

4.3. Findings related to lack of coordination in context

The most prominent gap in care that these families experienced was related to coordination of care (CoC). CoC is the organization of patient care activities between two or more participants (inclusive of the patient) to facilitate appropriate delivery of health care services. This includes marshalling the appropriate assessments and resources and facilitating transfer of information.[88] Many of the struggles that families experienced were related to a lack of coordination. They perceived the lack of a unified plan, incomplete information sharing (both with the family and between health care providers), poor logistical coordination and fragmentation of care.

CoC is a frequently recognized deficiency in the health care system, particularly related to the management of complex, chronic conditions and rare diseases.[50,89,90] Even

compared to other studies of rare disease, an outstanding need for CoC was a greater focus for the parents in this population. Patients with ultra-rare and undiagnosed conditions are at risk of deficiencies in coordination of care, since criteria for designated programs continue to rely mainly on categorical diagnoses.[91] This has significant implications for equity and quality of care, since CoC is associated with improved disease outcomes and patient experience.[92,93]

Parents in this study are currently taking the burden of care coordination, from making sure all diagnostic avenues and possibilities have been explored, to keeping track of necessary follow-up appointments, to ensuring that their child's medical information is being shared appropriately. However, they are not provided with many of the vital tools and skills to optimally enact this role, such as ready access to their child's medical information and the ability to engage their child's physicians. Other studies have also demonstrated patients and parents/caregivers placed in the role of care coordinator.[94] However, there are many other models of providing care coordination, through different care models and professionals including family physicians.[30]

In many ways, the experiences of parents of children with rare and undiagnosed conditions resemble those with other forms of chronic disease and disability. In all cases, parents can struggle to partner effectively with health care providers and require CoC. They advocate for their child's needs and cultivate strong self-management skills.[95–97] The accumulation of these roles, in addition to parenting a child with special needs, can easily lead to exhaustion and burnout.[98] Balance must be sought between empowering and engaging parents, while ensuring that they have the time, tools, resources and support to fill these important roles. The unique aspects of the parent

experience in the setting of rare and undiagnosed conditions is the layering of the persistent and complex nature of their uncertainty onto existing challenges with the medical system.

4.4. Focus on medical and physician care

Although the interviews were initially framed to identify themes relevant to a broad array of health care services and experiences, the resulting data was weighted to a discussion of medical and physician care. There are likely a number of possible reasons for this focus. First of all, it is possible that many of the most remarkable experiences of the families, either positive or negative, had been with physicians and these may have been drawn out in the interviews. For instance, physicians would have suggested or confirmed diagnoses, experiences that are known to carry significance with patients and families. Another possibility is that patients and families were attracted to the My Care Web study due to difficulties they encountered regarding their access to medical care and relationship to physicians. The My Care Web study was designed to facilitate access to care, and for most families, the barriers in this area were mostly related to physicians. Finally, the interviewer's identity as a physician cannot be ignored as a potential source of bias. Participants may have felt that medical and physician care was the primary interest, regardless of the framing of the questions.[99,100] Of note, there was no apparent difference in emphasis on physician care depending on whether the interviews were conducted in the home, by phone or in the hospital setting.

4.5. Variability within the study population

There was significant variability within the study population that likely gave rise to different perspectives on the themes outlined above. A few examples of population subsets will be discussed below.

Two of the children had treatable (although not curable) metabolic conditions. Both were engaged with subspecialist physicians who had considerable expertise in their condition and were connected to disease-specific support groups. However, their experiences were certainly not uniform, particularly as they relate to communication and relationships with physicians. Mother #2 had experienced distrust and relationship breakdown with one of her child's community physicians. She desired increased transparency with her child's physicians and more direct one-on-one communication. Mother #1 had a longstanding, positive relationship with her child's main physician, a subspecialist. This might have related to her child's early diagnosis. She also prioritized provider-to-provider communication and did not express a desire for direct access to her child's information. These two families had children with the most similar conditions yet their needs and wishes were quite different, likely related to their prior experiences.

Families #3 and #10 had children with the most severe neurodevelopmental impairment. These two children also had epilepsy and gastrostomy tube feeds. Their physical conditions were more fragile and their day-to-day care needs were greater than the other children in the study. These parents were both less focused on diagnosis and less likely to direct their own efforts to this endeavour. Each had found strong non-diagnosis based peer groups who provided mostly social-emotional and practical support, the

latter focused on navigating social systems. Of all participants, these two actually had the weakest connection to the physicians involved in their child's care. It's possible that the child's day-to-day care and developmental needs took attention away from a focus on diagnostic pursuits. Again, however, perspectives were not wholly similar. In particular, mother #10 was confident in the care and knowledge of her son and struggled with professional recognition of her expertise. Mother #3 was less confident and often felt burdened by physicians' expectations of her knowledge.

4.6. What is the meaning of a diagnosis

As introduced briefly earlier, the meaning of "diagnosis" is complex and multifaceted. The traditional disease perspective relies on a biomedical paradigm that applies treatments to halt or reverse the underlying pathophysiology of a disease. This is often the focus of rare disease research, a field that grew out of the desire to develop targeted treatments and develop orphan drug policies.[3,4,101] However, for many conditions, a direct connection between diagnosis and a specific therapeutic intervention is not a realistic expectation. For instance, many diagnoses related to developmental or mental health conditions are based on developmental-functional criteria; their underlying biological bases are often unknown and treatments are typically supportive and general. Even for diagnoses that are based on a known medical etiology, many do not have targeted treatments.[15]

There is growing interest in alternative and complementary perspectives that will be useful in describing, characterizing and supporting distinct groups of patients with a variety of conditions. Miller and Rosenbaum describe the disability perspective (in

contrast to the disease perspective) which focuses on the individual (function at the level of body, person and person-in-society) as opposed to the disease.[15] This idiographic knowledge of individual cases also relates back to the “expert in the child” role played by parents in this study. Other non-disease based descriptors are also gaining recognition, such as children with medical complexity and children with special health care needs. These categories are focused on the physical, psychological, developmental and social needs of the child and family and in many ways are more useful than a specific etiological diagnosis.[102,103]

Many themes identified in this study are not unique to parents of children with rare or undiagnosed conditions. The children in this study also invariably have complex conditions and neurodevelopmental impairment and they share important similarities with these populations in terms of their health care experience. For instance, the desire for increased coordination of care is mentioned in studies of children with a variety of conditions and may be more related to complexity of care than to the presence of a rare disease.[15,89,104,105] Although there are unique aspects to the ultra-rare and undiagnosed population such as uncertainty related to the long diagnostic odyssey, there is also significant heterogeneity within the group and overlap with other populations with chronic disease and disability. This highlights the need to address each patient as an individual, addressing their specific needs and goals in a manner that is mindful of both disease-specific and functional factors.

4.7. Limitations

This study had notable limitations with respect to the selection methods for the study population, generalizability and validity.

Selection bias for the study population

One main limitation of this study was the selection bias introduced with the recruitment procedures and source of the study population. These children were affected by the rarest of conditions, culminating in their referral to the TIDE-BC Complex Diagnostic Clinic – a limited resource of the highest-level of available diagnostic testing. In order to even obtain this referral, these parents or their physicians must have exhibited considerable motivation to find a diagnosis for their child. Their perspectives might differ substantially from parents who did not pursue diagnosis to the same degree, or who were perhaps not even aware that the option existed. They also likely differed in fundamental ways from parents of children who were diagnosed more easily or earlier in life. Both of these comparison groups could have had very different experience of care and interactions with medical professionals. To that end, study results may be most applicable to undiagnosed patient populations.

Since these interviews were conducted during recruitment for the My Care Web study, the participants were aware of, and had already agreed to participate in, an intervention addressing care coordination and collaboration. A significant proportion of families, more than half, declined to participate in the My Care Web study. Important factors were involved in this decision, most notably time constraints, language barriers and fragile

condition of the child. It is possible that this study missed incorporating the experiences of the families who were struggling the most or whose children had the most complex needs. There were a high number of families for whom we did not get a response at all to the study invitation. There are a variety of possible explanations including families not seeing a need for the intervention, reluctance due to negative experiences with health care and/or the CDC clinic or challenging personal circumstances. There are likely other possibilities as well that cannot be accounted for in these study results.

Since the My Care Web study was focused on improving communication and coordination, the study population may alternatively have experienced more challenges in these areas than others who had declined participation. In addition, some of the results obtained reflected the reasons that the participants agreed to participate in the My Care Web study. The My Care Web study was designed to address the perceived need for greater transparency in accessing medical information, easier access to physicians involved in their child's care and improved coordination of care service. Such a selection bias has likely resulted in stronger opinions regarding these issues, and perhaps a more dissatisfied view of the health care system, than would be expressed in other populations. Indeed, one family cited their satisfaction with current health care delivery as a reason for declining to participate in the My Care Web study and others may have felt the same. A study involving this group of more "satisfied" families may have produced quite different results.

Heterogeneous study population

Another limitation is the small and relatively heterogeneous group of patients; first, regarding the conditions themselves. For most children, their lack of diagnosis, or presumptive diagnosis of an ultra-rare condition, resulted in a paucity of information, a lack of centralized expertise for care and few specific treatment options. These conditions invariably were accompanied by developmental delay, but over a broad spectrum from mild to severe. Some children had additional neurological or medical manifestations such as epilepsy, gastro-esophageal reflux disease or ataxia. Most children were felt to have a genetic basis for their disease but this could have been inherited or developed de novo. The fragility and care requirements of these children varied significantly, which undoubtedly had an impact on how their parents' perceived their medical care.

Although the diagnostic experience to date was shared to some degree, by virtue of having ended up in a rare disease clinic, the individual experiences were still unique. Surely, the application of even a provisional diagnosis changed the experience of these families to some degree. Children in the study ranged widely in age, from those diagnosed thirteen years ago at age five months, to still being undiagnosed at seven years of age. Thus, the amount of time families had spent "in the system" and with or without a diagnosis varied widely. However, with the exception of the first family, all had struggled to find a diagnosis for at least one year and, by virtue of their referral to CDC, had exhausted many investigations.

The study population also exhibited significant differences in personal-social factors. Parents came from diverse cultural and socio-economic groups. Their educational and professional backgrounds were broad, encompassing a spectrum from high school to post-graduate education. Other important factors such as degree of social support, relationships with co-parents, spouses and family members, and personal experiences of illness and wellness were not fully explored but likely contributed to many of their reflections. Where possible, contextual information was provided to assist the reader in considering other sources of bias and personal circumstances that could have influenced the participants' perspectives.

Methodological limitations

The validity of the data is limited due to the lack of triangulation. The data came from a single source, the interviews with study participants, at a single point in time. In addition, analysis was conducted by a single researcher (T.D.). The single researcher design in the conduct and analysis of the interviews, in addition to the recruitment and conduct of the My Care Web study also allowed for greater depth of involvement and understanding both with the study participants and the data. Involvement of additional researchers in analysis would have allowed for different perspectives on the results and would potentially minimize any bias introduced by the researcher. Inclusion of other data sources, from other populations or derived in different formats (i.e. focus groups), would have further enhanced the results obtained. In addition, the results were not reviewed with the study participants, which would also enhance validity.

4.8. Implications

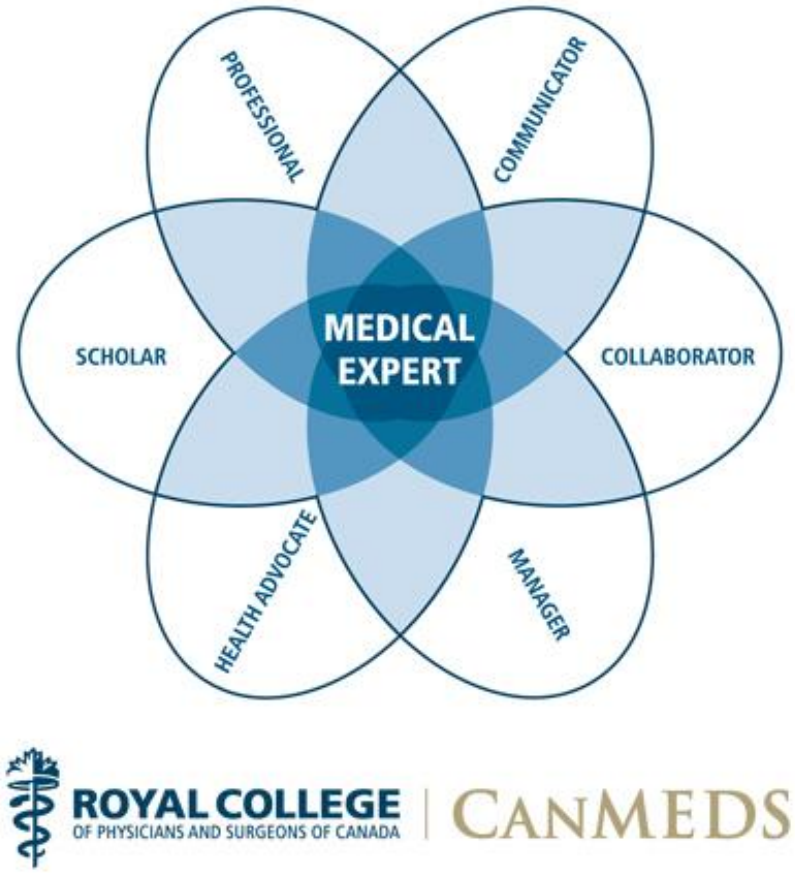
Uncertainty is a persistent and complex phenomenon that both precedes and follows the assignment of a diagnosis. Parental expectations must be understood during these long diagnostic odysseys and addressed in ways that are sensitive, respectful and realistic. Further, satisfaction with discovering an ultimate diagnosis is strongly related to the prior expectations of parents.[52] Even in patients who have a diagnosis, specific therapy is not always available and ongoing therapeutic and management trials are often the mainstay. In both diagnostic and therapeutic journeys, the planning of investigations and interventions must be tailored to the parent's underlying priorities and include communication about expectations and shared goal setting that is realistic. The anticipation of persistent uncertainties in disease manifestations, accuracy of diagnosis, management and prognosis should be incorporated as key topics when guiding families through this process. A parallel focus on non-categorical evaluation and management should attend to the child's functional status, needs, service gaps and the child/family quality of life.[15] Peer support should also be encouraged and facilitated outside of disease-specific networks with efforts to connect families based on geography, disease manifestations or disability.

Parents and physicians often struggle in the determination of their roles and developmental of their therapeutic alliance in the setting of rare or undiagnosed conditions. The reality that at least half of participants in this study had experienced a relationship breakdown with a main physician involved in their child's care, and even more were expressing dissatisfaction with this care, is concerning. There needs to be

fluidity and responsiveness in role distributions with a focus on effective partnership that best utilizes the contribution from parents and physicians. As in the case of other patient groups, communication, engagement and self-management are important areas of focus.

Physicians need to focus attention on roles that do not depend on disease-specific expertise. This is already an impetus in medical education through the CANMeds framework, a comprehensive description of the abilities physicians require to provide optimal care for patients. Many of these closely approximate the physician roles that participants in this study prioritized. The central competency remains that of the Medical Expert . This should not only refer to the provision of diagnosis-specific information and treatment, but should also include the ability to frame, contextualize and explain information that may not fit within familiar diagnoses – or help direct families to other sources of expertise. Another emerging area of physician expertise is in the care of complex and chronic conditions, many of which come with similar challenges, symptoms and comorbidities. Other competencies in CANMeds relate strongly to non-expert roles, such as Health Advocate, Collaborator and Communicator.[106] This study further validates the need for attention to these specific skills, particularly in the context of partnering effectively with the expert parent.

Figure 4.1. CANMeds Competency Framework



Even with improvements in support and development of new models of care, parents will still engage in advocacy, building expertise and managing their child's care. However, there are many existing barriers that inhibit parents from effectively implementing these roles. Parents often do not have direct access to their child's medical information and are not receiving timely responses from physicians, resulting in futile efforts at care coordination. In some cases, health care service eligibility could perhaps be based on need and not on diagnosis. Enabling the full participation of parents whose children have a wide variety of common, rare and undiagnosed

conditions in designing models of care and service delivery will ensure that all of their children's needs will be met.

Finally, an important conclusion from this study stems from the inherent heterogeneity and variability of the study population. Although they share many similarities in terms of their child's condition and their experience of illness, the perceptions and needs of parents varied widely. Much of this variability was difficult to predict or explain even when a number of important features were taken into account. It is not straightforward to elucidate the roles parents have or would like to assume, nor do they have uniform expectations from their child's physicians and other professional caregivers. This underscores the importance of individualization, not only of care and treatments, but of roles and relationships. Through honest, transparent and ongoing discussions, relationships can be formed that will optimize the parents' skills and involvement, while supporting them and providing the services they need for their child.

4.9. Recommendations for future study

These results give valuable insight into the parent perspective and experience with respect to children with ultra-rare and undiagnosed conditions. An important next step is to broaden this patient population to include parents who have not pursued extensive diagnostic testing or for whom it was not available. These parents might have distinct differences in their approach to the child's illness, such as a focus on quality of life and function, often in addition to diagnosis, that should be recognized and supported. Capturing parents at different stages of their child's illness trajectory and diagnostic journey would be valuable, as would following families longitudinally. The needs,

perspectives, roles and responsibilities perceived by parents undoubtedly evolve over time.

A deeper understanding of the role of the physician in the care of ultra-rare and undiagnosed patients requires input from the physicians themselves. Physicians could self-identify challenges and complexities in navigating their relationships with expert parents that may differ substantially from those viewed by the parents. A comparison of how each side conceptualizes the physician's role may point to important discrepancies that could lead to miscommunication and lack of clear expectations. Detailed interviews with physicians, or even paired interviews with parents and their child's physician(s) would be one way to explore these themes in more detail. These insights could give rise to education and mentoring opportunities that would better equip physicians to support families during these long diagnostic odysseys.

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Appendix A: Semi-Structured Interview Template

Semi-structured Interview Framework

Introduction and Context

Social and Family
Work/School

- *Can you tell me a bit about your family and its members?*
- *What does your family like to do together?*
- *Tell me about your child's experience at school and other social activities?*

Chronic Disease

Illness narrative
Stressors and coping

- *How would you describe your child's medical condition(s).*
- *Tell me about how you first learned of your child's illness.*
- *In what way does your child's medical condition impact your life/work/family?*
- *What are some of the things that really helped you?*
- *What have been your major challenges or frustrations?*

The Health Care Team

Access to the health care system
Current communication strategies
Relationships with health care providers
Perspectives on communication
Perspectives on connections

Care Mapping Exercise

- *Tell me about the experience of dealing with your child's illness and accessing care.*
- *Regarding your journey to this point in having a child with special needs, what has worked the best or helped the most?*
- *What have been your biggest challenges?*
- *Is there someone you would describe as a "point person" or an advocate?*
- *What would you change about the story you've told me, if you could?* Would do you think would have helped along the way?
- *What advice would you give others starting this journey?*

Technology Profile

Current use of technology
Perspectives on technology, security, privacy

- *What technology do you use now? In what settings do you*
- *How do you use computers? Cell phones? Other technology?*
- *Do you use computers for online shopping or banking? What do you think about internet privacy and security?*
- *How do you manage your online information (passwords, etc.)? What would make technology more useful to you?*
- *What might be some of your concerns about technology, if any?*

Appendix B: Sample Care Map