INTEGRATING THE RARE DISEASE CONTEXT INTO MEDICAL TRAINING MODELS

by

Celine Bruce-Lepage

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Abstract

Patient centred care (PCC) and evidence-based medicine (EBM) are cornerstones in the education of primary care providers. While teaching these approaches are intended to improve patient care, EBM and PCC were not developed with rare disease (RD) in mind, and therefore the tenets of those models may not hold up as expected in the treatment of RD. In the RD context patients are often more informed about both the disease and illness experience (Babac et al., 2019); there are rarely guidelines to evaluate, and physicians often have very limited knowledge about the disease. This presents a new problem to the current notions of expertise and care. The purpose of this research is to obtain caregiver perspectives about RD healthcare management and how those perspectives can improve RD healthcare by better integrating the RD context into existing models used in medical training (i.e. PCC and EBM). This study used a Qualitative Descriptive Design (Sandelowski, 2000) and data was collected through twelve semi-structured interviews. The analysis and the results, represent a complex interrelated system where a parent must navigate, learn, and manage their way through to secure high quality care. The findings in this study could inform the overall system in RD healthcare.

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Chapter 1

Introduction

My interest in healthcare education began seven years ago, when my daughter was diagnosed with a rare genetic disease. She was diagnosed at six weeks with Prader Willi Syndrome (PWS) a rare genetic disorder that affects approximately one in 15,000 births and like many rare diseases (RD), involves the coordination of several specialists attending to numerous co-morbidities. Leading to her diagnosis, we spent three weeks in the neonatal intensive care unit (NICU), witnessing daily rounds, slowing becoming acquainted with the Canadian healthcare system. Managing my daughter's medical needs enlightened me to the complexities of creating healthcare goals while managing a rare disease. Two years after her diagnosis my interest evolved into patient advocacy, which has provided me with the opportunity to support families across Canada navigating RD healthcare. As a result, I have first and second-hand knowledge of the healthcare climate for RD caregivers. The entry point for the development of this research is intertwined with my lived experience.

Rare diseases (RDs) are defined as conditions that affect fewer than 1 person in 2000, combined there are over 7000 known RDs; as a result, 1 in 12 Canadians are affected (CORD, 2021). Rare Disease (RD) is characterized by its low prevalence and frequently presents with numerous co-morbidities, which require multidisciplinary healthcare (Hannemann-Weber, 2011, Van Groenendael et al. 2015). And yet, RD patients typically experience siloed healthcare, wherein specialists rarely engage with one another (Hay et al., 2020).

Patient centred care (PCC) and evidence-based medicine (EBM) are cornerstones in the education of primary care providers. Sackett et al. (1996) described EBM as "the conscientious, explicit, and judicious use of current best evidence in making decisions about the care of individual patients" (p. 71). According to Stewart et al. (2014), EBM is a method for physicians to evaluate evidence, whereas PCC is a method to ensure patients' experiences and perspectives are included in the development of healthcare goals (p.37). Shared decision-making (SDM) models were developed to achieve PCC by fortifying patient perspectives in healthcare management decisions (Elwyn et al. 2012). While teaching these approaches are intended to improve patient care, EBM and PCC were not developed with RD in mind, and therefore the tenants of those models may not hold up as expected in the treatment of RD.

In standard practice physicians and their patients work together to make treatment goals. EBM requires physicians to use current best evidence, such as treatment guidelines, to make recommendations, on the other hand, illness experience forms the patient's perspective. Both represent valid evidence towards the aim of identifying the problem and goals for treatment. In practice, however, integrating hard science 'evidence' with patient preference represents the final, and elusive step to achieving patient centred care.

In the RD context patients are often more informed about both the disease and illness experience (Babac et al., 2019); there are rarely guidelines to evaluate, and physicians often have very limited knowledge about the disease. This presents a new problem to the current notions of expertise and care. There is little known about how

physicians navigate RD healthcare and treatment planning in the absence of evidence, and even less is known about the patient/caregiver's perspective.

Purpose

Integrating concepts of evidence-based medicine with patient perspective continues to be a challenge in medical training models. EBM emerged as a solution to better clinical decision-making, but the implicitly narrow view of 'acceptable evidence' according to EBM models, effectively marginalizes patient populations that are not reflected in randomized control trials (RCT). There is a scarcity of RCT in RD research, in part, due to low prevalence, therefore evidence in this context must be reconceptualized. Researchers have studied clinical reasoning, decision-making and the flaws which precipitate bad decision-making, but the patient perspective is lacking. The purpose of this research is to obtain caregiver perspectives about healthcare management and how those perspectives can improve RD healthcare by better integrating the RD context into existing models used in medical training (i.e. PCC and EBM).

Research Questions

The following research questions were used to guide my study, which aims to fill the current gap in RD healthcare:

1. How do caregivers of RD patients experience treatment planning in the absence of evidence?
2. How can those perspectives inform the training of EBM and PCC to better integrate the RD context?

Rationale

This research builds on existing medical training model literature and provides a new contribution by capturing the voices of parents caring for children affected by rare disease. Furthermore, findings of this study have the potential to influence policy recommendations for improving medical training models and the implementation of those models in the RD context.

Outline of Thesis

This thesis is comprised of six chapters. In Chapter 1, I provided a description of the factors which led to my interest in medical education and the development of my research questions. In Chapter 2, I describe two foundational concepts of medical education in relation to rare disease (RD) and discuss known barriers to high quality healthcare identified in the literature. In Chapter 3, I discuss the qualitative descriptive design I used in this study, before I move on to the presentation of results in Chapter 4. The results are organized according to the two main themes identified in the study and in Chapter 5, I provide an in-depth discussion of the results. Lastly, in Chapter 6, I provide the implications for research and practice, limitations of this work, and some concluding thoughts.

Chapter 2

Literature Review

In the following literature review, I begin by describing two foundational concepts of medical education: Patient Centred Care (PCC) and Evidence-Based Medicine (EBM). I first discuss PCC, followed by a brief description of Share Decision-Making (SDM) models that emerged from PCC, as well as critiques of their realization. Next, I discuss EBM and its relationship to PCC as well as critiques of their compatibility. Lastly, I introduce the rare disease (RD) context and discuss barriers to high quality healthcare identified in the literature.

Patient Centred Care

Evidence-based Medicine (EBM) and PCC are cornerstones of medical education and yet their compatibility is still heavily debated (Babac et al., 2019; Barratt, 2008; Braschi et al., 2020; Weaver, 2015). Once viewed as a soft science, PCC presupposed a mindset change, wherein physicians were taught to move away from paternalistic approaches, toward collaborative techniques that empower the patient and improve both the objective and subjective experience of healthcare (Stewart et al., 2014). The aim of PCC is to center the patient by empowering them and balancing relations in healthcare decision-making. Patient Centred Care (PCC) has been shown to not only improve patient satisfaction, but also lower testing expenditures, and improve patient outcomes (Dwamena *et al.*, 2012; Epstein et al., 2005; Stewart *et al.*, 2011). Patient-centred care (PCC) has formed the basis of most medical educational curricula since the 1990's, and shared decision-making (SDM) models emerged shortly after that to improve

PCC by giving the patient a central role in decision-making about their clinical care (Stewart et al., 2014).

Shared Decision-Making

Charles et al. (1997), developed a shared decision-making (SDM) framework to improve patient-centred medical decision-making. The framework was created to foster a collaborative process between patient and provider, based on a discussion of options, evidence, and potential benefits and harms. Later, Elwyn et al. (2012, 2017) developed the Three Talk Model (choice talk, option talk, decision talk), which simplified SDM steps making it more user friendly. Shared decision-making (SDM) frameworks are, according to proponents of PCC, most aligned in "finding common ground" (Stewart et al, 2014, p. 32) when patient and practitioners engage in decision-making. Key to the SDM process is the informed patient. Successful execution of SDM involves meaningfully offering choices to an informed patient. However, "acceptable treatment options" (Hasnain-Wynia, 2006, p. 4) are generally decided by the practitioner and in RD scenarios failure to broaden informational searches leave patients few, if any, options to consider. This can exacerbate power dynamics, which often privilege physicians as experts, and the patient/caregiver as unqualified to meaningfully weigh in on complex medical deliberations about treatment planning.

Critics have also claimed there is very little consensus about what constitutes PCC, making its conceptualization, institutionalization, and operationalization difficult to achieve (Dubbin et al., 2013; Tanenbaum, 2015). Tanenbaum (2015) identified four iterations of patient-centred models and explored the compatibility and incompatibly each had as they related to improving the U.S healthcare system. The thoughtful

deconstruction and historical trajectory of each PCC model highlighted the importance of clearly defined terms. Although all four versions of PCC discussed in the paper seemed to place patients at the center, closer examination of each revealed differing priorities. On the one hand, PCC can be used to lower expenditures (Stewart et al., 2011), on the other hand it can be used to generate patient satisfaction (Stewart et al., 2014), the course to arrive at either is unlikely to be the same (Tanenbaum, 2015). PCC has, according to Tanenbaum (2015) changed overtime, which is why adherence to its principles may not be consistently executed.

Critics have also asserted that shifts in modern healthcare demands require providers do more with less resources, and patients have been reconceptualized as "consumers of medical services" (Dubbin et al., 2013, p. 114, Shim, 2010). As healthcare models increasingly moved toward a patient-centred approach, patients have increasingly been expected to participate in their treatment planning. Although, patients with more sophisticated medical knowledge have been able to adapt to the changing landscape, many others do not have the capacity to meaningfully participate (Shim, 2010).

Despite ongoing debates about the defining elements of PCC, Dubbin et al. (2013) argued that the construct of PCC could be captured in three broad domains: "a provider's understanding of the patient's biopsychosocial context, shared understanding of the condition, and shared power and responsibility" (Dubbin et al., 2013, p. 114). Achieving an understanding of the patient's biopsychosocial context is complicated by the patient-provider's "socially structured and differentially distributed resources and competencies" (Dubbin et al., 2013, p. 115). In their review, the authors found that habitus, a concept based on Bourdieu's cultural capital (e.g. habits, skills, even language a person brings to

social interactions) shape both the patient and healthcare provider's (HCPs) biopsychosocial context; moreover, habitus affected patients and HCPs' ability to connect with each other and subsequently the quality of PCC administered (Dubbin et al., 2013, p.116). Finding a shared understanding of the condition was highly influenced by the healthcare provider's impression of the patient and their relative "ability to display biomedical ways of thinking about their health condition" (Dubbin et al., 2013, p. 117). In other words, the quality of PCC was contingent on the providers' impression of the patient's cultural health capital (CHC). Lastly, achieving shared power and responsibility required patients and providers to find common ground in the development of treatment and care planning (Dubbin et al., 2013). The study found that patients needed highly developed CHC to be perceived by their physician as capable of meaningful collaboration (Dubbin et al., 2013, p. 119). The authors concluded that physician bias maintained the power dynamics between patient-providers that PCC was meant to address (Dubbin et al., 2013).

Evidence-based Medicine

Given the above critique, it is not surprising that EBM has become a "widely entrenched notion in clinical medicine across North America and Europe" (Lambert 2006, p. 2639). Evidence-based medicine (EBM) emerged in response to the growing recognition that physicians are not infallible, and treatment interventions should therefore be based on evidence of benefits rather than practitioners "tradition or preference" (Lambert, 2006, p. 2634). The founders of EBM were concerned that physicians were overly reliant on personal experience, when making clinical decisions and sought to "preserve clinical autonomy" (Lambert, 2006, p. 2640) by formalizing systematic

research and the critical appraisal of evidence. Guyatt et al. (1992) claimed that the fundamental principles of EBM were focused on critical appraisal of available evidence, ideally a randomized control trial (RCT), and emphasized that physicians must not only gather the best available evidence, but also attend to the values and preferences of the "informed patient" (Guyatt et al., 1992). When done correctly EBM should encapsulate clinical expertise, systematic research, and the "thoughtful identification and compassionate use of individual patient's predicaments, rights, and preferences" (Sackett et al., 1996, p. 71). Through EBM, physicians would learn how to, not only, find the best available evidence, but also consider the patient particulars and preferences in making those evaluations. However, founders did not explicate how patient particulars were to be brought into the process.

Considering the primacy of evidence in medical decision-making, both proponents and critics of EBM have been concerned with what constitutes 'evidence'. Lambert (2006) analyzed implicit notions of evidence in EBM and the critiques that have emerged as a result. According to Lambert (2006) critiques of EBM reside in 6 categories : 1) evidence derived from randomized trials are not easily applied in other patient populations; 2) privileging gold standard placebo-based trials and the demands to show evidence of effectiveness will necessarily disadvantage interventions and patient populations that are more complex (this included RD populations), and 3) EBM relies on a "strict hierarchy of acceptable forms of evidence" (Lambert, 2006, p. 2635) that excludes the subjective perspective of the patient, 4) EBM essentially codifies and regulates aspects of clinical practice that are already in use (i.e intuition, or expertise) turning diagnostic art or clinical judgement into mindless application of scientific

knowledge, 5) clinical guidelines erode clinical autonomy and effectively limit patient choice; and finally, 6) disseminating and implementing evidence in clinical practice is logistically difficult (Lambert, 2006, p 2634-2636). Put simply, the above critiques can be reduced to two essential claims: First, EBM is not compatible with PCC (critiques 1-3), and second, it is a logistical nightmare (critiques 4-6).

Compatibility of Evidence-based Medicine and Patient Centred Care

Debates regarding the synergy of PCC and EBM have been well explored (Hasnain-Wynia, 2006; Lambert, 2006; Barratt, 2008; Weaver, 2015). According to Hasnain-Wynia (2006), EBM "tends toward efforts to standardize clinical decisions" (p. 6), which effectively reduces individual discretion, whereas, PCC models seek to promote individual discretion and variability in clinical care. Although EBM has been institutionally accepted as a model for practice, critics contend evidence-based approaches are sometimes used to provide "healthcare management and organisations with ammunition to cut costs on grounds of insufficient evidence for certain health interventions" (Lambert, 2006, p. 2639). In complex and uncertain clinical circumstances, practitioners often turn to established protocols and perceive "patients in a stereotypic, formulaic ways" (Weaver, 2015, p. 1077). Critics of EBM claim its practice dehumanizes the patient in healthcare decision-making. Although the founders of EBM explicitly underscored the importance of integrating individual patient preferences (Sackett et al., 2000), guidelines to achieve that end have not been clear. Evidence-based medicine (EBM) frameworks have effectively created a hierarchy of evidence wherein, "epidemiology in making up the 'evidence base'" (Lambert, 2006, p. 2640). Because evidence is narrowly conceived as epidemiological, patient-centred approaches to

evidence, such as Narrative-Based Medicine (NBM) or Cultural Competence Medicine (CCM), are not yet accepted as medically relevant (Lambert 2006; Hasnain-Wynia, 2006). Evidence-based medicine (EBM) uses the patient or 'case' as a starting point when reviewing population-based evidence, however, patient narratives themselves are not considered evidence (Lambert, 2006).

Logistics

Critiques concerned with logistical obstacles to EBM, entail implementation, redundancies, and a disregard for patient perspective (Barratt, 2008; Braschi et al., 2020; Lambert, 2006). There have been persistent debates that EBM leads to "basing clinical decisions on randomized trials, rather than on clinical experience" (Barratt, 2008, p. 408). So-called "cookbook medicine" (Lambert, 2006, p. 2637) erodes clinical autonomy and patient participation in decision-making; On the other hand, the phenomena of 'EBM mimics' shows that the application of EBM does not guarantee against "the distorted use of EBM to support personal opinion (Braschi et al., 2020, p. 176). In the RD context, due to the "low awareness of RDs among physicians" (Bokayeva et al., 2021, p. 91), knowledge itself is often in question.

Patients as experts of their own disease (Bokayeva et al., 2021) have unique disease experiences that do not necessarily align with participant groups in RCTs. An essential step to the successful execution of EBM requires that HCPs create treatment pros and cons according to individualized patient risk profile (Barratt, 2008). Evidence-based medicine (EBM) was not developed to distance physicians from the humanity of their patients, but rather formalize the skills necessary to critically appraise original literature and/or standard approaches. However, EBM's narrow view of 'acceptable'

evidence, effectively marginalizes patient perspectives in decision-making by legitimizing the primacy of RCT.

In the RD context there is usually a dearth of evidence to evaluate, rendering RD patients and their healthcare providers (HCPs) outside the bounds of model EBM. According to Barratt (2008) improving clinical consultation is at the heart of both EBM and SDM, therefore, integrating these two models will lead to achieving that end. All medical decision-making, rare or not, involves some level of uncertainty. Scarcity of guidelines and RCTs in the RD context exemplifies the unpredictability of medical decision-making; however, SDM models could be used to reconceptualize the notion of what constitutes 'best evidence'.

Rare Disease

Rare diseases (RD) are defined as conditions that affect fewer than 1 person in 2000, and combined there are over 7000 known RDs, as a result, 3 million Canadians are affected (CORD, 2021). Studies in healthcare education have focused on improving diagnostic timeframes (Alfaro et al, 2021; Vandeborne, 2019) and RD knowledge gaps (Sarrafpour et al., 2021; Walkowiak & Domaradzki, 2021), which result in HCPs being unfamiliar and ill-equipped to deliver consistent and high quality healthcare. Rare Diseases (RDs) frequently present with numerous co-morbidities, which require a multidisciplinary healthcare team approach. However, RD patients typically experience siloed healthcare, wherein specialists rarely engage with one another (Hay et al., 2020). Due to the multifactorial nature of most rare diseases, RD patients frequently engage in healthcare decision-making in the absence of evidence derived from RCT.

Evidence-based medicine (EBM) and SDM models were not designed with rare disease in mind. According to Stewart et al. (2014), SDM is contingent on patients making informed decisions, and the content of those decisions ought to be evidence-based (Barratt, 2008). However, information in the RD context is scarce, and application of EBM as defined by Guyatt et al. (1992) and Sackett et al. (1996) is unrealistic; integrating SDM into EBM could be the key to synergizing PCC and EBM, as well as improving RD healthcare decision-making.

Babac et al. (2019) examined the implementation of SDM in Germany with an aim to locate potential efficiencies in RD healthcare. The study analyzed the relationship between patients and physicians engaging in medical decision-making and found that successful collaboration in the RD context was contingent on trust. According to the authors, a trusting relationship between patient-physicians was a pre-requisite for effective communication and consequently SDM. Lack of time during the diagnostic process impinged on patient-physician's ability to build good trusting relationships and those issues persisted during disease management. Transparent communication of RD knowledge as well as meaningful engagement "in the form of transfers or time invested" (Babac et al., 2019, p. 11) also emerged as crucial. Patients reported that once diagnosed, information, proper care instructions, and even searching for RD experts, was left to the patients to do themselves (Babac et al., 2019, p.7). Due to physicians' lack of knowledge about RD, patients and their families "preferred to consult other affected families" (Babac et al., 2019, p. 9) and physicians were primarily consulted for general medical advice. Physicians lost their monopoly on health information and instead developed a new role as a "sorter and structurer of available rare disease information" (Babac et al., 2019, p. 11).

In a context rife with uncertainty, it is not surprising that patients tended towards internet-based information. Babac et al. (2019) claimed that HCPs tended toward paternalistic approaches when working with RD patients, because the lack of information in the RD context was analogous to emergent scenarios, which require decisive action. However, integrating patient's knowledge with physicians' professional knowledge could "soften conflict and strengthen the rare diseases network approach at its core" (Babac et al., 2019, p. 12). The expertise developed by patients and their families could be fostered in the SDM process (Babac et al., 2019), allowing physicians to gain a better understanding of the disease.

Rare Disease (RD) patients' approach to information collection was also studied by Kauw et al. (2015). The authors analyzed social media posts to gain insight into why RD patients used online forums and what role they might play in PCC models. Healthcare providers (HCPs) often lack expertise due to the relatively low occurrence rate of RDs, however, virtual information exchanges were shown to effectively make rare diseases less rare. Kauw et al. (2015) concluded that the forums were primarily used for informational support and that the information shared on these forums could be useful in the development of healthcare plans.

Studies investigating RD healthcare have also shown that innovation, networking, and knowledge sharing among HCP represent key interventions in the pursuit to improve RD healthcare (Hannemann-Weber, 2011; Van Groenendael et al., 2015). Stable and consistent healthcare teams, as well as interdisciplinary knowledge sharing in lieu of RCT not only produced better results for patients, but also improved patient's amenability to new strategies (Hannemann-Weber, 2011). Evidence, in the RD

context must be reconceptualized so that treatment planning can be organized in response to the specific patient needs.

In summary, a review of the literature reveals important limitations of the prevailing medical education foundations: EBM and PCC. A review of the literature has highlighted gaps and limitations related to RD healthcare specifically, and patient participation in healthcare management more broadly. Research has demonstrated that although patient preferences are ostensibly part of the EBM approach, in practice, they are not routinely incorporate into the medical decision-making process. There is a strong need for healthcare educators to reflect on the limits of EBM and to re-conceptualize 'acceptable' evidence, especially in the RD context.

Chapter 3

Method

In this chapter, I describe the study design including participants, data collection, methodological approach, and analysis procedures. I finish this chapter with a reflexive statement regarding my positionality.

Participants and Data Collection

I selected participants that had experience with the phenomenon under study and to yield thick and rich data (Morse, 2015). I used a poster (Appendix D) and an email campaign delivered through the patient group, Foundation for Prader Willi Research Canada (FPWRC), to reach my ideal demographic. The following criteria was be used to target parents or caregivers of a child with the rare genetic disorder Prader Willi Syndrome (PWS): (1) They are a parent or caregiver of a child (four years or older) with a genetic diagnosis of PWS; (2) the child is being followed by one or more specialists. Inclusion/exclusion criteria was based on the following factors: A child diagnosed with PWS is typically seen by several specialist, however, referrals can take up to one year and frequency of appointments typically occurs three to six months apart. As a result, a two-year-old patient, for example, may have only been seen by a specialist two to six times respectively.

Data was collected through twelve semi-structured interviews and participants were given a minimum of two weeks to review the transcript of their interview and to add any outstanding thoughts. Although the sample size was relatively small, theoretical saturation (Palinkas et al., 2013) was achieved by targeting caregivers from a homogenous patient population and focusing on PWS specific experiences, which

reduced variation and simplified analysis. This adjustment was necessary because the inquiry under question occurs in a relatively rare context.

Methodology

The present study has followed qualitative descriptive design procedures as described by Sandelowski (2000). Qualitative descriptive design allows researchers to give a "straightforward descriptions of experiences and perceptions…in areas where there is little known about the topic under investigation" (Doyle et al., 2020). Given that the topic involves rare disease healthcare, this approach was appropriate. I chose this design because unlike other kinds of research descriptions, which require the researcher to use theoretical and abstract language when interpreting their data (e.g., phenomenological, ethnographic, narrative, etc.), qualitative descriptive design allows the researcher to present "the facts of the case in everyday language" (Sandelowski, 2000, p. 336). The aim of this method is to achieve descriptive validity by staying close to the data and "to the surface of words and events" (ibid.). As a result, I was able to "present the findings in a way that directly reflects or closely resembles the terminology used" (Doyle et al., 2020, p. 444) by my participants during the coding process and in my results. Qualitative descriptive design also allows researchers some flexibility in data collection and analysis by integrating methods from other qualitative designs to best answer the research question (Doyle et al., 2020).

Interview Protocol

When developing my interview protocol (see Appendix C), I drew on Elwyn et al.'s (2012) Three-Talk Model, a shared decision-making (SDM) model created to improve patient-centred care (PCC) as a sensitizing concept. According to Bowen (2006)

sensitizing concepts are best used as “interpretive devices” or as a “starting point” for qualitative studies (p. 2). The interview guide was piloted prior to the commencement of this study to ensure questions would elicit constructive data. The first question allowed participants to provide some contextual information (i.e. age of child at diagnosis). The second question asked participants to report the number of specialists involved in their child’s care and to reflect upon the overall organization of that care. Participants were asked to consider whether they believed their specialists provided them with adequate information allowing them to engage in informed decision-making (Questions 5); whether they believed their doctors meaningfully included them in their child’s HC decision-making (Question 6) and what the quality of those decision-making conversations were (Question 7). The last three question in the protocol asked participants to reflect on the barriers and burdens of RD HC, and finally what they believed could address those obstacles.

Preparation of raw data files (data cleaning)

Interviews were transcribed verbatim and to save time, Zoom transcription was used during the interview. Following the interview, transcripts were reviewed to assure accuracy and to de-identify contextual information. Upon completion of the transcript, each participant was given a minimum of two weeks to review the transcript and make any necessary edits to ensure their perspective was capture authentically. When transcripts were returned, they were immediately uploaded and stored into NVivo 12 for organization and coding. The first few transcripts and initial coding were shared with my committee to facilitate possible modifications to the interview guide; however, the questions were found to yield good data and the protocol was left unchanged.

Close reading of text

Prior to initiating coding, each transcript was read a minimum of three times, which allowed me to familiarize myself with the raw data. Transcripts were then reviewed question by question to ensure understanding of the overall themes emerging from the responses.

Creating of categories and overlapping coding

Transcript were uploaded and categories were first organized according to interview questions. Initial codes were created based on participant's responses. Once transcripts were coded, and themes began to emerge in each category, I reviewed across categories to find any new findings that may have been missed. Themes were identified using participants language (Sandelowski, 2000) and similar themes were grouped together. There were 76 codes, and eventually codes were refined into two main themes and three subthemes.

Validity, Credibility, Trustworthiness

The following procedures were undertaken for the purpose of rigor and trustworthiness (Lincoln & Guba, 1985). Member checking was used to establish credibility and participants were able to review the transcripts to ensure accuracy; I also used reflective interviewing methods (e.g. reiterating interviewee statements) to ensure my understanding of their experiences were accurate. In order to facilitate transferability of the study results, participants were selected based on their in-depth understanding of the process being studied, specifically the negotiation of treatment goals and healthcare management of PWS. Therefore, only parents or live-in caregivers who met this criterion

were included in the study. Although PWS is a unique genetic disorder, the disease burden parallels many RD trajectories.

Positionality and Reflexivity

I am a candidate in the Master of Education program at Queen's University, Kingston, Ontario, Canada and a parent of a child diagnosed with a rare disease. The purpose of this research is to obtain caregiver perspectives about healthcare management and how those perspectives can improve RD healthcare by better integrating the RD context into existing models used in medical training (i.e. PCC and EBM). Having shared similar experiences with the participants, I am positioned as an 'insider', making me well equipped to understand nuance and implied content. This positionality fostered open, honest, and candid discussion, and greatly improved the feasibility of collecting quality data because respondents were more willing to share their experiences with me, and I was able to draw on my knowledge "about potentially helpful and informative resources" (Berger, 2013, p. 220). The risk of projecting my bias was lessened by the aim of the study itself. Positive and negative perspectives about healthcare management are both useful toward the objective of using caregivers' perspectives to better integrate the RD context into existing models used in medical training. Therefore, I was not pursuing a specific perspective about healthcare; all data was welcome.

Ethical Considerations

This study received ethical clearance from the General Research Ethics Board (GREB) at Queen's University. The clearance letter provided by GREB for this study is found in Appendix A. The Letter of Information/Consent Form is located in Appendix B.

To maintain confidentiality, participants are identified numerically to ensure anonymity. Upon completion of member checking, participants were given a $15 Starbucks card in thanks for their participation. Information was securely stored under password protection in online and electronic contexts and a USB copy was secured in a locked file drawer when not in active use.

Chapter 4

Results

Twelve participants were interviewed as part of this thesis and asked to share their experiences in rare disease (RD) healthcare (HC) management and treatment planning. The terms used in the results are a reflection of participants description of their healthcare experiences. The terms specialist, doctor, and physician are used interchangeably in reference to medical physicians that work under individual specialties. Specialists were defined as medical physicians who specialize in a particular field, such as endocrinology, orthopedics, neurology, respirology, and other related disciplines. Additionally, when discussing coordination of care, the participants considered the various specialists working with their child as members of their healthcare team (HC team). The following table provides a summary of the participants, the age of their child at the time of interview, and specialists for those that disclosed.

Table 1

Participant Demographics

Participants	Caregiver	Age of Child	Number of Specialists	Disclosed Specialist
P 001	Mother	6	2	Endocrinologist, Paediatrician
P 002	Mother	11	7	Endocrinologist, Neurologist, Ophthalmologist, Orthopedist,

Participants	Caregiver	Age of Child	Number of Specialists	Disclosed Specialist
P 003	Mother	11	6	Otolaryngologist, Paediatrician, Respirologist Endocrinologist, Neurologist, Neuro-psychiatrist, Ophthalmologist, Orthopedist, Pediatrician
P 004	Mother	9	5	Endocrinologist, Dentist, Orthopaedist, Paediatrician, Respirologist
P 005	Mother	15	10	Not disclosed
P 006	Mother	16.5	4	Endocrinologist x 2, Orthopedist, Respirologist
P 007	Mother	4	9	Not Disclosed
P 008	Mother	7.5	4	Endocrinologist, Neurologist, Ophthalmologist, Respirologist
P 009	Mother	6	2	Endocrinologist x 2
P 010	Mother	14	3	

Participants	Caregiver	Age of Child	Number of Specialists	Disclosed Specialist
P 011	Mother	16	3	Endocrinologist, Orthopaedist, Respirologist
P 012	Mother	4	5	Endocrinologist, Paediatrician, Psychologist
				Endocrinologist, Gastroenterologist, Orthopedist-hips, Orthopedist-spine, Pediatrician

The analysis and the results, represent a complex interrelated system where a parent must navigate, learn, and manage their way through to secure high quality care. There were two main themes with three subthemes identified and presented in this section: The first theme, *Lack of Coordination*, included the subthemes *Coordination Through HC Team Collaboration, Coordination Through Collaboration with Parents* and *Coordination as Accessing Expert Opinions.* The Subthemes *Coordination Through Collaboration with Parents* and *Coordination as Accessing Expert Opinions* also fit under the second theme of *Inadequate PWS Knowledge*. In what follows, results are organized by theme (*Lack of Coordination* and *Inadequate PWS Knowledge)* followed by

recommendations for improving RD HC and lastly, highlights of complexities that became apparent during analysis.

Lack of Coordination

All twelve participants expressed concerns about the inadequacy of PWS knowledge (systemic and individualistic) and lack of coordination in HC (systemic and individualistic). Coordination was discussed as three separate but interrelated themes: The first (*Coordination Through HC Team Collaboration*) concerned a global approach to HC, wherein the particularities of disease management are meaningfully incorporated into the care through interdisciplinary collaboration. The second (*Coordination as Collaborating with Parents*) concerned the role parents play in RD HC management as both lay experts of the RD and experts about their own child. The third (*Coordination as Accessing Expert Opinions*) concerned PWS knowledge and the importance of collaborating with experts to ensure that HC decisions and treatment planning are based on best practice and the best available evidence. Subthemes two and three are interrelated and are therefore discussed concurrently.

Coordination Through Healthcare Team Collaboration

All but two participants described frustrations about the lack of communication between their specialists; of the two participants that did not (P 001 and P 009), both had recently lost a member of their HC team and were focused on their gap in care. A global approach to HC is described as treating the whole patient as opposed to individual morbidities. Participants in this study described fragmented care plans which largely flowed from a siloed HC approach. One participant explained:

> "[My son]…was followed actively by almost nine specialists, and every time we saw one of them...it wasn't about, you know, the global aspects it was about, you know, one specific point, one symptom that we were treating. So, we're not treating Prader Willi" (P 007).

Siloed HC is commonly experienced in the RD context and can be ameliorated through policy and administrative improvements, such as requiring team meetings (Hay et al. 2020). Cross-disciplinary communication have been used with success in other areas of RD HC (Gall et al., 2022; Geng et al., 2021; Van Groenendael et al., 2015,), however, according to participants in this study, interdisciplinary communication rarely occurred:

> "There is no network (laughs). They all work individually" (P 002).
>
> "That labour is falling on the parent to be the communication gateway to another person in the care" (P 008).
>
> "I'm the link basically and if I…if I don't tell them what happened in this or this or this uh…they don't know" (P 002).

This seemingly innocuous failure to collaborate using an interdisciplinary approach created a role for parents, which I have termed the *Quarterback.* The *Quarterback* is a coordinator, an advocate, a researcher, and in this case intermediary communicator.

Participants believed that siloed care was, in some instances, evidence of an overtaxed system and at other times, due to individual practitioner practice. For instance, four participants described attending multiple clinics that had centralized communication tools, allowing their specialists to view clinic notes and requisitions (e.g., blood work or scans) from other clinics, however, those files were not routinely accessed:

> “He's like yeah we could check any clinics that you been at in this hospital. I was like and in like 11 years nobody has ever done that” (P 003).
>
> “The parent is required to be the manager of these services. You have to keep track of when you've seen so and so, and when you're required to follow up” (P 012).

Concerns regarding lack of coordination ranged from being inconvenient to speculation that potentially significant HC matters were being overlooked.

Coordination as Collaborating with Parents and Accessing Expert Opinions

PWS is associated with a higher incidence of scoliosis (i.e. prevalence rages from 15% to 86%, Crinò et al., 2022); nine of the twelve participants were followed by an orthopedic specialist for scoliosis and/or orthotics. Despite their children being followed for PWS related morbidities, participants found that cross-disciplinary communication and accessing expert opinions were largely left to parents to coordinate. Participants agreed that the quality of HC was improved when specialists coordinated with PWS experts. However, efforts to access expert opinions varied according to individual practitioner practice.

In most cases first contact with PWS experts was initiated by participants. Participants had concerns about the lack of coordination and/or poor disease knowledge their doctors demonstrated, and the effects those deficits had on the quality of care their child was receiving: “I'm in charge. Like I’m in charge of all of it, and that, I feel, is the burden to me sometimes. Like it's hard. It's really hard” (P 006). Participants did not expect their doctors to have expert knowledge about PWS, but they did want their doctors

to acknowledge the limits of their knowledge and take on the labour of coordinating with experts once they were located.

Inadequate Prader Willi Syndome Knowledge

Some participants (P 006 and P 007) did experience a level of coordination, wherein specialists had knowledge of other providers on their team, however, insufficient PWS knowledge exacerbated the siloed effect of the care. The low prevalence of PWS, according to participants, led to significant variability in individual practitioner knowledge. Participants that *Quarterbacked* their child's HC ensured connections between disciplines were made and mitigated poor disease knowledge by locating expertise when necessary:

> "Being responsible for everything, as the mother. Uh, worrying you're going to miss something. Worrying that you, you only have a small window of time to try and make change" (P 005).

Ten of the twelve participants sought expert opinions and/or care outside of their local communities. Several participants travelled to the U.S. to meet with a world-renowned expert in PWS (who will be referred to as Dr. PWS for the remainder of this discussion), and others accessed expert opinions remotely. Two participants (P 008, P 010) were advised by their local doctor to travel for PWS specific expertise, whereas other participants sought expertise to mitigate inadequate PWS knowledge demonstrated by their specialists. All twelve participants described feelings of frustration and stress from working with doctors that had poor disease knowledge and the impact the lack of knowledge had on the quality HC they experienced. Participants described the burden of keeping their doctors informed to ensure their children were receiving adequate HC:

> “I mean she knows how to be a doctor, but she doesn't know the nuances of Prader Willi and when we start delving into behavior or hyperphagia…doesn't know how to manage that. She doesn't know what to tell me to do” (P 003).
>
> “Always explaining and telling the doctors what the symptoms are rather than them saying, oh, she’s got PWS, and here's what we can do with that type of thing” (P 009).

Inadequate PWS knowledge was another factor, which precipitated the need for parents to take up the role of the *Quarterback.*

Individual practitioner practice and the effort to improve disease knowledge improved HC experiences. Two participants (P 001, and P 011) believed that despite their specialist having limited knowledge about PWS at the time of their first meeting, a demonstrated interest in developing their knowledge provided the space needed to work collaboratively:

> “She was a bit resistant...and she was very frank and said I’m just not completely comfortable, but I’m willing to do it if that's what you really want...once that started everything was fine, there was no more resistance” (P 001).

On the other hand, specialists that showed a lack of interest or, worse, refused to improve their disease knowledge worsened HC experiences. Nine participants described circumstances, which led them to change doctors due to inadequate PWS knowledge. One participant, who traveled to the U.S. to meet Dr. PWS, was disappointed when her Canadian endocrinologist refused to collaborate: “I went in with a binder, and it was all tabbed out with, here's what we're doing and here's dosages, and here's...and here's the

research. And here's what [Dr. PWS] says, and here's what so and so it says, and she wouldn't even look at it. She wouldn't even read it" (P 012).

Similarly, another participant, who took part in a pilot project for coordinating HC, described her frustration when her son's physician refused to participate in the project:

> "the endocrinologist, who's supposed to be the most involved, never came...we had two meetings, never came to either one of them. Was also given an opportunity to phone this specialist in the United States, and her time would be paid. She refused to do that" (P 005).

Participants felt bewildered when doctors refused to develop their PWS knowledge, which ultimately left parents with a sense of skepticism about the quality of HC their child was receiving.

Physicians with poor disease knowledge forced parents to fiercely advocate for their child's HC. When acting as the *Quarterback,* parents would engage in research to ensure their doctors were informed and that their medical advice was up-to-date: "How safe is it to be discussing medical interventions if you don't understand the underlying problem with the…disease and disorder" (P 005). Participants believed that the necessity of *Quarterbacking* was a failure in HC coordination at the systemic level (standard of care and guidelines for PWS have not been effectively disseminated in the HC sphere) and at the individualistic level (those guidelines and standards could be access by doctors, who choose to contact PWS experts and/or patient advocacy groups).

> "The fact that we have to do the research to find things out. It's really, really stressful. I feel like I fail all the time because I'll find out something new that

may...to me is new, but I didn't know about it. No one mentioned to me. It's like, only if I go seeking it, and you really have to stay on top of it" (P 010).

Participants believed that doctors that were unwilling to pursue PWS knowledge, increased the caregiver burden and maintained the need for parents to advocate and engage in additional labour as the *Quarterback* of their child's care:

> "It's really exhausting to...present yourself as the, um, expert like the informed, you know, researcher in the room, when really you're just a parent who wants to get care for your kid" (P 008).

However, participants also believed that working with doctors that did not have PWS expertise, but were willing to learn and increase their PWS knowledge decreased their overall burden.

Meaningful Improvements to Rare Disease Healthcare

Participants described the work of managing their child's RD HC as arduous. Despite having no formal training or authority, participants developed a set of sophisticate skills to ensure their child had the best care possible. Parents barely had a chance to process the heartbreak of their child's diagnosis before transforming into the *Quarterback,* responsible for coordinating, researching, and advocating for high quality healthcare.

When participants were asked to reflect on what changes would improve their experiences with HC management and treatment planning, their responses reflected the above discussed themes. Knowledge sharing coordination through team communication, accessing expert opinions, and meaningful involvement of parents were all suggested to improve HC delivery.

All twelve participants shared experiences about working with doctors that were uninformed and/or uninterested in learning about PWS. There was a sense from some participants that when physicians demonstrated inadequate PWS knowledge it was evidence of poor preparation: “I would say that before they enter a room, they go look up the syndrome, and it's pretty easy to do that” (P 005); others believed it was due to low prevalence and lack of resources. Participants believed that a centralized standard of care or guidelines would be instrumental in mitigating both issues of disease knowledge and communication. Participants suggested that guidelines created and updated by PWS experts would greatly benefit patients followed by specialists that lack sufficient knowledge due to inexperience and/or disinterest.

Due to the low prevalence of PWS, participants also believed that centralizing guidelines would improve care in areas that are less populated, and standard of care is poorly disseminated. One participant suggested that if physicians had “a milestone line that shows different things that might happen along that milestone within the child's life and...the different types of treatment available...that would be a very holistic view of things” (P 001). Another participant suggested a “one pager that they share with their team, and if you do have a patient with Prader Willi Syndrome, you just read the page before you meet the patient” (P 007).

Participants also proposed that administrative and logistical *Quarterbacking* tasks ought to be under the purview of a HC coordinator. This coordinator would be the team leader and facilitate cross-disciplinary communications and ensure that members of the HC team were collaborating and consulting with experts when necessary. Guidelines and the implementation of a team coordinator would facilitate cross-disciplinary

communications as well as cross-border communications thereby ensuring PWS patients benefit from expertise from around the world.

Improved coordination was theoretical for all but one participant, who had the opportunity to participate in a pilot project providing HC coordination. Throughout the pilot project, the participant was able to experience cross-disciplinary collaboration between her son's physicians, as well as coordination with PWS experts:

> "That's been the most powerful intervention for us. Is when we had all those players community-based and medical, hospital-based people got together in one room and talked about the idiosyncrasies of Prader Willi Syndrome, and the effect it was having on [my son's] health" (P 005).

Ultimately, improving the knowledgeability of specialists about PWS was found to be essential to improve HC for this patient population.

The final recommendation made by participants was for meaningful collaboration with parents. Parents are often more familiar with their child's RD than the clinicians they are working with. Participants believed that doctors ought to respect the unique set of skills parents bring to the table. Although the RD context frequently means there is a lack of good available evidence, this was not always the case, according to participants; parents found that there was PWS specific research and experts to locate, the challenge was getting their doctors to access those resources. Parents agreed that meaningful involvement in their children's HC decisions was essential, but the burden of medical expertise should not also rest on a parent's shoulders. Specialists must improve their contribution to treatment planning by researching current literature, as well as

interdisciplinary knowledge sharing with both PWS experts and other members of the HC team:

> "I'd like to know what I don't know" (P 001).
>
> "I would love a situation where I'm, I'm learning from doctors as much as they're learning from me" (P 008).
>
> "We don't know what we're missing with our daughter's care because we don't know everything, and we have to do the research" (P 010).

Acting as *Quarterback* gave parents a sense of purpose but was also exhausting. Participants expressed a sense of satisfaction in their ability to secure the best care for their children, but also struggled with the knowledge that without their ongoing interventions their children's HC would undoubtedly suffer. One participant noted: "If I did not take on that role of informed advocate, quasi, like, amateur researcher (laughs)...my son would have no care." (P 008).

Highlights of Complexities

Two major interrelated threads were identified across the themes and can be considered barriers to adequate HC: System related barriers and individual practitioner practice related barriers. System related barriers are features of the healthcare that were often viewed by participants as hassles and burdensome aspects that needed to be worked around or overcome to support their child's HC needs. Individual practitioner practices represented the spectrum of individual healthcare practices that enabled and/or hindered care. Participants often had to navigate both the system and the individual specialist's process, and in many instances individual practitioner practice was shown to improve or worsen the systemic barriers. As such they appear to be intertwined in the care of PWS.

The findings in this study revealed that parents of children with PWS routinely took on responsibilities commonly held by physicians; as such, improving HC experiences would require a more equitable balance of responsibilities. All twelve participants ended the interview with an expression of gratitude for the opportunity to share their stories and an expression of hope that meaningful improvements might be made to RD HC management. In the following chapter, I discuss the findings in relation to the existing literature and revisit the noted complexities in greater details.

Chapter 5

Discussion

The focus of this thesis was to investigate how parents of children with rare diseases (RDs) experience the practice of healthcare (HC) and how those perspectives might inform the training of physicians and improve RD HC. The two main themes identified in this study were: *Lack of Coordination* and *Inadequate PWS Knowledge*. This chapter is comprised of five sections, beginning with a discussion of the findings in relation to the literature, followed by a discussion of the complexities of healthcare management and treatment planning for families living with RD, the limitations of this research, the implications for research and practice, and concluding thoughts.

Issues of coordination and knowledge both created additional labour for parents, which led to participants taking on so-called *Quarterbacking* tasks. These findings reflect Currie and Szabo's (2019) work, which also investigated challenges faced by parents of children living with RDs. Currie and Szabo (2019) found that lack of RD knowledge along with the lack of coordination between healthcare providers' (HCPs) and community services significantly increased the burden of care for parents. Similar to the findings in the present study, Currie and Szabo (2019) also found that physicians were at times insufficiently prepared for scheduled appointments, including failing to do recent literature searches prior to meeting patients.

Gaps in physician knowledge about rare diseases have been linked, in part, with insufficient preparation on the part of both medical students and professionals (Bokayeva et al., 2021; Currie & Szabo, 2019). However, a more predictable factor is the lack of representation of rare diseases in medical curricula and postgraduate training (Walkowiak

& Domaraddzki, 2021). Most RDs are not part of the general medical education curriculum. Parents and patients with RDs often find that they are the experts of their own condition and physicians that admit to the limits of their knowledge, but also "make the appropriate contacts to compensate" (Babac et al., 2019, p.5) for those gaps are able to nurture a trusting relationship and provide high quality care. Lack of PWS knowledge provoked several participants to contact experts for PWS specific insights into their children's HC plan. However, as was reflected in the work of Currie and Szabo (2019), parental contribution to healthcare planning was not always welcomed by physicians.

The tenets of evidence-based medicine (EBM) require that physicians use current best evidence, such as treatment guidelines, to ensure their recommendations are not overly reliant on personal experience (Lambert, 2006). Participants that experienced paternalistic and/or adversarial decision-making conversations, suspected that specialists were more concerned with maintaining decision-making power than they were with pursuing the best outcomes. A 50-year content analysis of collaboration-related articles from the journal - *Medical Education* - found that despite collaboration being heralded as essential to improving HC outcomes, the language used in those articles regularly perpetuated existing hierarchies that impede collaboration (Paradis et al., 2017). According to the authors, there is an intrinsic tension between the goal of instituting collaboration and the preservation of existing hierarchies between physicians and so called "non-doctors" (Paradis et al., 2017, p. 869). The tendency to prioritize existing hierarchies has also been found to extend to the type of evidence legitimized by EBM frameworks (Lambert, 2006).

Patient Centred Care (PCC) models champion the meaningful inclusion of patients' experiences and perspectives, when developing healthcare goals (Stewart et al., 2014). In paediatric care parents, as advocates, contribute to the patient perspective. However, pedagogical approaches to medical education also tend to reinforce siloed approaches to healthcare and impede coordination (Stewart et al., 2014). Discipline-specific approaches to teaching make it difficult for medical students to "think like members of an interprofessional team" (Steward et al., 2014, p. 288). Instead, medical students are encouraged to "accept a major responsibility for decision-making and to see themselves as team leaders" (Ibid., p. 287). Scholarship that examines collaboration further reinforces this siloed approach by continuing to position doctors as implicitly or explicitly "the leaders of teams and in ultimate control of patient care" (Paradis et al., 2017, p. 868). As is shown in the present study, discipline specific HC goals that disregard a global approach to patient care tend to minimize the parent's perspective and the quality of HC decreases.

Failure to coordinate care was viewed by participants as evidence of both systemic and individualistic barriers. Participants suspected that their specialists were unwilling to take up the role of coordinator because they viewed their contribution to the care plan as specific to their specialty. This absence of leadership resonated with Geng et al.'s, study (2021), which concerned consultative medicine for patients with complex conditions involving multi-organ systems. According to the authors, primary care providers (PCPs) tended to "defer to specialist for their expertise, while specialists may assume the PCP [would] drive the development of a cohesive plan of care" (Geng et al., 2021, p. 2479). Participants in the present study found that neither their specialist nor

their PCP were able to act as coordinator, which left parents to take up the labour of coordination themselves or suffer the consequences of fragmented care.

Social-Economic Status and Gender

Social-economic status (SES) and gender were not the primary focus of this study, yet it is important to acknowledge their representation in the sample. All twelve participants were mothers and most of the participants believed their SES significantly affected their ability to effectively Q*uarterback*. The gendered representation of the sample is perhaps unsurprising considering that caregiving task are often viewed as part of "women's work" (Revensen et al., 2016). All twelve participants were mothers; some worked in HC and/or had a science background, two participants had PhDs, another worked for disability support services. Three participants left professional careers because the coordination, research, and advocacy required to *Quarterback* required a full-time commitment. In short, the participants in this study were privileged enough to be pursue adequate HC. The present study is consistent with previous findings (Abel 2008; Dubbin et al., 2013; Gentler, 2014; Gage-Bouchard; 2017, Hardeman et al., 2015; Missinne et al., 2014; Shim 2010) that showed links between SES and HC outcomes. Participants believed high quality HC was contingent on their ability to effectively *Quarterback,* and physician's responses to their *Quarterbacking* skills elicited better or poorer healthcare experiences.

Why should instruction emphasize coordination?

Although, issues of collaboration and coordination in HC are not specific to RD, they are particularly salient in this context. When participants believed their specialists demonstrated inadequate PWS knowledge and/or failed to coordinate with other members

of their HC team, the burden of *Quarterbacking* increased. Participants believed they were responsible for compensating shortfalls in critical communications between their specilialists and routinely found that initiating contact with PWS experts was largely left to parents as well. Coordination in RD HC not only improve patient outcomes by minimize common issues that result from physicians' relative unfamiliarity with RDs (Van Groenendael et al., 2015), but also decrease parents' stress around the coordination of that healthcare (Gall et al., 2022).

According to the principles of EBM, HCPs are expected to use the most current and reliable evidence available to make medical decisions. Pioneers of both PCC (Stewart et al., 2011) and EBM (Guyatt et al. 1992; Sackett et al. 2000) prescribed integrating patient's perspective and preferences into medical decision-making processes. Given that up-to-date RD knowledge often requires accessing information from formal and informal resources (Bokayeva et al., 2021), parents could prove to be useful knowledge brokers "linking researcher with knowledge users" (Donnelly et al., 2014, p. 39). Although RD is not explicitly covered by the above-mentioned models, the findings in this study have revealed that the pathway to providing patient-centred EBM may not be so different in the RD context.

The findings in this study can inform the overall system in rare disease healthcare and can be summarized in three points. First, *Quarterbacking* task described by participants represent a significant burden for RD caregivers, which not only created logistical inconveniences (e.g., trying to keep various specialists informed about the global picture of HC), but also led to significant distrust about the quality of the HC itself.

Second, participants attributed failures to provide high quality RD care with system related barriers and individual practitioner practice related barriers. For instance, participants believed that system related barriers such as limited resources and lack of time likely impacted the quality of care their specialist could reasonably offer. However, when individual specialists referenced in this study failed to perform perfunctory literature searches and were resistant to updating their disease knowledge and/or working with disease experts, they effectively created obstacles to quality HC.

Previous research has linked poor RD care with insufficient preparation on the part of both medical students and the physicians that train them (Bokayeva et al., 2021, Currie & Szabo, 2019) as well as pedagogical approaches to medical education (Stewart et al., 2014), including the underrepresentation of RDs in medical curricula (Sarrafpour et al., 2021; Walkowiak & Domaradzki, 2021). Although medical encounters that lack clear evidence do demand more from physicians, high quality care is still possible with effective collaboration and coordination.

Lastly, the themes and proposed changes to RD HC identified in this study are supported by the tenets of PCC and EBM, and represented in the 2015 CanMEDS framework. The framework, identifies six roles that physicians ought to embody in order to achieve the integrated role of *medical expert*; those roles are *communicator, collaborator, leader, health advocate, scholar,* and *professional* (Frank et al., 2015). The realization of high-quality care in the RD context does not involve reinventing the wheel, but rather a reinforcing this frameworks. Meaningful collaboration between specialists and parents (Babac et al., 2019; Bokayeva et al., 2021), as well as coordinated interdisciplinary collaboration within the HC team and with disease experts (Curie &

Szabo, 2019; Gall et al., 2022) are essential to improving HC experiences and lessoning the burden on parents. While these three points are intended to inform RD HC, enacting will enhance the overall quality of care for patients and strengthen the healthcare system.

Chapter 6

Future Directions

This study investigated how parents of children with rare diseases (RDs) experience the practice of healthcare (HC) and how those perspectives might inform the training of physicians and improve RD HC. However, there are some limitations that need to be considered in future research. First, participants in the sample were homogenous (Caucasian mothers with post-secondary education) and therefore do not represent the broader population. As such, the barriers and suggested remedies would likely differ somewhat in a more diverse sample. Second, the perspectives captured in this study were of the parents alone and did not include physicians' viewpoint. Given that participants believed individual practitioner practice could increase or decrease caregiver burden, a better understanding the physicians' perspective would be of benefit. Lastly, therapist (i.e., Speech and Language Pathology, Occupational Therapy, and Physiotherapy) are integral members of a HC team supporting a child living with PWS. However, participants were directed to share perspectives and experiences from encounters with medical physicians. Given the importance of therapy in the development of a patient centered healthcare plans, failure to capture those stories is a clear limitation.

Implications for research and practice

Future research should include more diverse samples to better understand how barriers and burdens may differ for people that are racialized and/or are marginalized due to their socio-economic status. Additionally, future research should capture the voices of all members of the HC team including therapist, specialists, and caregivers to better understand the various factors that lead to gaps in care. The present study corroborates

previous findings (Babac et al., 2019; Bokayeva et al., 2021; Budych et al., 2012; Curie & Szabo, 2019; Gall et al., 2022; Geng et al., 2021; Van Groenendael et al., 2015) that coordination is essential to delivering high quality care in the RD context.

Ideally, future studies will draw on the work of Donnelly et al. (2014) and Gall et al. (2022), to create coordination and collaboration-based HC interventions that can be modified and attuned to the RD context. The results of this study provide a clear, and yet, narrow view of what is needed to improve RD HC. The suggestions made by parents could, ostensibly be implemented by physicians individually, however, systemic barriers experienced by those same doctors have not been meaningfully investigated. A knowledge translation-informed evaluation model (Donnelly et al., 2014) combined with a coordination model (Gall et al., 2022) could address logistical limitations experienced by physicians as well as "facilitate the integration of patient-specific and contextually based knowledge" (Donnelly et al., 2014, p. 52). Possible pedagogical approaches to medical education could incorporate patient-present teaching (Chema et al., 2021), which directly aligns with one of the three recommendations made by participants in this study, namely greater collaboration with parents. Another useful approach is simulation exercises that include role-play with patient educators (Sanges et al., 2020). Involving patients in the medical education process enhances patient centredness (Chema et al., 2021) and allows students to learn about "clinical signs that are not easily reproduced" (Sanges et al., 2020, p. 7) as well as the psycho-emotional aspects of that disease.

Conclusion

Although I have recommended that the voices of physicians be captured in future research, interventions aimed at improving HC should always center the perspectives of

patients and caregivers as the ultimate users of those services. The recommendations made are deliberately one-sided because the participants in this study are already contributing significantly to the HC management of their children; shortfalls in HC did not flow from lack of effort or compliance on the part of these mothers, but rather systemic and individual practice related barriers. Physicians working with complex and RD patients may need to shift their approach from presumed expert to collaborator. I would also argue that there needs to be greater emphasis placed on the application of PCC and EBM in the RD context throughout medical training. Physicians working with RD pateints may need to rely more heavily on consultative processes to provide high quality care.

The recommendations made by the participants in this study align with the tenets of PCC and EBM and are already required by the college of physicians as outlined in the current CanMEDS framework (Frank et al., 2015). Scholars have attributed the marginalization of patients' perspectives with the emphasis EBM models place on the primacy of RCTs (Lambert et al., 2006), others have shown that power dynamics between doctors and patients have the tendency to complicate authentic and informed SDM conversations (Braschi et al., 2020, Stewart et al., 2014); both explanations substantiate there is a need for RD context specific training of these models.

When physicians learn they have a patient with a rare disease, a thorough review of the case and literature review as well as efforts to locate an expert may be necessary to deliver high quality care. Patients with RDs, like most patients, are not homogenous. Rare diseases like PWS are spectrum disorders, meaning each case will bring its own complexities and dynamics, including disease presentation and even reactions to

medication. When you add individual family dynamics and SES to the equation, the incompatibility of using a formulaic approach becomes clear. Training physicians in the RD context will therefore require personalized individualized medicine and a consultative approach.

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Appendix A

GREB Clearance Letter

June 20, 2022

Ms. Celine Bruce-Lepage
Queen's University

Title: "GEDCU-1109-22 Medical training models in the Rare Disease Context;" TRAQ # 6036328

Dear Ms. Bruce-Lepage:

The General Research Ethics Board (GREB), by means of a delegated board review, has cleared your proposal entitled " **GEDCU-1109-22 Medical training models in the Rare Disease Context** " for ethical compliance with the Tri-Council Guidelines (TCPS 2) and Queen's ethics policies. In accordance with the Tri-Council Guidelines (Article 6.14) and Standard Operating Procedures (405), your project has been cleared for one year.

You are reminded of your obligation to submit an annual renewal form prior to the annual renewal due date (access this form at http://www.queensu.ca/traq/signon.html/; click on "Events;" under "Create New Event" click on "General Research Ethics Board Annual Renewal/Closure Form for Cleared Studies"). Please note that when your research project is completed, you need to submit an Annual Renewal/Closure Form in Romeo/traq indicating that the project is 'completed' so that the file can be closed. This should be submitted at the time of completion; there is no need to wait until the annual renewal due date.

You are reminded of your obligation to advise the GREB of any adverse event(s) that occur during this one-year period (access this form at http://www.queensu.ca/traq/signon.html/; click on "Events;" under "Create New Event" click on "General Research Ethics Board Adverse Event Form"). An adverse event includes, but is not limited to, a complaint, a change or unexpected event that alters the level of risk for the researcher or participants or situation that requires a substantial change in approach to a participant(s). You are also advised that all adverse events must be reported to the GREB within 48 hours.

You are also reminded that all changes that might affect human participants must be cleared by the GREB. For example, you must report changes to the level of risk, applicant characteristics, and implementation of new procedures. To submit an amendment form, access the application by at http://www.queensu.ca/traq/signon.html; click on "Events;" under "Create New Event" click on "General Research Ethics Board Request for the Amendment of Approved Studies." Once submitted, these changes will automatically be sent to the Ethics Coordinator, GREB, at University Research Services for further review and clearance by GREB or the Chair, GREB.

On behalf of the General Research Ethics Board, I wish you continued success in your research.

Sincerely,

Professor Dean A. Tripp, PhD
Chair, General Research Ethics Board (GREB)
Departments of Psychology, Anesthesiology & Urology
Queen's University

Appendix B

Letter of Informed Consent

Study Title: Integrating the Rare Disease Context into Medical Training Models

Name of Principal Investigator: Celine Bruce-Lepage, Faculty of Education, Queen's University

Name of Supervisor*:* Dr. Saad Chahine

I am inviting parents and caregivers of children with PWS to take part in a research study. The purpose of this project is to improve the training of physicians by gathering patient perspectives about healthcare management and to integrate the rare disease context into existing models used in medical training (i.e. Patient Centred Care and Evidence Based Medicine).

If you agree to take part, I will interview you for 30-45 minutes using Zoom. The interview will be audio/video recorded and later transcribed verbatim. There are no known risks to participating. If you feel upset after the interview, please call Crisis Services Canada at 1-833-456-4566 or Text 45645. There are no direct benefits to you as a participant. You will receive a $15 Starbucks gift card for participating. Following the interview you will be sent a copy of the transcript and asked to review and add or remove any other thoughts you may have. Once completed and returned, you will receive the gift card via email.

Participation is voluntary. You do not have to answer any questions you do not want to. You can stop your participation at any time by telling me you would like to stop or simply closing the video conferencing application. Stopping your participation will have no negative impact on you. You may request to have your data withdrawn from the study up until you complete your review of the transcript by contacting me at celine.bl@queensu.ca.

Your confidentiality will be protected to the extent permitted by applicable laws. I will do this by replacing your name with a pseudonym in all study records. Direct quotes may be used as part of research dissemination and we will do our best to de-identify contextual information. However, there is always a possibility that you could be identified through your quotes. To mitigate this possibility, you will have a chance to review your entire transcript as part of the research process, at which point you can amend any of the content of your transcript.

The study data will be stored on an encrypted hard drive on Queen's University servers. Your real name will never be linked to your pseudonym in any file. Access to study data is limited to myself, my supervisor Dr. Saad Chahine, and my committee member Dr. Ian Matheson. Additionally, deidentified data may be used in publications, conferences and

other dissemination activities. GREB is bound by confidentiality and will not disclose any personal information.

I will keep your data securely for at least five years per Queen's University Policy, after which all recordings, transcripts, and data will be destroyed. The code file identifying your pseudonym and study ID number will also be destroyed five years after study closure.

I plan to publish the results of this study in academic journals and present them at conferences. I will not include any personally identifying information from the interviews when presenting my findings. I will never include any real names with quotes. I will make every effort to ensure quotes do not identify participants. During the interview, please let me know if you say anything you do not want me to quote.

The Queen's General Research Ethics Board (GREB) may request access to study data and/or all other study materials used in this research to ensure that we (the research team) have or are meeting our ethical obligations in conducting this research. GREB is bound by confidentiality agreements and will not release any personal information.
If you have any ethics concerns please contact the General Research Ethics Board (GREB) at 1-844-535-2988 (Toll free in North America) or email chair.GREB@queensu.ca. Call 1-613-533-2988 if outside North America. If non-English speaking participants wish to contact the Chair for ethics concerns, translation assistance may be necessary, as the REB Chair communicates in English only.

If you have any questions about the research, please contact me at celine.bl@queensu.ca. You may also contact my supervisor Dr. Saad Chahine at saad.chahine@queensu.ca

This Letter of Information provides you with the details to help you make an informed choice. All your questions should be answered to your satisfaction before you decide whether or not to participate in this research study. Keep one copy of the Letter of Information for your records and return one copy to the Researcher, Celine Bruce-Lepage.

You have not waived any legal rights by consenting to participate in this study.

By verbally consenting, I am verifying that: I have read the Letter of Information and all of my questions have been answered.

☐ Yes, you have my permission to use quotes
☐ Yes, you have my permission to record my interview.
☐ No, you do not have my permission to use quotes.
☐ No, you do not have my permission to record my interview.

_____	_____	_____
Signature of Person Conducting the Consent Discussion	PRINTED NAME & ROLE	Date

Appendix C

Interview Guide

The purpose of this study is to improve the training of physicians by gathering patient perspectives about healthcare management and to integrate the rare disease context into existing medical training models (i.e. Patient Centred Care and Evidence Based Medicine). Questions for the interview will be focused on the function of treatment and healthcare planning. There is no level of expertise required to participate in this study. My hope is to gather a wide spectrum of perspectives. If you have any questions or concerns please feel free to contact me directly at celine.bl@queensu.ca

Interview Questions:

1. At what age was your child diagnosed?
Sample probe: How many years have you been managing your child's medical decision-making?

2. How many specialists follow your child currently?
Sample probe: Can you tell me a bit about how they fit into your child's global healthcare management plans?

3. Can you describe what happens when you start working with a new specialist?

4. Do you find that your specialists have a strong understanding of PWS and its particularities?
Sample probe: Can you describe some specific experiences you've had?

5. When discussing next steps, or treatment planning do your physicians "describe" and/or make "clear the pros and cons of different options"?

6. To what extent do you feel you are included in setting healthcare goals for your child?

7. Do decision-making conversations feel collaborative or adversarial?
Sample probe: How do you feel when you engage in these types of conversations?

8. What do you think are the greatest barriers to treatment planning or healthcare management?
Sample probe: If you haven't experienced barriers, can you tell me what is working well?

9. What do you find is the most burdensome, or difficult aspect of managing your child's healthcare needs?
Sample probe: If you don't feel burdened, can you share how you've managed this task successfully?

10. The purpose of this research is to improve the training of physicians when treating patients with a rare disease like PWS. What do you think is missing or needs to be included to improve your experience in managing your child's healthcare needs? Sample probe: What has worked well that you think might help other people?

Appendix D

Recruitment poster