Parent perceptions of health care networks for children with inherited metabolic diseases: a mixed methods study

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Preface

Approvals

Chapter 2 involved primary data collection and secondary analysis of an existing dataset from an established cohort study, for which permission to collect, access and use the data was obtained. The original cohort study (Families' Health Care Experiences for Children with Inherited Metabolic Diseases) obtained ethics approvals from the Children's Hospital of Eastern Ontario Research Ethics Board (CTO 1955) and the University of Ottawa Research Ethics Board (H-04-20-5757), as well as each participating Canadian metabolic centre. Zobaida Al-Baldawi (ZA) was added to the study as a student on the University of Ottawa Research Ethics Board protocol (certificate, Appendix A).

Author Contributions

The student (ZA) was the primary author on the manuscript in this thesis, which was co-authored by her co-supervisors Dr. Beth K Potter (BKP) and Dr. Ian Graham (IG), thesis advisory committee member, Dr. Pranesh Chakraborty (PC), and collaborator, Dr. Ann Jolly (AJ). The student (ZA) was responsible for planning the methodology, collecting primary qualitative data, conducting analyses of quantitative and qualitative data, interpreting the findings, and drafting the manuscript. Each of these processes was done with guidance from BKP, IG, and PC, each of whom also provided critical feedback on the draft manuscript. The additional co-author, AJ, provided methods expertise (social network analysis) that informed the analysis of the data, assisted with the interpretation of the findings, and provided critical feedback on the draft manuscript.

Abstract

Objectives: The aim of this study was to gain a thorough understanding of parents' perceptions of and experiences with the care networks surrounding young children (≤ 12 years) with inherited metabolic diseases (IMDs).

Methods: In this mixed methods study, parent participants created a 'care map' depicting their child's network of care providers. We analyzed care maps using social network analysis. A subset of parents participated in a semi-structured interview. We analyzed interviews thematically and integrated quantitative and qualitative results narratively.

Results: Sixty parents contributed care maps and 10 participated in interviews. Parent-drawn care networks were large with few connections between providers. Parents felt responsible for creating and maintaining care networks and for coordinating care. They valued providers who trusted them as part of their child's health care team.

Conclusions: Our findings highlight the complexity of care for children with IMDs and can inform the design of interventions to improve care.

(**150**/150 words)

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List of Abbreviations

CIMDRN	Canadian Inherited Metabolic Diseases Network
IMDs	Inherited Metabolic Diseases
IQR	Interquartile Range
REDCap	Research Electronic Data Capture
SEPH	School of Epidemiology and Public Health

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Chapter 1 – Introduction

1.1 Background

1.1.1 Rare inherited metabolic diseases

Inherited metabolic diseases (IMDs) (also known as inborn errors of metabolism) are a heterogeneous group of rare genetic diseases that impact health and development.[1] Specifically, IMDs represent a group of over 1000 single-gene disorders, in which a specific gene mutation results in a deficiency or insufficient activity of a gene product – an individual enzyme, a structural protein, or a transporter molecule – in an intermediate metabolic pathway.[2,3] IMDs can generally be classified according to the substrate involved, such as disorders affecting carbohydrates, proteins, lipids, nucleic acids, and other organelles (e.g., lysosomal and mitochondrial disorders).[1,4]

The clinical course for a child with an IMD differs depending on the specific disease as well as on patient and environmental characteristics.[5] For example, for some IMDs, children are at risk of experiencing acute episodes of illness that can be life-threatening. An example IMD that follows this course is the fatty acid oxidation disorder, medium-chain acyl-CoA dehydrogenase (MCAD) deficiency. Children with MCAD deficiency are at high risk during times of catabolic stress, such as during prolonged fasting or when experiencing illness that includes fever; and they may require frequent emergency care to prevent metabolic crises.[6,7] Other IMDs tend to have a more chronic but non-progressive clinical course. An example is the amino acid disorder, phenylketonuria (PKU). Children with PKU require a highly specialized and difficult to manage diet to avoid the development of severe intellectual disabilities and they experience frequent blood draws and interactions with metabolic specialists to monitor their treatment.[8–10] Finally, a third trajectory includes IMDs that are chronic, multi-system, and progressive. An example is

mucopolysaccharidosis type I (MPS I), which is highly variable in severity but in general leads to progressive skeletal, cardiac, digestive, respiratory, and sometimes nervous system manifestations. Children with MPS I tend to require increasing care with multiple pediatric specialists over time.[11]

Most IMDs have an autosomal recessive inheritance pattern. IMDs are individually rare conditions; however, collectively, they represent a group of genetic diseases that are an important cause of morbidity and mortality, often beginning in early childhood.[2] The global incidence of IMDs is estimated to be 1 in 2000 live births and IMDs account for approximately 0.4% of all child deaths globally.[12] The heterogeneous clinical presentation of IMDs, and their overlap with other diseases, can lead to the diagnosis being missed.[12] However, many treatable IMDs for which early treatment is associated with improved outcomes are incorporated into population-wide newborn screening programs.[5]

The early detection of IMDs through newborn screening programs has led to improved outcomes for IMDs where there are effective treatments.[13] Treatment options are tailored to the biochemical and clinical features of the IMD and the unique characteristics of the patient. Traditionally, management of many IMDs relied on diet therapy and supportive care interventions but scientific advances have expanded options in this patient population to include disease-modifying interventions such as enzyme replacement therapy, cell and organ transplantation, and gene therapy.[13] Some current strategies for the management of IMDs involve removal of toxic metabolites, provision of optimum vitamins and cofactors, special dietary management, and supportive therapy in emergencies (e.g. cardiorespiratory support).[14] Successful implementation of these strategies requires continuous monitoring from a variety of health care professionals.

1.1.2 Care needs and experiences of children with IMDs and their families

While the health care needs of children with IMDs are dependent on the clinical course of the disease and the treatment strategies used, many children with IMDs require highly specialized diets and medications to manage at home and have frequent interactions with both hospital- and community-based health services. Previous studies about the health care experiences of children with IMDs and their families, from the perspectives of representatives from patient groups [15] or parents and caregivers themselves,[16, 17] revealed concerns about the lack of familiarity with and knowledge about IMDs among health care providers who were not specialists in metabolic care, and about poor health care coordination.

The challenges specific to this population reflect broader issues with health care delivery for children with other complex and chronic illnesses who similarly require frequent interactions with the health care system.[18,19] One recent study of children with complex medical needs found that on average they received care from 13 different physicians across six specialties.[18] Given the high number of services and providers involved in the care of children with complex or chronic illness, some studies have found that care tends to be fragmented.[20] Specifically, communication between the numerous providers is frequently poor so that family caregivers feel responsible for transmitting information and generally coordinating care among providers, settings, and services.[14,15] Overall, implementation of interdisciplinary, family-centred care attuned to the needs of patients with complex and chronic illness is lacking.[17, 19]

1.1.3 Patient and family-centred IMD care

Patient experience has been recognized as a target for improving the quality of health service delivery.[22] Including patient experience as a measure of quality in health care has multifaceted importance both because of its intrinsic value and its relevance to improving patient

safety and clinical effectiveness.[23] Patient experience and patient-centred care are broad concepts that encompass several dimensions and have been operationalized slightly differently across health care institutions. The Institute of Medicine, Picker Institute, and National Institute for Health and Care Excellence (NICE) have all produced patient experience frameworks which share the following common principles: access, coordination and continuity of care, physical comfort, emotional support, clear communication, and involvement of family and carers in decisions. [22,24,25] In pediatrics, the concept of patient-centred care is expanded to patient and family-centred care. Family-centred care is appropriate in pediatrics since it accounts for children's developmental trajectories and their dependence on parents, emphasizing the central role the family plays in all aspects of pediatric care.[26] There is no consensus definition of family-centred care; however, as a concept, it emphasizes collaboration and partnership between care providers and it views families as experts in the care delivery process.[27] The common elements in family-centred care as conceptualized by several groups are information sharing, respect and honoring differences, partnership and collaboration, negotiation, and care in the context of family and community.[27]

1.1.4 Care coordination and relational continuity

Operationalizing family-centred care in the ambulatory setting, particularly in the United States, has revolved around the Medical Home model, which specifies that care for all children should be accessible, coordinated, comprehensive, family-centred, culturally competent, continuous, and compassionate.[26,27] Specifically, care coordination is highlighted as an essential component to the delivery of high-quality health care for children with complex medical needs.[28–30] Children with complex medical needs are often targeted by care coordination interventions due to their disproportionately high need for health care services.[31] Care coordination can be interpreted and operationalized differently across health care settings. According to the framework

published by Antonelli et al. and reiterated by the American Academy of Pediatrics, care coordination can be defined as:

> *Patient and family-centered, assessment-driven, team-based activity designed to meet the needs of children and youth while enhancing the care giving capabilities of families. Care coordination addresses interrelated medical, social, developmental, behavioral, educational and financial needs to achieve optimal health and wellness outcomes.*[26,28]

Care coordination is an essential component of the Medical Home model as it emphasizes partnerships across settings.[26,28] In one study, care coordination improved communication between health care providers and family satisfaction with care.[32] Another study linked care coordination with receiving family-centred care, satisfaction with services, lower financial family burden, less impact on parental employment, fewer emergency department visits and improved parental partnerships with professionals.[33]

Further, care coordination can contribute to facilitating relational continuity with key providers.[33,34] Haggerty et al. define relational continuity as an ongoing, trusting therapeutic relationship between the patient/family and a health care provider.[35] Caregivers of children with medical complexity report a need for relational continuity.[20,34] In a study exploring care coordination across care networks for children with chronic health conditions, caregivers emphasized that the concept of relational continuity is important across the entire network of services and providers.[20] Caregivers believed that relational continuity was important for optimum care as it generated a 'complex, contextualized appreciation of the child and family' which in turn, increased clinical accuracy.[20] Similarly, other studies have shown that caregivers have confidence in the quality of care by providers who have a thorough knowledge of their child.[34,36–38] Overall, both coordination of care and relational continuity are essential components of the delivery of high-quality, family-centred care.[29,30,39,40]

1.1.5 Supporting family-centred care using care maps

A 'care map' is a graphical representation of the care and services received by a patient as well as interactions between formal and informal service providers.[20] A care map can be drawn on paper or created electronically. In a pediatric setting, a care map consists of the child and family placed in the centre of the map, with the organizations, teams or individuals radiating outward.[41] Care mapping has been defined as a family-driven process that can be useful to highlight the 'big picture' about the family's lived experience and specific details about resources and needs for each family.[41] Care maps have been suggested as a tool to support care coordination efforts as they can visually bring into focus what is needed both medically and socially to support families.[30,42]

For children with medical complexity, care maps have been used in clinical care, for example, alongside care plans. In such cases, a care map can highlight the family experience in ways that a medical plan cannot, and inform actions to address gaps related to patient and social issues.[43] Further, care maps can be used as a tool to create meaningful partnerships between providers and parents.[30,43] Specifically, in some studies, parents of children with medical complexity perceived the integration of care maps into clinical practice as way to have their voices heard and ensure that their preferences and needs were addressed.[30,43] From health care providers' perspectives, care maps have been perceived as a useful tool that can aid in their understanding of the family's journey of care and the family's perspectives; in some studies, they believed that this understanding allowed them to facilitate better patient and family-centred partnerships. [30,44]

In addition to the application of care maps to facilitate family-centred care in clinical settings, a number of studies have employed care maps as a research tool to study topics pertaining to health service delivery. For example, one study used care maps to understand the care networks of asthma patients and the patient role in multidisciplinary care.[45] Another study assessed the

personal social support networks of families with children with medical complexity using care maps.[46] Other examples include using care maps to understand the networks of care surrounding cancer palliative patients[47] and social support in home pediatric palliative care.[48]

1.1.6 Study context: The Canadian Inherited Metabolic Diseases Research Network

The Canadian Inherited Metabolic Diseases Research Network (CIMDRN) is a pan-Canadian, multidisciplinary network that focuses on generating high quality evidence to improve health care and outcomes for children diagnosed with IMDs.[49] The CIMDRN has to date focused mainly on a subset of 31 targeted IMDs (Appendix B).[49] This network is currently undertaking a new multi-phase set of studies that will ultimately lead to the development and experimental evaluation of complex interventions to improve patient and family experiences with care for children with IMDs, including but extending beyond the 31 that had been studied to date.[50] Phase I of this work aims to understand gaps in the delivery of family-centred care for children with IMDs. Later phases will focus on provider perspectives and on developing evidence-informed interventions. Guided by the family-centred care philosophy, this multiphase program of work emphasizes patient and family engagement, whereby patients/families are involved as partners and advisors in every stage of the research, including the eventual co-design of interventions. This is especially important due to patients'/families' expertise in rare disease care,[51] and this approach is aligned with the aforementioned central role of families in family-centred care

One part of phase I of this research program aims to develop an understanding of the care networks of children with IMDs from parents' perspectives. This is the focus of my thesis project.

1.2 Summary of Rationale

The limited literature specific to IMDs reflects a broader knowledge base regarding care for children with chronic diseases, which identifies a need to improve health care experiences for

children and their families. [19,31] Given the need for frequent interactions with the health care system to address medical and social needs, families of children with IMDs, in common with families of children with chronic diseases generally, face fragmented and impersonal care.[16] To inform the development of interventions tailored to the needs of children with IMDs that address issues of care coordination, relational continuity, and other elements of family-centred care, we first need to better understand current health care needs and gaps in care specific to this population.

1.3 Objectives

The overall aim of this study is to gain a thorough understanding of parents'/guardians' (hereafter "parents") perceptions of the care networks of young children with IMDs and how those networks are experienced. The following are the specific study objectives:

1) Quantitative objective: To describe the providers and services included in care networks for children with IMDs from parents' perspectives, the connections they identify between providers, the adequacy of care coordination, and the degree of relational continuity with providers perceived as most important.

2) Qualitative objective: To gain an in-depth understanding of parental perceptions of care networks for children with IMDs, including: i) experiences with the care network as a whole; ii) the identification of characteristics (i.e., actions and attributes) of a 'key provider' and the role they play in the child's IMDs care; and (iii) care coordination among providers including perceived gaps in coordination and their impact; and

3) Mixed methods integration: To merge the quantitative and qualitative findings related to children's care networks to arrive at a nuanced and thorough understanding of the nature of those networks and how they are experienced and assessed by parents.

1.4 Guiding Concepts

In line with previous studies of health care experiences,[52–55] principles of family-centred care, such as information sharing, respect, and involvement of families, were used to guide data collection and analysis.[27] In particular, relational continuity and care coordination for children with IMDs served as sensitizing concepts. We applied these guiding concepts by asking direct questions relating to them in data collections tools (e.g., questionnaires, interview guide). During analysis, we identified themes relating to concepts of family-centred care.

1.5 Category and Format of Thesis

This thesis involves a secondary analysis of existing data (care maps) and primary data collection (qualitative interviews). The thesis has three chapters and is written using a thesis by article(s) format in accordance with guidelines provided by the School of Epidemiology and Public Health (SEPH). Chapter 1 has described the literature on the topic and other key concepts, as well as the rationale and objectives for the thesis. Chapter 2 is a standalone manuscript and describes the completed mixed-methods study that addresses objectives 1-3. We used a quantitative analysis of care maps created by participants in a larger cohort study and semi-structured interviews with a subset of those participants to gain a nuanced understanding of parents' perspectives on the care networks of children with IMDs. Finally, chapter 3 presents an elaborated discussion summarizing and interpreting the study findings and their implications for designing health services delivery interventions and future research.

References

1. Ferreira CR, Karnebeek CDM Van, Vockley J, Blau N, Centre AM. A proposed nosology of inborn errors of metabolism. *Genet Med*. 2019;21(1):102-106. doi:10.1038/s41436-018-0022-8.A
2. Pampols T. Inherited Metabolic Rare Disease. In: *Advances in Experimental Medicine and Biology*. ; 2010:397-431. doi:10.1007/978-90-481-9485-8_23
3. Sanderson S, Green A, Preece MA, Burton H. The incidence of inherited metabolic disorders in the West Midlands, UK. *Arch Dis Child*. 2006;91(11):896-899. doi:10.1136/adc.2005.091637
4. Rajappa M, Goyal A, Kaur J. Inherited metabolic disorders involving the eye: A clinico-biochemical perspective. *Eye*. 2010;24(4):507-518. doi:10.1038/eye.2009.229
5. Ellaway C, Wilcken B, Christodoulou J. Clinical approach to inborn errors of metabolism presenting in the newborn period. *J Paediatr Child Health*. 2002;38(5):511-517. doi:10.1046/j.1440-1754.2002.00047.x
6. Grosse SD, Khoury MJ, Greene CL, Crider KS, Pollitt RJ. The epidemiology of medium chain acyl-CoA dehydrogenase deficiency: An update. *Genet Med*. 2006;8(4):205-212. doi:10.1097/01.gim.0000204472.25153.8d
7. Wilcken B, Haas M, Joy P, et al. Outcome of neonatal screening for medium-chain acyl-CoA dehydrogenase deficiency in Australia: a cohort study. *Lancet*. 2007;369(9555):37-42. doi:10.1016/S0140-6736(07)60029-4
8. Vockley J, Andersson HC, Antshel KM, et al. Phenylalanine hydroxylase deficiency: Diagnosis and management guideline. *Genet Med*. 2014;16(2):188-200. doi:10.1038/gim.2013.157
9. MacDonald A, Van Rijn M, Gokmen-Ozel H, Burgard P. The reality of dietary compliance in the management of phenylketonuria. *J Inherit Metab Dis*. 2010;33(6):665-670. doi:10.1007/s10545-010-9073-y
10. Hartnett C, Salvarinova-Zivkovic R, Yap-Todos E, et al. Long-term outcomes of blood phenylalanine concentrations in children with classical phenylketonuria. *Mol Genet Metab*. 2013;108(4):255-258. doi:10.1016/j.ymgme.2013.01.007
11. D'Aco K, Underhill L, Rangachari L, et al. Diagnosis and treatment trends in mucopolysaccharidosis I: Findings from the MPS I registry. *Eur J Pediatr*. 2012;171(6):911-919. doi:10.1007/s00431-011-1644-x
12. Waters D, Adeloye D, Woolham D, Wastnedge E, Patel S, Rudan I. Global birth prevalence and mortality from inborn errors of metabolism: a systematic analysis of the evidence. *J Glob Health*. 2018;8(2). doi:10.7189/jogh.08.021102
13. Fukao T, Nakamura K. Advances in inborn errors of metabolism. *J Hum Genet*. 2019;64(2):65. doi:10.1038/s10038-018-0535-7
14. Mohamed S. Review Article Treatment strategies for acute metabolic dis- orders in neonates. *Sudan J Pediatr*. 11(2):6-13.
15. Khangura SD, Tingley K, Chakraborty P, et al. Child and family experiences with inborn errors of metabolism: a qualitative interview study with representatives of patient groups. *J Inherit Metab Dis*. 2016;39(1):139-147. doi:10.1007/s10545-015-9881-1
16. Siddiq S, Wilson BJ, Graham ID, et al. Experiences of caregivers of children with inherited metabolic diseases: a qualitative study. *Orphanet J Rare Dis*. 2016;11(1):168. doi:10.1186/s13023-016-0548-2

17. Chow AJ, Pugliese M, Tessier LA, et al. Family Experiences with Care for Children with Inherited Metabolic Diseases in Canada: A Cross-Sectional Survey. *Patient*. 2022;15(2):171-185. doi:10.1007/s40271-021-00538-8
18. Cohen E, Berry JG, Camacho X, Anderson G, Wodchis W, Guttmann A. Patterns and Costs of Health Care Use of Children With Medical Complexity. *Pediatrics*. 2012;130(6):e1463-e1470. doi:10.1542/peds.2012-0175
19. Dewan T, Cohen E. Children with medical complexity in Canada. *Paediatr Child Health*. 2013;18(10):518-522. doi:10.1093/pch/18.10.518
20. Miller AR, Condin CJ, McKellin WH, Shaw N, Klassen AF, Sheps S. Continuity of care for children with complex chronic health conditions: parents' perspectives. *BMC Health Serv Res*. 2009;9(1):242. doi:10.1186/1472-6963-9-242
21. Kuo DZ. A National Profile of Caregiver Challenges Among More Medically Complex Children With Special Health Care Needs. *Arch Pediatr Adolesc Med*. 2011;165(11):1020. doi:10.1001/archpediatrics.2011.172
22. Institute of Medicine. *Crossing the Quality Chasm*. National Academies Press; 2001. doi:10.17226/10027
23. Doyle C, Lennox L, Bell D. A systematic review of evidence on the links between patient experience and clinical safety and effectiveness. *BMJ Open*. 2013;3(1):e001570. doi:10.1136/bmjopen-2012-001570
24. Picker Institute. Principles of Patient-Centered Care. Accessed January 10, 2018. http://pickerinstitute.ipfcc.org/about/picker-principles/
25. February EU. Patient experience in adult NHS services Evidence Update February 2014. 2014;35(February):839-849.
26. Turchi RM, Antonelli RC, Norwood KW, et al. Patient- and Family-Centered Care Coordination: A Framework for Integrating Care for Children and Youth Across Multiple Systems. *Pediatrics*. 2014;133(5):e1451-e1460. doi:10.1542/peds.2014-0318
27. Kuo DZ, Houtrow AJ, Arango P, Kuhlthau KA, Simmons JM, Neff JM. Family-Centered Care: Current Applications and Future Directions in Pediatric Health Care. *Matern Child Health J*. 2012;16(2):297-305. doi:10.1007/s10995-011-0751-7
28. Antonelli, R; McAllister, J; Popp J. Making Care Coordination a Critical Part of the Pediatric Health System. 2009;(1277). http://www.commonwealthfund.org/publications/fund-reports/2009/may/making-care-coordination-a-critical-component-of-the-pediatric-health-system
29. Turchi RM, Antonelli RC, Norwood KW, et al. Patient- and Family-Centered Care Coordination: A Framework for Integrating Care for Children and Youth Across Multiple Systems. *Pediatrics*. 2014;133(5):e1451-e1460. doi:10.1542/peds.2014-0318
30. Adams S, Nicholas D, Mahant S, et al. Care maps for children with medical complexity. *Dev Med Child Neurol*. 2017;59(12):1299-1306. doi:10.1111/dmcn.13576
31. Cohen E, Kuo DZ, Agrawal R, et al. Children With Medical Complexity: An Emerging Population for Clinical and Research Initiatives. *Pediatrics*. 2011;127(3):529-538. doi:10.1542/peds.2010-0910
32. Wood D, Winterbauer N, Sloyer P, et al. A Longitudinal Study of a Pediatric Practice-Based Versus an Agency-Based Model of Care Coordination for Children and Youth with Special Health Care Needs. *Matern Child Health J*. 2009;13(5):667-676. doi:10.1007/s10995-008-0406-5
33. Turchi RM, Berhane Z, Bethell C, Pomponio A, Antonelli R, Minkovitz CS. Care

Coordination for CSHCN: Associations With Family-Provider Relations and Family/Child Outcomes. *Pediatrics*. 2009;124(Supplement 4):S428-S434. doi:10.1542/peds.2009-1255O
34. Baird J, Rehm RS, Hinds PS, Baggott C, Davies B. Do You Know My Child? Continuity of Nursing Care in the Pediatric Intensive Care Unit. *Nurs Res*. 2016;65(2):142-150. doi:10.1097/NNR.0000000000000135
35. Haggerty JL. Continuity of care: a multidisciplinary review. *BMJ*. 2003;327(7425):1219-1221. doi:10.1136/bmj.327.7425.1219
36. Carter B, Cummings J, Cooper L. An exploration of best practice in multi-agency working and the experiences of families of children with complex health needs. What works well and what needs to be done to improve practice for the future? *J Clin Nurs*. 2007;16(3):527-539. doi:10.1111/j.1365-2702.2006.01554.x
37. Espezel HJE, Canam CJ. Parent-nurse interactions: care of hospitalized children. *J Adv Nurs*. 2003;44(1):34-41. doi:10.1046/j.1365-2648.2003.02765.x
38. Fisher MJ, Broome ME. Parent–Provider Communication During Hospitalization. *J Pediatr Nurs*. 2011;26(1):58-69. doi:10.1016/j.pedn.2009.12.071
39. Berwick DM, Nolan TW, Whittington J. The Triple Aim : Care , Health , And Cost Reproduced with permission of the copyright owner . Further reproduction prohibited without permission . *Health Aff*. 2008;27(3):759-769.
40. Stille C, Turchi RM, Antonelli R, et al. The Family-Centered Medical Home: Specific Considerations for Child Health Research and Policy. *Acad Pediatr*. 2010;10(4):211-217. doi:10.1016/j.acap.2010.05.002
41. Antonelli RC, Lind C. Care Mapping : A How-To Guide for Patients and Families How do I make my care map for my child or family member ? How do I use my care map ? :2-4.
42. Adams S, Cohen E, Mahant S, Friedman JN, MacCulloch R, Nicholas DB. Exploring the usefulness of comprehensive care plans for children with medical complexity (CMC): a qualitative study. *BMC Pediatr*. 2013;13(1):10. doi:10.1186/1471-2431-13-10
43. Adams S, Nicholas D, Mahant S, et al. Care maps and care plans for children with medical complexity. *Child Care Health Dev*. 2019;45(1):104-110. doi:10.1111/cch.12632
44. Batalden M, Batalden P, Margolis P, et al. Coproduction of healthcare service. *BMJ Qual Saf*. 2016;25(7):509-517. doi:10.1136/bmjqs-2015-004315
45. Cheong LHM, Armour CL, Bosnic-Anticevich SZ. Primary health care teams and the patient perspective: A social network analysis. *Res Soc Adm Pharm*. 2013;9(6):741-757. doi:10.1016/j.sapharm.2012.12.003
46. Varda DM, Talmi A. Social Connectedness in Family Social Support Networks: Strengthening Systems of Care for Children with Special Health Care Needs. *eGEMs (Generating Evid Methods to Improv patient outcomes)*. 2018;6(1):23. doi:10.5334/egems.232
47. Jarrett N, Porter K, Davis C, et al. The networks of care surrounding cancer palliative care patients. *BMJ Support Palliat Care*. 2015;5(4):435-442. doi:10.1136/bmjspcare-2014-000782
48. Lindemann D, Borasio GD, Führer M, Wasner M. Visualizing social support in home pediatric palliative care using network maps. *Palliat Med*. 2020;34(3):378-386. doi:10.1177/0269216319870673
49. Potter BK, Chakraborty P, Kronick JB, et al. Achieving the “triple aim” for inborn errors

of metabolism: a review of challenges to outcomes research and presentation of a new practice-based evidence framework. *Genet Med*. 2013;15(6):415-422. doi:10.1038/gim.2012.153

50. Chow AJ, Iverson R, Lamoureux M, et al. Families' healthcare experiences for children with inherited metabolic diseases: protocol for a mixed methods cohort study. *BMJ Open*. 2022;12(2):e055664. doi:10.1136/bmjopen-2021-055664
51. Potter BK, Khangura SD, Tingley K, Chakraborty P, Little J. Translating rare-disease therapies into improved care for patients and families: what are the right outcomes, designs, and engagement approaches in health-systems research? *Genet Med*. 2016;18(2):117-123. doi:10.1038/gim.2015.42
52. Luthy C, Gerstel PF, Pugliesi A, Piguet V, Allaz AF, Cedraschi C. Bedside or not bedside: Evaluation of patient satisfaction in intensive medical rehabilitation wards. *PLoS One*. 2017;12(2):1-11. doi:10.1371/journal.pone.0170474
53. Byczkowski TL, Gillespie GL, Kennebeck SS, Fitzgerald MR, Downing KA, Alessandrini EA. Family-Centered Pediatric Emergency Care: A Framework for Measuring What Parents Want and Value. *Acad Pediatr*. 2016;16(4):327-335. doi:10.1016/j.acap.2015.08.011
54. Wong ELY, Leung MCM, Cheung AWL, Yam CHK, Yeoh EK, Griffiths S. A population-based survey using PPE-15: Relationship of care aspects to patient satisfaction in Hong Kong. *Int J Qual Heal Care*. 2011;23(4):390-396. doi:10.1093/intqhc/mzr037
55. Peytremann-Bridevaux I, Kolly V, Perneger T V. The association between body mass index and patients experiences with inpatient care. *Int J Qual Heal Care*. 2010;22(2):140-144. doi:10.1093/intqhc/mzq005

Interface

The following article, titled "*Parents' perceptions of health care networks of children with inherited metabolic diseases (IMD): a mixed methods study*," presents research that included a secondary analysis of experiential data (care maps and care map questionnaire) and primary data collection (semi-structured interviews). For the purposes of my thesis, I completed a secondary analysis of the care maps. However, I have been working as a research assistant on the cohort study prior to the start of data collection for that study, and I have been partially responsible for supporting participant enrollment and data collection. As a research assistant, I co-developed and implemented procedures for digitizing care maps and communicating with families about their care maps. As part of my thesis project, I developed and implemented processes for entering and analyzing the care map data.

The secondary analysis of care maps and questionnaire addressed objective 1 of this thesis, investigating the structure and organization of care networks of children with IMDs and satisfaction with care coordination and relational continuity, from parents' perspectives. Specifically, we: i) investigated the frequency with which each provider type was included in parent-drawn care maps for children with IMDs, the frequency of identification of each provider type as a key health care provider, and, for each of the most common key health care providers, parental perceptions of relational continuity and care coordination; and ii) used social network analysis to understand network size and connections between providers, from parents' perspectives. The semi-structured interviews about the care maps addressed objective 2 of this thesis. We explored parental perceptions of care networks for children with IMDs in greater depth, including how networks were experienced overall as well as relationships with providers and connections between providers. We used mixed-methods integration to address objective 3 of this

thesis in order to gain a nuanced and thorough understanding of the nature of children's care networks and how they are experienced and assessed by parents. In chapter 2, I briefly describe patient partner involvement in all stages of this work as co-investigators and advisors who have contributed since the beginning. Before publication of the study, patient partner investigators involved in this study will have the opportunity to provide feedback on the manuscript. At that stage, we will report on the full patient engagement strategy using the Guidance for Reporting Involvement of Patients and the Public 2.0 (GRIPP2).[1]

The cohort study in which this thesis was embedded received ethics approval from the Children's Hospital of Eastern Ontario Research Ethics Board, the University of Ottawa Research Ethics Board, and research ethics boards at each participating metabolic centre. This article is formatted for submission to the journal *Health Expectations*. The article presented in this thesis contains more details in the Methods and Results sections than what would normally be suitable for a journal publication. This is to ensure that thesis examiners have sufficient details regarding the study to thoroughly assess the work. The version submitted for publication will follow the word limits imposed by *Health Expectations*.

Author contributions: ZA, BKP, PC, IDG, and AJ conceived of the study idea and planned the methodology. ZA conducted data analyses and drafted the manuscript. All authors assisted with the interpretation of data and results and provided feedback and suggested critical revisions to the final manuscript. Additional co-investigators, including patient partners, as well as research coordinators who have already contributed to the study in which this thesis is embedded will also eventually meet all co-authorship criteria by critically reviewing the manuscript after the thesis defence. For the purposes of the thesis evaluation these additional authors have been excluded.

Chapter 2 – Parents' perceptions of health care networks of children with inherited metabolic diseases (IMDs): a mixed methods study

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Abstract

Introduction: Children with inherited metabolic diseases (IMDs) and their families face challenges in receiving high quality, family-centred health services. To inform the development of interventions to improve their health care experiences, we need to better understand care needs from families' perspectives. We sought to understand parental perceptions of the health care networks of children with IMDs.

Methods: This explanatory sequential mixed-methods study was embedded in a prospective cohort study of parents/guardians of children ≤12 years of age with IMDs, enrolled from one of 11 participating Canadian centres. Participants drew a 'care map' depicting their perceptions of their child's network of care providers and coordination between providers, followed by a tailored survey about relational continuity and care coordination. We used social network analysis to describe network size, density (a measure of connectedness between providers) and centralization (extent to which connections centred on one specific provider) as well as connectedness of individual providers. A subset of participants participated in a semi-structured interview to elaborate on their care map. We analyzed interviews thematically and integrated quantitative and qualitative results narratively.

Results: Sixty parents provided care maps and 10 participated in interviews. Parents identified a median of 14.5 providers in their children's care networks; metabolic doctors and lab technicians were most commonly included (92% of care maps each). Networks had a median density of 0.08, meaning that approximately 8% of possible pairwise connections between providers were identified by parents. Networks had a median centralization of 0.23, meaning that the most connected provider was included in approximately 23% of pairwise connections. A majority of participants (80%) identified at least one key provider who knew the family very or fairly well; parents we interviewed strongly valued this relational continuity. Parents generally perceived that their children's care needs were being met but described that this required extensive effort by families to establish and coordinate care. Parents often sought a "go-to" provider in the network who could share information and support them in accessing care.

Conclusion: These findings highlight gaps in family-centred care for children with IMDs that place a high responsibility on parents as informal care "managers".

(350/350 words)

Patient or Public Contribution:
Three patient partners are co-investigators on the cohort study in which this study is embedded and led the patient engagement strategy. They were involved in decision making at all stages of this care map component of the study, including development of the data collection instruments, development of the recruitment strategy, and the interpretation of the findings. Eleven family and patient advisors, recruited from family advocacy and support organizations, provided feedback on the Care Map instructions and all study questionnaires.

Keywords (maximum 7): inherited metabolic diseases; mixed methods; social network analysis; care coordination; relational continuity; caregivers; family-centred care

2.1 Introduction

Inherited metabolic diseases (IMDs) are a heterogeneous group of over 1000 single gene disorders that lead to deficiencies in enzymes needed in biochemical pathways.[2] IMDs are individually rare conditions; however, collectively, they are an important cause of morbidity and mortality, with clinical manifestations that frequently begin in early childhood.[3] For some IMDs there are disease-modifying treatments that yield improved outcomes when implemented in early life and thus IMDs are the most common targets of population-wide newborn screening programs.[4] The clinical course for a child with an IMD depends on the specific disease, and individual child's characteristics and can encompass acute episodic illnesses with or without accompanying chronic multi-system sequelae or a chronic clinical course with or without progressive multisystem disease.[4]

While the health care needs of each child are dependent on the specific disease, child age, and other characteristics, there are some commonalities in the health care needs and experiences of children with IMDs and their families. For example, children with IMDs often require highly specialized diets and medications and have frequent interactions with hospital- and community-based health services. Some of the reported challenges faced by families include uncertainty with the diagnosis and prognosis, poor care coordination and discontinuity of information, lack of familiarity with IMD care among some providers, and substantial caregiver impacts with respect to the time needed to provide care at home and organize care within the health care system.[5,6] These challenges are echoed by caregivers of children with medical complexity who often report a need for care coordination and relational continuity across the entire network of services and providers.[7,8] A few studies have shown that caregivers have confidence in the quality of care by providers who have a thorough knowledge of their child.[7,9–11] Overall, both coordination of care

and relational continuity are essential components of the delivery of high-quality, family-centred care.[12–15]

The limited literature specific to IMDs reflects a broader knowledge base regarding care for children with other chronic diseases which identifies a need to improve health care experiences for children and their families. To inform the development of interventions tailored to the needs of children with IMDs that address issues of care coordination and relational continuity, we first need to better understand current health care needs and gaps in care specific to this population.

The overall aim of this mixed-methods study is to gain a thorough understanding of parents' perceptions of the structure of care networks for young children with IMDs and how those networks are experienced. The specific study objectives are:

1) Quantitative objective: To describe the providers and services included in care networks for children with IMDs from parents' perspectives, the connections they identify between providers, the adequacy of care coordination, and the degree of relational continuity with providers perceived as most important;
2) Qualitative objective: To gain an in-depth understanding of parental perceptions of care networks for children with IMDs, including: i) experiences with the care network as a whole; ii) the identification of characteristics (i.e., actions and attributes) of a 'key provider' and the role they play in the child's IMDs care; and (iii) care coordination among providers including perceived gaps in coordination and their impact; and
3) Mixed methods integration: To merge the quantitative and qualitative findings related to children's care networks to arrive at a nuanced and thorough understanding of the nature of those networks and how they are experienced and assessed by parents.

2.2 Methods

This study received ethics approval from the Children's Hospital of Eastern Ontario Research Ethics Approval (REB), the University of Ottawa Research Ethics Board, and the Research Ethics Boards of all participating hospital centres.

2.2.1 Study design

We used an explanatory sequential mixed-methods design (QUANT → QUAL).[16] We first invited participants to co-construct a care map, starting with a participant-provided drawing, and to provide information about care coordination and relational continuity in relation to the providers shown on the care map. We invited a purposively selected subsample of these participants to participate in a qualitative semi-structured interview. The term "sequential" is applied at the level of the individual participant as opposed to the group level, whereby the qualitative interview questions focused and expanded upon the care maps, thus enhancing, and explaining the quantitative data.

In this study, caregivers of children with rare disease and/or individuals with rare diseases were engaged as patient partners and contributed at all stages of the work.

2.2.2 Study sites and participants

This research project is embedded in a prospective cohort study where participants were parents or legal guardians ("parents") of children 12 years of age or younger diagnosed with an IMD (list of targeted IMDs is summarized in Appendix B) and receiving care at one of 11 participating Canadian metabolic treatment centres, located in academic pediatric hospitals in six provinces. To form a heterogeneous sample, we implemented a purposive maximum variation sampling approach[17] for the cohort study[18], to achieve diversity on six pre-identified categorical variables that we hypothesized may be associated with health care needs and/or experiences for

children with IMDs: study centre, travel time from home to study centre (in broad categories), child's sex assigned at birth**,** child's age (years), and typical clinical trajectory of the IMD. With respect to clinical trajectory, we classified each IMD diagnosis into one of three a priori clinician-informed groups that were deemed descriptive of the overall clinical course of IMDs: i) Group 1: chronic and non-progressive; ii) Group 2: acute and episodic; and iii) Group 3: multi-system and progressive. Treatment protocols and health care service availability and practice have been shown to vary by clinical needs, study centre, and/or distance to specialists.[19,20] Age has also been demonstrated to impact the frequency of health care encounters, with the most frequent encounters being in the first years following an IMD diagnosis.[5] Finally, sex differences in metabolic traits are widely reported.[21,22] Participants were enrolled on a rolling basis between November 2020 and March 2022.

2.2.3 Quantitative methods

2.2.3.1 Quantitative sample size

Given the descriptive purpose of the cohort study, there was no formal sample size calculation. The aim was to recruit approximately 100 participants to support descriptive analyses, achieve diversity on the sampling characteristics noted, and for feasibility.

2.2.3.2 Data collection: Care map, care map questionnaire, and baseline questionnaire

A 'care map' is a graphical representation of the care and services received by a patient as well as interactions with formal and informal service providers.[8] We define a care map in this study as a graphical representation of the network of health care providers surrounding a child with an IMD and their family, from the perspective of a parent. Previous research involving children with complex or chronic health conditions has defined and used care maps similarly.[8,14] In the cohort study, enrolled participants received an invitation to create a care map reflecting the parent's

perceptions of their child's care as one of the first data collection activities after enrollment. In the care map instructions (Appendix C), parents were asked to draw a care map that reflected how they saw their child's network of care providers, including which providers were involved in care provision, which providers were perceived to work together to coordinate their child's care, and which providers were considered to be 'key providers' (up to ten key providers could be listed). We defined a key provider as someone perceived by the parent to be important to their child's health care. We provided participants with a template, whereby the centre of the map included the child and family, with connections to categories or subcategories of services and providers involved in the child's care.[23] The participants drew the care map by hand. They then photographed the map and uploaded the photo to a survey software program used for the full study, REDCap[24,25], which was hosted on a secure server at the Children's Hospital of Eastern Ontario. REDCap accommodated secure communication back and forth between participants and the study team. The study team created an electronic version of the care map that sought to maintain the structure submitted by the family while also harmonizing some elements, for example, using standard terms for health care providers such as "dietitian" and "school psychologist" (guidelines that the study team used to create an electronic care map are summarized in Appendix D). The research team submitted the care map back to the participant to confirm the accuracy of the electronic version. Sometimes telephone calls or secure email communication were used in further interactions until we arrived at an electronic version of the care map that the participant felt accurately reflected their perceptions of the child's care network while retaining the elements needed for analysis. Once the electronic care map was finalized, the study team (ZA and a research staff member) double-entered the care map data into a secured data entry form on REDCap (guidelines for care map data entry are summarized in Appendix E).

Once a final version of the care map was submitted, participants were invited to complete a care map questionnaire (Appendix F). On the care map questionnaire, participants were asked two research team-developed questions for each of the health care providers on the care map who were designated as "key providers" by the participant (up to 10 key providers in total). Specifically, for each of the key health care providers, participants were asked to rate their perceptions of that provider's coordination with other providers ("How well do you think your child's key health care providers coordinate your child's care with other providers?") and their perceptions of relational continuity of that provider with the child and family ("How well does each of your child's key health care providers know your child?"). Both questions used a 5-point Likert-type scale (0, "Not well at all" to 4, "Very well").

Once the care map questionnaire was submitted, participants were invited to complete a baseline questionnaire. In this study we reported the characteristics of the parent, the child and the household from the data used for purposive sampling and from the baseline questionnaire.

2.2.3.3 Quantitative Analysis

We reported the characteristics of participants and their children descriptively. For child characteristics, we reported age, sex, IMD diagnosis, presence of other chronic conditions, and metabolic centre providing care; for parent characteristics, we reported the relationship to the child, gender identity, education, employment status, and immigration status; and for household characteristics, we reported income and travel time from the household to the metabolic centre. We planned to treat missing data with casewise deletion.

We analyzed the care map data using an adapted form of social network analysis. Briefly, social network analysis is a quantitative methodology that focuses on relationships between and among social entities, measuring and mapping relationships and flows between people, groups, or

organizations.[26,18] The adapted form of social network analysis we used is known as egocentric social network analysis. An egocentric map is composed of only a focal actor (ego) and a set of component actors and their direct ties to the ego as well connections between the component actors.[26,27] We deemed this to be the most appropriate form of social network analysis given that participants' care maps were centred around a child with an IMD (the ego) and perceived connections to providers (the component actors); and given that we were analyzing each child's care map separately rather than making connections between the families in the study. In an egocentric social network analysis, the component actors or individuals who are connected to the ego (providers of care in this case) are called alters. The data analysis was conducted using UCINET software.[28]

Analysis of network characteristics. We reported social network analysis metrics appropriate for egocentric networks. Specifically, we calculated network size, network density, and network centralization for each child's care network.[26, 19] *Network size* refers to the number of alters that are present in a network (i.e., the number of care providers). *Network density* represents the actual number of ties in a network (the term "tie" is used to reflect a connection between two alters) as a ratio to the total maximum ties that are possible with all the alters of the network. In a network with A alters and A_t ties, density is calculated as: $\frac{2\,A_t}{A(A-1)}$. Network density ranges from 0-1 and is interpreted as the proportion of all possible pairwise connections between providers that were perceived as connections by parents. *Network centralization* is measured by summing the difference between the degree centrality value (degree centrality is the number of connections a provider has to other providers, as defined below) of the most central alter ($c(a^*)$) and all other alters ($c(a)$) then dividing by the maximum possible such sum for a network with N alters: $\frac{\sum_{i=1}^{N}(C(a*)-C(a))}{(N-1)(N-2)}$. Network centralization ranges from 0-1 and is interpreted as the proportion

of all parent-identified pairwise connections in a network that included the single provider with the most connections. Network density and centralization are complementary measures; network density allows us to understand the overall cohesiveness of a network while centralization helps to explain the extent to which the connectedness of a network is organized around a particular alter (in this case, a care provider).[26] We reported the median and interquartile range (IQR) for network size, density, and centralization for the sample. We investigated the association of child age, child IMD trajectory group, and travel time to the metabolic centre with each of these network characteristics by reporting the median and IQR for each category of each variable and by using the Kruskal-Wallis test. Finally, as a post-hoc exploratory analysis, we used the distribution of networks by size, density, and centralization to create "archetypes" or groups of networks with similar features. We described these groups by child age, IMD trajectory group, and travel time to the metabolic centre.

Analysis of provider characteristics and connections between specific providers. We descriptively reported on the frequency with which each provider type was included in the care maps and the frequency of identification of each provider type as a key health care provider. We also analyzed the care map questionnaire data to determine, for each of the most common key health care providers, parental perceptions of relational continuity and care coordination. Finally, we reported the social network analysis metrics of degree-centrality and share for each provider type in the care networks.[26] *Degree-centrality* refers to the number of ties that a particular alter has with other alters in the network (i.e., the number of other providers to whom each provider is perceived to be connected). The degree centrality of an alter, d(i), is mathematically calculated as: $d(i) = \Sigma m_{ij}$, where $m_{ij} = 1$ if there is a link between alters i and j, and $m_{ij} = 0$ if there is no such link. We calculated degree centrality for each provider type across the sample and described the

median and IQR for degree centrality across the sample for each provider type included on at least five participants' children's care maps. *Share* is the degree centrality measure of an alter divided by the sum of all the alter centralities in the network. Thus, shares of all alters in a network must sum to 1 and can be interpreted as the proportion of perceived connections that include a specific provider type. We calculated the share for each provider type across the sample and described the median and IQR of the share for each provider type included on at least five participants' children's care maps.

2.2.4 Qualitative methods

2.2.4.1 Qualitative sample

A sub-sample of participants from those who completed the care map and care map questionnaire were invited to participate in the care-network focused interviews. To form a heterogenous sample, we identified and recruited participants to achieve diversity on child's age, travel time to the metabolic centre, IMD clinical trajectory, and metabolic centre. Participants were sampled after completing the care map questionnaire and while still contributing data to the cohort study. On a rolling basis, participants were invited via e-mail to participate in a one-on-one, semi-structured interview.

2.2.4.2 Qualitative sample size

A common guiding principle informing the termination of data collection in qualitative research is the concept of saturation. Our view of saturation is centred around the idea of 'informational redundancy.'[29,30] We followed Hennink, Kaiser & Marconi's conceptualization of saturation as 'the point when we fully understood issues, and when no further dimensions, nuances, or insights of issues can be found.'[31(p.594)] We operationalized thematic saturation by looking at

the new content codes identified in the analysis of each interview and stopped interviews when we found no new content codes.

2.2.4.3 *Data collection: Qualitative interviews*

The purpose of the qualitative interviews was to enhance our understanding of parents' perceptions of and experiences with the care networks of children with IMDs. The study team developed the interview guide concurrently with the early care map data collection in the quantitative study (Appendix G). The sample of care maps submitted by enrolled participants as well as literature on care maps influenced the questions explored in the interview guide.[32,33] Feedback on the interview guide from the study patient partners about wording of questions was incorporated into the final interview guide. The interview guide queried the following topics: (i) how parents experienced the process of drawing the care map, (ii) how they selected providers to include on the care map, (iii) characteristics of a 'key' health care provider and the role they played in the child's IMD care, (iv) the nature of the connections between providers and the impact of those connections on the family, and (v) the adequacy of the care network in meeting the child's and family's needs, including parental perceptions of the need for improvements in the care network. The interview guide was tested with practice interviews involving three study patient partners to ensure that the questions were clear and logical and to receive feedback on the overall experience of being interviewed. ZA conducted the interviews using the Zoom platform by videoconference or by audioconference, depending on participant preference. The interviews were recorded using the Zoom platform and fully transcribed by a transcriptionist affiliated with the Children's Hospital of Eastern Ontario. ZA verified all transcripts and removed any identifying information (e.g., names and specific diagnoses prior to analysis).

2.2.4.4 Qualitative analysis

We analyzed the qualitative data concurrently with data collection so that later interviews could probe themes we identified in earlier interviews. The analysis of the qualitative interview data was guided by an interpretive description approach,[34] suitable for investigation of a clinical phenomenon with the goal of capturing themes and patterns within subjective perceptions and generating a description capable of informing clinical understanding.[34] Interpretive description is in philosophical alignment with interpretive naturalistic orientations. Thus, the following are key philosophical underpinnings for interpretive description study design: i) reality is complex, contextual, constructed and subjective; ii) the researcher and the participant interact to influence each other; and iii) no *a priori* theory could possibly encompass the multiple realities that are likely to be encountered.[34]

To guide the analysis and coding of the data, we implemented the six phases of thematic analysis proposed by Braun and Clark.[35] First, after each interview, ZA, BKP, and IG reviewed the transcripts to familiarize themselves with the study data by reading and re-reading the interview transcript, writing memos and generating initial codes. The study team met periodically during data collection and analysis to discuss the early findings and ensure that future interviews were used to probe and check on the initial codes identified. Second, ZA systematically coded across the entire dataset of interview transcripts. BKP reviewed the coding. Third, the codes were collated into potential themes and all data relevant to the potential themes were gathered. Fourth, ZA reviewed the generated themes against the original data and the initial codes. Fifth, ZA, BKP and IG refined the themes into a cohesive set with clear definitions. Finally, we selected data extracts to illustrate the themes. The data analysis was an iterative and reflective process and the team cycled through the different phases back and forth to ensure that findings were reflective of the

original data. We presented interim results of the qualitative analysis to three patient partners, a clinician (PC), and other members of our research team, in order to broaden our perspective on their interpretation.

To ensure qualitative rigor, we implemented several processes during different phases of the study that follow the criteria of credibility, transferability, dependability and confirmability.[36] To summarize, we employed verbatim transcription as a strategy to ensure rigor; we engaged with the study data for a prolonged period of time and reviewed the data and analysis with experts in qualitative research; we created thick descriptions of the themes; and we maintained an audit trail by keeping records of the transcripts and notes.

2.2.4.5 Positionality and reflexivity

ZA is a graduate student with previous experience working on qualitative- and quantitative-focused research teams. ZA has a rare disease, diagnosed in childhood. ZA's health care experiences differed markedly from children in this study, on grounds of both less intense and frequent interactions with the health care system. Nonetheless, her experiences provided her with a unique perspective when speaking to parents of children with rare diseases and may have influenced and aided in relating to the parents and understanding their perspectives. Thus, it was particularly important to operationalize reflexivity throughout the research process. To do so, ZA used memoing and reflection. Immediately following each interview, ZA wrote memos about any part of the interview that invoked opinions or thoughts based on her personal and professional experiences. ZA and BKP then discussed these interviews and reflected on how those opinions or thoughts might have influenced the interview and analysis process. BKP is an epidemiologist and health services researcher with formal training in quantitative and mixed methods. She leads a program of research that aims to generate evidence toward improving health care for children with

rare diseases. BKP is a parent who has experience with receiving care for her two children but not in the context of chronic disease**.** IDG is medical sociologist and health services researcher with formal training in qualitative and quantitative methods. He leads a program of research that focuses on knowledge translation and conducting applied research on strategies to increase implementation of evidence-informed practice. PC is a metabolic physician and leads a program of research that aims to generate evidence toward improving health care for children with rare diseases.

2.2.5 Data integration

We conducted mixed-methods integration at three levels: the design, methods, and interpretation and reporting (Figure 1). As described, at the design level, this study followed the explanatory sequential mixed-methods design, beginning with the quantitative data collection and continuing with qualitative data collection for a subset of participants. At the methods level, the components were integrated through the *connection* approach, whereby one type of data (care maps) links with the other through the sampling frame (care map interviews); i.e., the interview participants were selected from the participants who submitted a care map.[16] Finally, at the interpretation and reporting level, the data and findings were integrated *narratively*, whereby qualitative and quantitative findings were described in a single report. Specifically, a *narrative weaving* approach was followed, in which both quantitative and qualitative findings were written together on a topic-by-topic basis.[16]

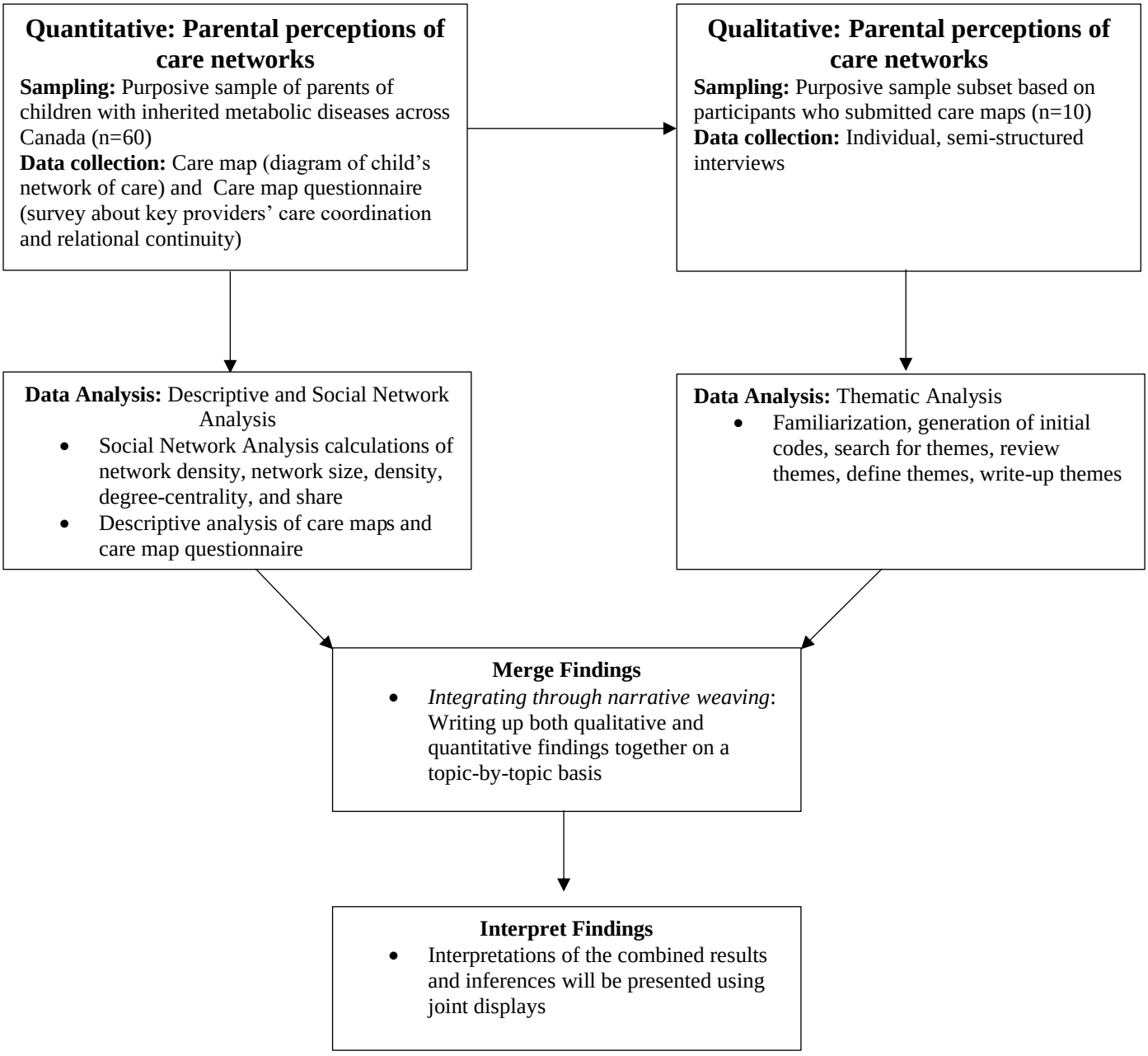

Figure 1. Mixed Methods Integration: An Explanatory Sequential Mixed-Methods Model

2.3 Results

2.3.1 Participant, child, and household characteristics

We analyzed care maps from 60 participants enrolled in the cohort study from November 2020 to March, 2022. There were no missing data on the variables we analyzed, with the exception of household income, for which four participants (7%) indicated that they preferred not to answer. All participants completing care maps and questionnaires were parents or stepparents of children with IMDs and most (88%) were mothers (Table 1). The majority of parents (78%) were college or university graduates and slightly under two thirds (65%) reported working part-time or full-time. More than half of participants (58%) reported a traveling time of one hour or less to reach the metabolic disease clinic. Half of the participants (50%) reported a total annual household income greater than $100,000. The most common IMD diagnoses were medium-chain acyl-CoA dehydrogenase deficiency (22%) and phenylalanine hydroxylase deficiency (18%) and close to half of the children of participants (47%) had a diagnosis that fell into Group 2 (episodic and acute disease). Over one fifth (22%) of parents reported that their child had another chronic disease beyond the IMD diagnosis. The number of participants enrolled across participating clinical sites ranged from 1 to 11 (Table 1).

We invited 15 participants enrolled in the cohort study on an ongoing basis, between June 2021 and February 2022, to participate in a semi-structured interview about their child's care map. In total, 10 participants consented to participate in the care map interviews. We determined that saturation had been reached by the eighth interview. The last two interviews added no new content codes. Hence, the decision was to stop at 10 interviews.

Nine of the 10 interview participants were mothers of children with IMDs (Table 2). Most parents we interviewed were college or university graduates (80%) and reported working part time

or full time (90%). The average interview length was 45 minutes. Characteristics that we used to achieve diversity sampling (travel time to a metabolic centre, child age, child sex assigned at birth, child IMD group, and treating centre) are summarized in Table 2.

We present the remaining results within three topic areas: parent perceptions of the characteristics of children's care networks (2.3.2), key care providers and relational continuity (2.3.3), and providers as part of a dynamic network, including care coordination (2.3.4). For each topic, we present the detailed quantitative and qualitative findings, followed by a joint display table that integrates the findings. A final section of the results (2.3.5) describes parental recommendations for improvements to children's health care networks drawn from the qualitative interviews.

Table 1: Participant, child, and household characteristics (overall cohort study)

	No. (%)*
Parent characteristics	
Relationship to child (n=60)	
Father (biological, legal, or adoptive)	6 (10)
Mother (biological, legal, or adoptive)	53 (88)
Stepmother	1 (2)
Highest education level completed (n=60)	
High school or GED	3 (5)
Some college or university education (no degree or diploma)	10 (17)
College	11 (18)
University	29 (48)
Graduate school	7 (12)
Working part/full time (n=60)	39 (65)
Landed immigrant, yes (n=60)	7 (12)
Household characteristics	
Household income (n=60)	
≤ $40,000	7 (13)
$40,001 - $60,000	6 (11)
$60,001 - $80,000	4 (7)
$80,001 - $100,000	11 (20)
$100,001 – $120,000	22 (39)
≥ 120, 001	6 (11)
Prefer not to answer	4
Travel time to the metabolic centre (n=60)	
1 hour or less	35 (58)
More than 1 hour and up to 3	12 (20)
More than 3 hours	13 (22)
Child characteristics	
Age (years) (n=60)	
≤1 – 3	26 (43)
4 – 6	16 (27)
7 – 12	18 (30)
Assigned Sex (n=60)	
Male	28 (47)
Female	32 (53)
Inherited metabolic disease diagnosis (n=60)	
Medium-chain acyl-CoA dehydrogenase deficiency	13 (22)
Phenylalanine hydroxylase deficiency	11 (18)
Other amino acid disorders	4 (7)
Other fatty oxidation disorders	6 (10)
Urea cycle disorders	6 (10)
Organic acid disorders	3 (5)
Mucopolysaccharidosis type 1 – 4 (MPS I – MPS IV)	9 (15)
Other	8 (13)
IMD Categories (n=60)	
IMD Group I (chronic and non-progressive disease)	17 (28)
IMD Group II (acute and episodic disease)	28 (47)
IMD Group III (progressive and multi-system disease)	15 (25)
Other chronic illness, yes (n=60)	13 (22)
Metabolic Centre where child received care (n=60)	
Alberta Children's Hospital, Calgary	6 (10)
BC Children's Hospital, Vancouver	4 (7)
Children's Hospital – Health Sciences Centre Winnipeg, Winnipeg	1 (2)

Children's Hospital – London Health Sciences Centre, London	4 (7)
Children's Hospital of Eastern Ontario (CHEO), Ottawa	9 (15)
Hospital for Sick Children, Toronto	3 (5)
IWK Health Centre, Halifax	11 (18)
Kingston General Hospital, Kingston	5 (8)
McMaster Children's Hospital – Hamilton Health Sciences**,** Hamilton	11 (18)
Montreal Children's Hospital, Montreal	1 (2)
Stollery Children's Hospital, Edmonton	5 (8)

*numbers rounded up to one decimal place

Table 2: Participant, child, and household characteristics (qualitative interviews)

Parent characteristics (n=10)	**No.** (%)
Relationship to child	
Father (biological, legal, or adoptive)	1 (10)
Mother (biological, legal, or adoptive)	9 (90)
Working part/full time	9 (90)
Household characteristics (n=10)	
Travel time to the metabolic centre (hours)	
1 hour or less	4 (40)
More than 1 hour and up to 3 hours	4 (40)
More than 3 hours	2 (20)
Child Characteristics (n=10)	
Age (years)	
≤1 – 3	4 (40)
4 – 6	3 (30)
7 – 12	3 (30)
Assigned Sex	
Male	6 (60)
Female	4 (40)
IMD Categories	
IMD Group I (chronic and non-progressive disease)	3 (30)
IMD Group II (acute and episodic disease)	3 (30)
IMD Group III (progressive and multi-system disease)	4 (40)
Metabolic centre where child received care	
Alberta Children's Hospital, Calgary	2 (20)
Stollery Children's Hospital, Edmonton	2 (20)
Children's Hospital – London Health Sciences Centre	1 (10)
Children's Hospital of Eastern Ontario	1 (10)
McMaster Children's Hospital	2 (20)
Hospital for Sick Children	1 (10)
Kingston General Hospital	1 (10)

2.3.2 Parent perceptions of the characteristics of children's care networks

2.3.2.1 Characteristics of children's care networks: quantitative findings

From the double entry of care map data, there was no more than a 2% discrepancy in entry of data between ZA and the second researcher entering the data for any single care map. All discrepancies were resolved by review and discussion.

Network size. Parent participants identified a minimum of three and a maximum of 43 care providers in their children's care networks (network size distribution, Appendix H). The median network size was 14.5 (interquartile range [IQR] 10-20) (Table 3). Older children had larger network sizes relative to younger children: parents reported a median of 11 providers for children aged 3 years or younger, 14 providers for children aged 4-6 years, and 20 providers for children aged 7-12 years. Parents also reported more providers in the care networks of children with diagnoses in IMD Group 3 (multi-system and progressive diagnoses, median 26) relative to Group 2 (acute and episodic diagnoses, median 14) and Group 1 (chronic and non-progressive diagnoses, median 10) (Table 3).

Network density. Care networks of children with IMDs had a median density of 0.08 (IQR 0.05-0.11), meaning that approximately 8% of all possible pairwise connections between providers were identified as observed connections by parents. Parents reported observing a higher proportion of connections among providers who could be connected in the care networks of children with diagnoses in IMD Groups 1 (median density 0.10) and 2 (median density 0.09) relative to Group 3 (median density 0.05) (Table 3). Network density was not significantly associated with child age nor with travel time to the metabolic centre.

Network centralization. The median centralization for children's care networks, based on parent perceptions of connections among providers was 0.23 (IQR 0.12-0. 31). This means that on

average, the most connected provider was involved in 23% of all pairwise connections in a care network. Network centralization was not significantly associated with child age nor with IMD group (Table 3). However, parents reported more centralized care networks for children who lived more than three hours (median centralization 0.30) relative to children whose travel time was an hour or less (median centralization 0.28) from a metabolic centre or one to three hours (median centralization 0.16).

Table 3. Network size, density, and centralization of networks for children with IMDs based on parent-contributed care maps (n=60)

	Network Size Median (IQR)	Density Median (IQR)	Centralization Median (IQR)
Overall	14.5 (10 - 20)	0.08 (0.05 - 0.11)	0.23 (0.12 - 0.31)
Travel time to the metabolic centre			
1 hour or less	14 (9 - 19)	0.07 (0.03 - 0.11)	0.23 (0.11 - 0.32)
More than 1 hour and up to 3 hours	20 (15 - 29)	0.07 (0.03 - 0.10)	0.16 (0.10 - 0.28)
More than 3 hours	16 (9 - 21)	0.10 (0.06 - 0.11)	0.30 (0.23 - 0.36)
p (sig)	0.08	0.42	0.047*
Age group (years)			
≤1 – 3	11 (8 - 17)	0.11 (0.06 - 0.19)	0.26 (0.14 - 0.32)
4 – 6	14 (10 -19)	0.07 (0.05 - 0.09)	0.24 (0.14 - 0.33)
7 – 12	20 (16 - 27)	0.06 (0.02 - 0.08)	0.15 (0.10 - 0.26)
p (sig)	0.002**	0.15	0.17
IMD group			
IMD Group I (chronic and non-progressive)	10 (8 - 14)	0.10 (0.03 - 0.14)	0.22 (0.11 - 0.30)
IMD Group II (acute and episodic)	14 (10 - 19)	0.09 (0.06 - 0.14)	0.25 (0.15 - 0.31)
IMD Group III (progressive and multi-system)	26 (18 - 36)	0.05 (0.02 - 0.07)	0.18 (0.12 - 0.33)
p (sig)	<0.0001***	0.02*	0.88

IQR indicates interquartile range. Data are presented as median (IQR), and p-values are from Kruskal-Wallis test.

*** *P <0.001*, ** *P <0.01*, * *P <0.05* according to Kruskal-Wallis test.

Provider types in networks. Parents identified 89 unique "types" of providers (where "type" is the provider role or title – e.g., metabolic doctor, teacher, friend, pediatrician) on their children's care networks. Of these, 64 were health care providers (e.g., nurse, doctor) while 25 were educational providers (e.g., educational assistant) or informal supports (e.g., friend). A summary of the most commonly identified providers and key providers is provided in Table 4 (full

list, Appendix I). Almost all participants (n=55, 92%) identified a metabolic doctor and lab technician on their child's network, with a range of 0-2 metabolic doctors and 0-1 lab technicians per participant. The next most commonly identified health care providers were: dietitians (n=45 children's networks), family doctors (n=41), nurses (n=40), and pediatricians (n=40). Informal supports that parents frequently identified on their children's care networks included friends (n=40 children's networks) and family members (n=23).

Settings identified in care networks. The most common locations that parents identified on their children's care networks included a metabolic clinic (n=58 participants, 97%), clinics located in the community (e.g., primary care clinics) (n=57, 95%) and blood labs, either in community or at hospital (n=55, 92%). Other settings and clinics that were identified by at least five participants include a school or daycare (n=33, 55%), an optometry or ophthalmology clinic (n=15, 25%), a cardiology clinic (n=10, 17%), an audiology clinic (n=9, 15%), a neurology clinic (n=9, 15%), an orthopedic clinic (n=9, 15%) and an emergency department (n=8, 13%). A full list of all settings identified on these networks is provided in Appendix J.

Table 4. Providers and supporters commonly involved in care networks of children with IMDs, from parents' perspectives

Provider	Children with this provider in network n (%)	Range per participant (n) [a]	Children with this provider as a key provider in network n (%)	Key provider range per participant (n) [b]
Metabolic doctor	55 (92%)	0-2	40 (67%)	0-1
Lab technicians	55 (92%)	0-1	6 (10%)	0-1
Dietitian	45 (75%)	0-2	33 (55%)	0-2
Family doctor	41 (68%)	0-3	14 (23%)	0-2
Nurse	40 (67%)	0-5	11 (18%)	0-1
Friends	40 (67%)	0-1	0	//
Pediatrician	39 (65%)	0-3	27 (45%)	0-1
Extended family	23 (38%)	0-1	2 (3%)	//
Social worker	22 (37%)	0-3	4 (7%)	//
Daycare staff	21 (35%)	0-1	2 (3%)	//
Dentist	20 (33%)	0-2	6 (10%)	0-1
Occupational therapist	19 (32%)	0-4	4 (7%)	//
Physiotherapist	18 (30%)	0-3	6 (11%)	0-1
Teacher	18 (30%)	0-4	3 (5%)	//
Pharmacist	17 (28%)	0-2	4 (15%)	//
Ophthalmologist	16 (27%)	0-1	5 (8%)	//
Neurologist	16 (27%)	0-1	8 (13%)	0-1
Cardiologist	14 (23%)	0-1	3 (5%)	//
Audiologist	13 (22%)	0-2	3 (5%)	//
Genetic counsellor	13 (22%)	0-1	2 (3%)	//
ER staff (Doctors, nurses)	13 (22%)	0-3	2 (7%)	//
Geneticist	12 (20%)	0-1	8 (13%)	0-1

[a] Among children with at least one of this provider type in their care network
[b] Among children with at least one key provider of this provider type in their care network

Network archetypes. During the analysis of the care maps, we recognized the potential value of exploring whether there were defined groups or archetypes of care maps with similar characteristics. We devised these archetypes based on the distributions of network size, density, and centralization. We used post-hoc visualization of these distributions and discussion among the team to identify three care map archetypes: 1) small networks (11 or fewer providers based on an obvious split in the distribution of network size in the sample); 2) large but sparse networks (>11 providers and lower or equal to the median for either density or centralization or both among large care maps); and 3) large *relatively* connected networks (>11 providers and higher than the median for both density and centralization among large care maps).

1) Small networks (examples, Figure 2). Slightly over one third of participants (n=23, 38%) drew a care network for their child with 11 or fewer providers. The median network size for small networks was 9 (IQR 8 - 11). The median density for small networks was higher than that for the overall sample at 0.13 (IQR 0.08 - 0.21), meaning that a median 13% of all possible connections between providers were perceived as connections by parents. The median centralization for small networks was also higher than for the overall sample at 0.28 (IQR 0.21-0.32), meaning that on average there was a provider that held 28% of the connections reported in the network.
2) Large and sparse networks (examples, Figure 3). Slightly over one third of participants (n=23, 38%) drew a care map for their child with 11 or more providers listed on them and with a density ≤ 0.06 and/or centralization ≤ 0.18. The median network size for this archetype was 20 (IQR 15 - 27). The median density was 0.04 (IQR 0.02 - 0.06) and median centralization was 0.13 (IQR 0.1 - 0.17).
3) Large and *relatively* connected networks (examples, Figure 4). Close to a quarter of participants (n=14, 23%) drew a care map for their child with 11 or more providers and density > 0.06 and centralization > 0.18. The median network size for this archetype was 18 (IQR 16 - 19). The median density was 0.08 (IQR 0.07 - 0.11) and median centralization was 0.27 (IQR 0.23 - 0.35).

Through a post-hoc exploratory analysis, we found that children with small networks tended to be relatively younger, spent less time traveling to the metabolic centre, and were diagnosed with a disease in IMD Groups 1 and 2 relative to those with large networks (Appendix K). Those with large and sparse networks were comparable to those with sparse and relatively

more connected networks with regards to age, travel time to metabolic centre and IMD clinical trajectory group.

Figure 2. Examples of participant care maps that can be classified as part of the 'small' network archetype

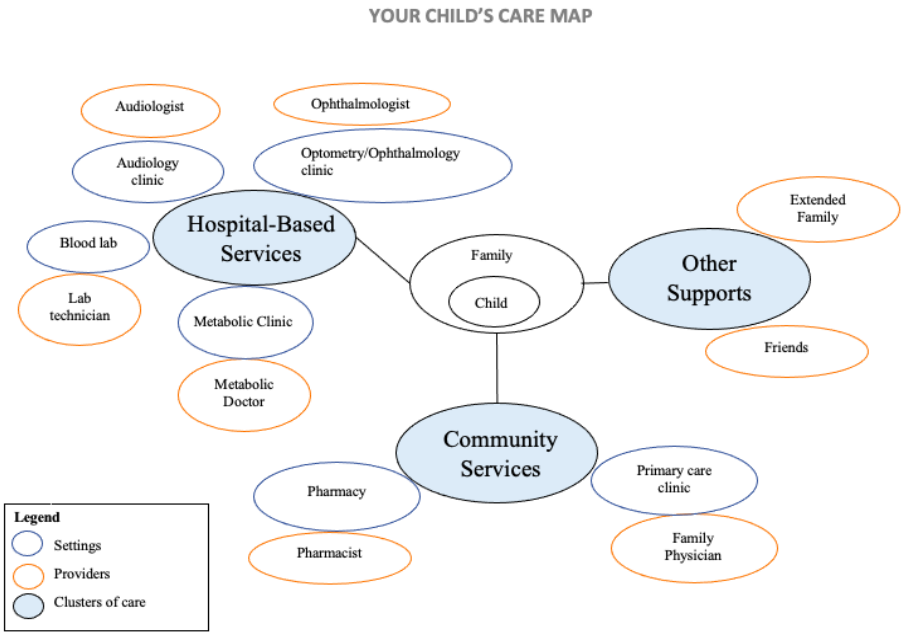

Figure 2 (a). Example #1 of a 'small' network (network of a child with an IMD in Group 1).

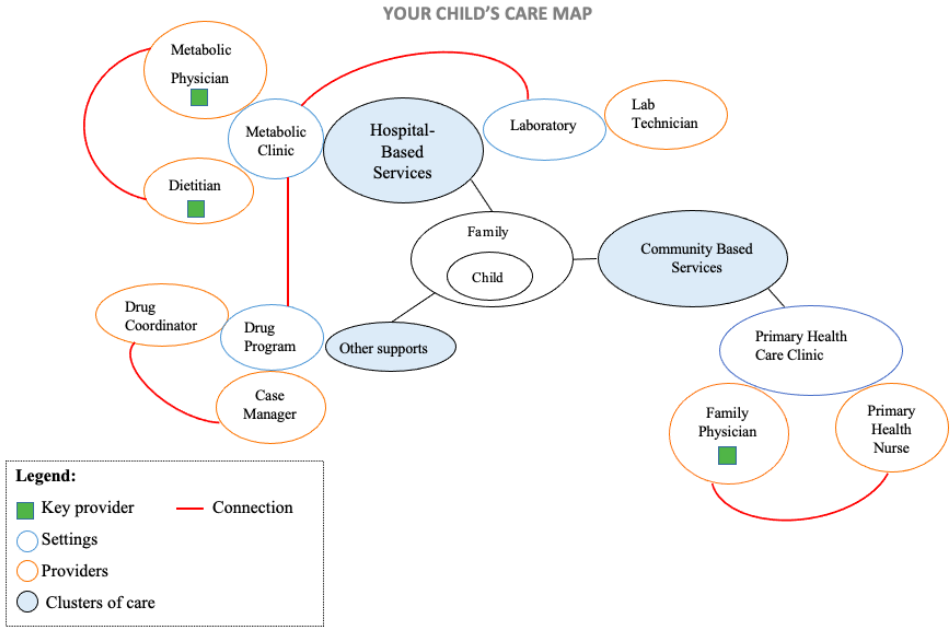

Figure 2 (b). Example #2 of a 'small' network (network of a child with an IMD in Group 1).

Figure 3. Examples of participant care maps that can be classified as part of the large and sparse network archetype

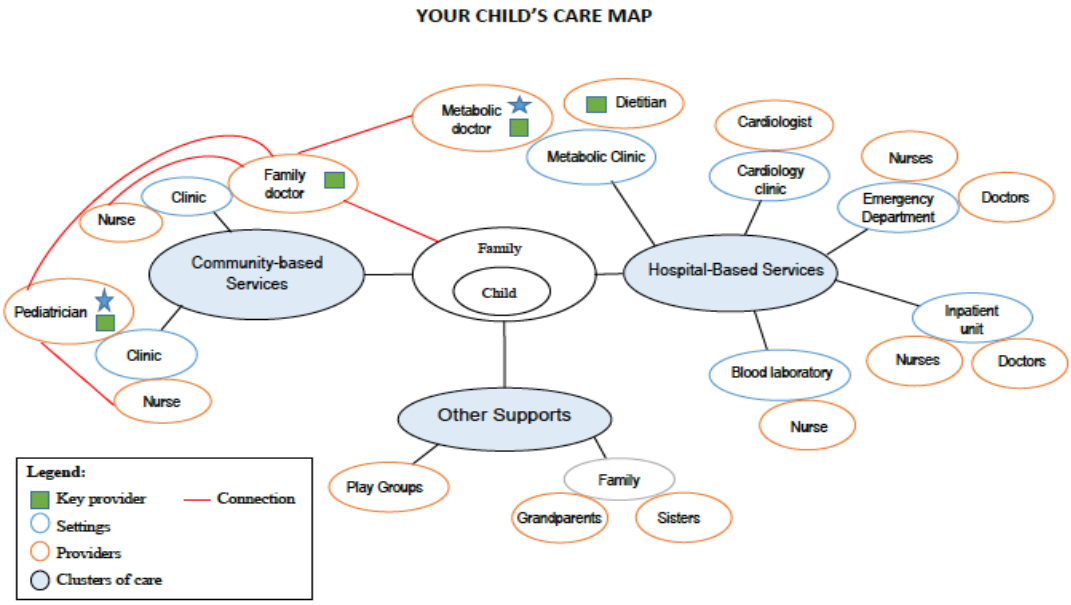

Figure 3 (a). Example #1 of a 'large and sparse' network (network of a child with an IMD in Group 2).

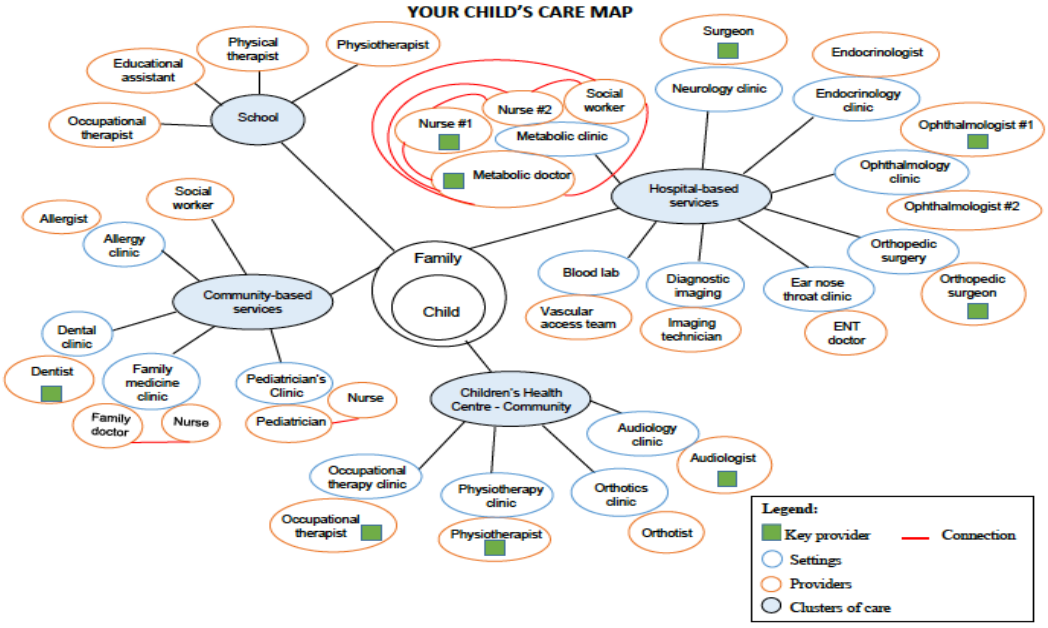

Figure 3 (b). Example #2 of a 'large and sparse' network (network of a child with an IMD in Group 3).

Figure 4. Examples of participant care maps that can be classified as part of the large and relatively connected network archetype

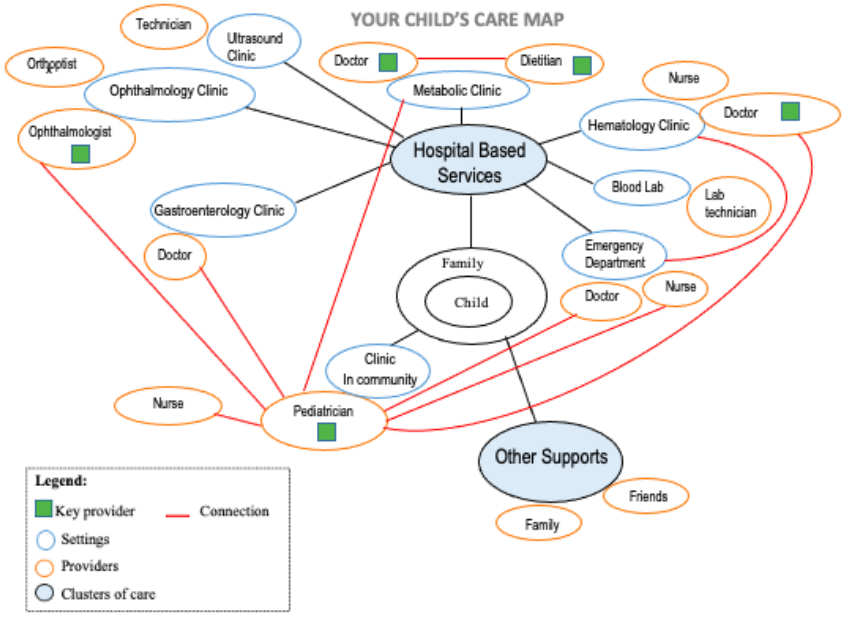

Figure 4 (a). Example #1 of a 'large and relatively connected' network (network of a child with an IMD in Group 2).

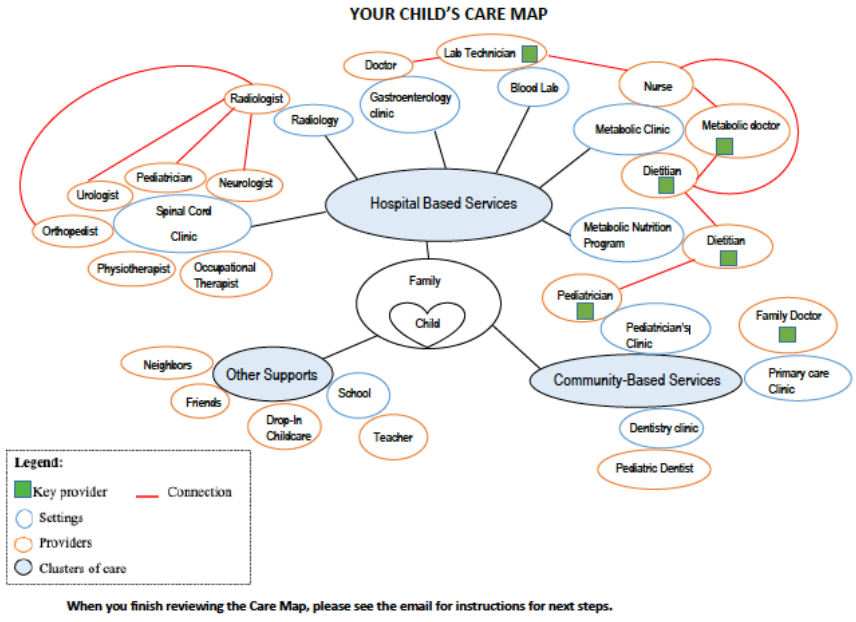

Figure 4 (b). Example #2 of a 'large and relatively connected' network (network of a child with an IMD in Group 1).

2.3.2.2 Characteristics of children's care networks: Qualitative findings

Parental reflection on process of drawing care maps. Most parents shared that the process of drawing a care map required reflection on their child's IMD trajectory, starting with care providers they have been seeing since the IMD diagnosis and considering who was added or removed from their child's care team along the way. Many parents noted that they tried to include all of the providers and services that are important for any aspect of their child's care (additional illustrative quotations supporting all qualitative themes, Appendix L):

> *"So, what I did was, um, kind of even went back to diagnosis, and how those people start, like, how that all started in our lives. So, that's where I went with Metabolics, right? Like, they were critical. Neurology was at the time [of diagnosis]. We gained a Pediatric doctor. The Ears, Nose and Throat was very important at the time. So, I started kind of there, and then thought, "Okay, who is not involved, really anymore" and really, they all still are in some way connected."* (Participant #1, Mother of child with an IMD in Group 3 and 'large and sparse' network)

Many parents we interviewed noted that when looking at the completed care map, they were somewhat surprised or in awe of the large number of providers and services involved in their child's care, the connections between the different providers and how the different parts of their child's network work together:

> *"It was interesting to, like, see the number of, like, connections we have for [child's name]. Like, there's a lot more people involved than I thought there was (laughs)."* (Participant #4, Mother of child with an IMD in Group 2 and 'large and sparse' network)

The dynamic nature of IMDs care We asked interviewed parents to take us through the care map, providing an overview of their children's care routines and the different care providers that they interact with on a regular basis. Parents often described some level of predictability in terms of a schedule of follow-up appointments with regular providers such as a metabolic physician, specialist, or pediatrician. However, when discussing these care routines and frequency of interactions with different providers, parents also frequently added that everything remained in

flux. We found from parents' descriptions that families have a 'journey' with the disease. As they navigate this journey, at certain points, their child's symptoms might be stable – so their care may consist mainly of routine check-ups. At other points, the child might be experiencing symptoms that require frequent interactions with the health care system. Parents described that children's care networks changed with age due to changes inherent to child development (e.g., starting school, attending summer camps) as well as due to changes specific to their IMD. Furthermore, changes happen because of system and provider factors (e.g., providers retiring, programs ending):

> *"Um, well now because she's… every blood test that we get it done and it comes back as normal, we don't get to see the doctors that much, maybe once a year now. Before it was maybe 2 or 3 times a year, but now because the blood tests came back normal, then it is like once a year."* (Participant #7, Father of child with an IMD in Group 2 and 'small' network)

Parental responsibility – parents as 'managers' of their children's care networks. Among the parents we interviewed, we found that, rather than acting only as recipients of care from the network of providers for their child and family, they typically felt responsible for establishing, adapting, and organizing their child's care network. This parental agency and effort started early in the disease journey as parents worked to identify services and providers that were needed to meet their child's care needs. Several parents explained that arriving at a network of care that met their child's care needs was a process that took a few years initially and was ongoing. Parents reported using several strategies to ensure that their child received needed care, including leveraging their social connections, seeking out opportunities in the community, using resources available to them and becoming advocates. This was described as time-consuming, overwhelming, and impactful:

> *"My role was, it took, I would say, 2 to 3 years after initial diagnosis for me to work the system here and there to set up this [care network]. I didn't realize, in the beginning, that this was what was going to be the end result of that. For instance, the [service name],*

> *finding that [services coordinator], took me going through several places…and asking questions regarding how, like, "This is really overwhelming. Do you know of something that might be of assistance?"…There was a lot of asking of questions, of feeling overwhelmed at the beginning, not being able to juggle a lot. I did end up having to leave my full-time job. There was no way at the beginning, that I would be able to work and make all of this happen."* (Participant #8, Mother of child with an IMD in Group 3 and 'large and relatively connected' network)

Participants who lived in rural communities where families sometimes personally know their providers outside of a health care context often discussed leveraging these social connections, which they described as positively impacting their comfort level with providers and allowing the family to access care more readily:

> *"Everybody always knows everybody, or you know, the clinic works with the pharmacist, or, um, like, our pharmacist is actually a relative, so (laughs). It's quite nice just to phone and, they know who we are, and they know [child's name], and um, if there is something that, like, if she had a prescription change or something and they weren't sure about it, they were good about being able to phone, or, or me phone."* (Participant #5, Mother of child with an IMD in Group 1 and 'small' network)

Parents acknowledged that their responsibility for the network did not end once a child's care network was established. Most parents had an active role in continually coordinating their child's care, especially by sharing information among providers and advocating to ensure access to services. In addition, due to the dynamic nature of children's care networks as described earlier, parents reported an ongoing need to adapt to change as their child's care needs changed and their providers changed (e.g., due to retirements or providers moving practices):

> *"And then, we've had a huge turnover in our [community services] team regarding people that are in those particular positions, and that's difficult because of the rare disease aspect of it…So, I am finding myself doing a lot of education right now, and I am managing a lot of the [community services] on my own again right now."* (Participant #8, Mother of child with an IMD in Group 3 and 'large and relatively connected' network)

Given the effort required by parents to coordinate and continually manage their children's care networks, many parents expressed relief about experiences where they felt that they could trust that a provider was able to coordinate their child's care without parental involvement.:

> *"I have very much trust for both of them. That they didn't need me in the middle, right? Like, they just, I didn't have to, you know, call the Dentist office, and you know, double check something, or make sure something else was, you know, aligned, or call the doctor. Like, I just had complete trust that this was going to, they gave it to them, they took care of it, and I didn't need to be in any way consulted or (laughs). You know, I did not need to follow up with anything, and I think that is what is critical."* (Participant #1, Mother of child with an IMD in Group 3 and 'large and sparse' network).

2.3.2.3 Characteristics of children's care networks: Mixed-methods integration

We integrated the quantitative and qualitative findings regarding the characteristics of children's care networks as perceived by parents (Table 5). This revealed that there was variability in the structure of children's networks associated with child age and IMD Group. However, overall, parents tended to perceive a large number of providers and lack of connectedness, necessitating an intensive 'manager' role. They used resources available to them, including social connections, to improve care.

Table 5. Joint display: Integrated summary of characteristics of children's care networks

Summary	*Quantitative Findings*	*Qualitative Findings*	*Summary quotation*
Parents of children with IMDs described health care networks that were variable in size. Networks were dynamic due to children's changing needs and changes to provider and service availability. Parents felt responsible for establishing their child's care network and this role was ongoing.	○ Parents identified a median of 14.5 providers and 89 provider types. ○ Most common providers: metabolic doctors, lab technicians, dietitians. ○ Larger networks were associated with older children and progressive, multi-system disease.	○ As informal 'managers', parents felt responsible for their child's care network. ○ This was often described as overwhelming.	*"At the beginning we felt, we were in the learning phase, learning all about the condition, and what it means for our family, and once we had a better grasp of that and felt a little bit more on our feet with the information, then we started to ask more questions about how we could do things better. And that's how we ended up being connected to the pediatrician locally, so that we could do things like blood work without the drive. Those types of things, yes, I think we had a role in that."* (Participant #10, Mother of child with an IMD in Group 1 and 'large and sparse' network)
While there was variation across networks in the numbers and types of connections between providers, parents generally perceived networks to be sparsely coordinated. Rather than solely being recipients of care, parents felt they had to step in and provide a coordinating role.	○ Networks had a median density of 0.08 (only 8% of possible pairwise connections were identified by parents) and a median centralization of 0.23 (the most connected provider was involved in approximately 23% of the pairwise connections).	○ Parents had to compensate for the lack of care coordination. ○ Parents frequently described adopting an advocacy role and leveraging resources available to them to improve access to care for their child.	

2.3.3 Key providers and relational continuity

2.3.3.1 Key providers and relational continuity: quantitative findings

Key providers. Parents identified 55 unique types of "key" providers on their children's care networks. Of these 55, 46 were health care providers (e.g., nurses, doctors) while 9 were informal supports and educational providers (e.g., educational assistant, friends). The majority of participants identified a metabolic doctor (n=40, 67%) and a dietitian (n=33, 55%) as key providers on these networks of care. Close to half of participants (45%) identified a pediatrician as a key provider. A summary of the most commonly identified key providers is provided in Table 4.

Relational continuity. We measured the relational continuity of those key providers who were health care providers (rather than educational providers and informal supports) by asking the parent how well a provider knew the child and family. A majority of participants (80%) reported having at least one health care provider in the network that knew their child and family fairly well or very well. A high proportion of participants (94%) who identified a dietitian as a key provider indicated that at least one dietitian in their child's network knew their child and family very well or fairly well (Figure 5). A majority of participants who identified a metabolic doctor (75%) or a pediatrician (70%) as key providers also indicated that at least one of these provider types knew their child and family very well or fairly well. Remaining key providers were rated by a minority of participants as knowing the child and family very well or fairly well (Figure 5).

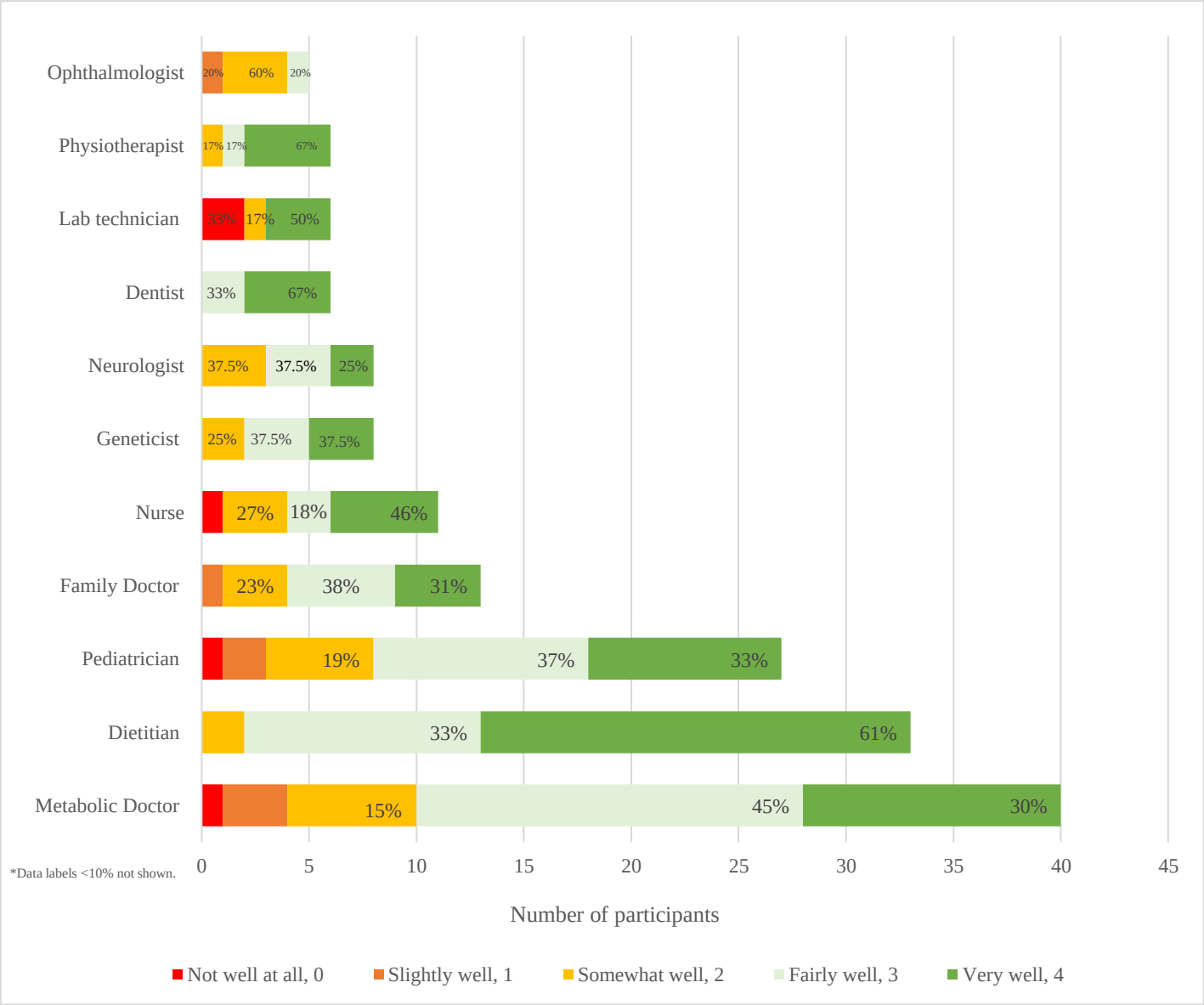

Figure 5. Relational continuity ratings for key health care providers identified on care maps by at least 5 participants

2.3.3.2 Key providers and relational continuity: qualitative findings

The characteristics of key providers. As identified in the descriptive analysis above, metabolic doctors, dietitians and pediatricians were most commonly identified as key providers. During the interviews, we asked parents what makes someone a key provider or how they chose to designate certain providers in the network as key to their child's care. Parents identified several provider characteristics that were important to this designation. Some providers were considered

key even if they had only one of these characteristics while others exhibited many of the characteristics.

One of the characteristics that parents felt made someone a key provider was their expertise in an area of child's health that a parent described as impactful in the child's daily life or disease progression (e.g., monitoring of a specific symptom or aiding in the management of diet). Sometimes this was a specialist in a discipline important to the child's care and other times this was a provider who took the time to research and learn about the child's disease:

> *"I think, just because [child's] condition is so rare, she had to do a lot of work to research the condition itself and how it is to be treated, and she has taken on a big responsibility, in reaching out to other doctors at other facilities, to find information and bring it back to us and talk to other care providers in other centres, to compare and learn, and so I think, just on the whole knowledge base, learning and understanding, and all of that, that's why she's so key in his care."* (Participant #9, Mother of child with an IMD in Group 3 and 'large and relatively connected' network)

Another important characteristic that parents reported having considered in designating someone as a key provider on their child's care map was their availability. Due to the nature of IMDs, parents noted that they often had questions regarding aspects of care management such as diet prescriptions, medications, and other therapies; in some cases, these questions arose in response to changing symptoms that could become medical emergencies. Many parents identified that they appreciated and sought out providers who were responsive and could address their concerns and questions in a timely way:

> *"I would actually put that the hematologist's nurse is also a key provider, because if I have any issues, like, if my daughter was not feeling well, and I am not sure what to do, it is the nurse I would talk to. I would call them, and they would call me back, sometimes within like minutes, sometimes they are answering the phone, like, it's very easy to get a hold of them and talk to them day to day. One time when my daughter was sick, I talked to them, like, ten times in a day, and, you know, and then we were just keeping track of her temperature through the day"* (Participant #2, Mother of child with an IMD in Group 1 and 'large and relatively connected' network)

Some parents identified that they designated a provider as a key provider because of their role as a central person in the network of care that connects the family with other providers and services or in the coordination of different services and providers:

> *"She also, as the Metabolic doctor, is coordinating with all of those other clinics that you see on Hospital-Based Services. She's the one who puts in a referral for all those different services, and then they report their findings back to her, so she collects all of the information from all of those other hospital resources from hospital #1 and keeps it in his file. Although I do talk to them, she also then goes over the findings with me and relates it back to his overall care."* (Participant #8, Mother of child with an IMD in Group 3 and 'large and relatively connected' network)

Finally, another attribute that parents indicated was important was that a care provider had a 'human side' to them – they are warm and friendly in their interactions with the family:

> *"Yeah, and [dietitian's] just a very like, kind person, and not ever like, hard on us if we are not like, sticking to his diet properly (laughs) and just very understanding that yeah, we have a whole other life going on, and if like, we're not strictly following her rules (laughs), then she's not mean to us, so."* (Participant #4, Mother of child with an IMD in Group 2 and 'large and sparse' network)

Relational continuity. As reported above, a high proportion of participants who identified a dietitian or a metabolic doctor as key providers also rated at least one of these provider types as knowing their child and family very well or fairly well. Many parents elaborated that they appreciated when a provider was fully aware of their child's medical history as it meant that they did not have to repeatedly retell their child's IMD journey. However, relational continuity was more than not having retell their story. Parents told us that they valued providers that they had an ongoing relationship with as this could also help them establish an emotional connection and a level of familiarity with the child and family:

> *"We have spent quite a bit of time with that particular person over the span of several years. She is familiar with all the ins and outs of our family and our son. Like, having a child with this particular metabolic disorder, they have to be kind of familiar with what his home life is like, and what kind of services he is receiving outside of their hospital-base, so that's how she knows just about everything about what we are up to, and what my son does*

> *when he is not inside that hospital. She has an interest in that too"* (Participant #8, Mother of child with an IMD in Group 3 and 'large and relatively connected' network)

Having the same provider over time was not always associated with relational continuity. Sometimes parents described a provider whom they saw repeatedly over time but who did not seem to know the child and family well:

> *"Like, I don't think she would recognize any of us if we passed her in the hallway at the hospital. She doesn't know a lot about the metabolic issues specifically, but she is a specialist in what is wrong with his eyes, so yeah. She doesn't know him well, but I'm okay with that, because she knows her stuff (laughs)."* (Participant #4, Mother of child with an IMD in Group 2 and 'large and sparse' network)

Mutual trust. We learned from the parents we interviewed that trust is an important requirement for developing positive relationships with their child's care providers. Many of the parents reported that they strongly valued care providers who trusted family members when they provided input about their child's IMDs care. In parallel, parents also valued providers in whom they could place their trust. Parents described this in terms of providers who saw "the whole child", those whose knowledge and expertise were trusted by the family, and those who reliably followed through on commitments so that the parent did not need to double check (e.g., on whether information was shared as promised, referrals or prescriptions sent, appointments set, etc.):

> *"And so much, that his, the outcome of that phone call was that I knew [son] best, and if I felt like, over a duration of time, like, you know, like, I'm not seeing any of those symptoms subside, that, um, to give him a call, that he would call in the increase. Like, I could go ahead and start with the medication I had, and that was, like, he is trusting, right?"* (Participant #1, Mother of child with an IMD in Group 3 and 'large and sparse' network)

2.3.3.3 Key providers and relational continuity: mixed methods integration

We integrated the quantitative and qualitative findings regarding parents' perspectives on the roles of key providers and the concept of relational continuity (Table 6). This revealed that

there was a variety of providers that were designated as key. Parents designated someone as a key provider due to their impactful role in their child's care network, their availability, and their affect. The most commonly identified key healthcare providers were metabolic doctors, dietitians, and pediatricians. A high proportion of participants who identified a metabolic doctor, dietitian, or pediatrician as a key provider indicated that at least one of these provider types in their child's network knew their child and family very well or fairly well. Parents emphasized the importance of relational continuity and mutual trust with care providers.

Table 6. Joint display: Integrated summary of key providers and relational continuity

Summary	*Quantitative Findings*	*Qualitative Findings*	*Summary quotation*
Parents of children with IMDs described various types of key providers but dietitians, metabolic doctors and pediatricians were most commonly named. These providers were selected because of their expertise, or central role, among other characteristics.	○ Parents identified 55 unique types of key providers. ○ A majority of participants (80%) reported having at least one health care provider in the network that knew their child and family fairly well or very well.	○ Characteristics leading to a provider being designated as key included one or more of: disease expertise, availability, warm demeanour, and central role in coordinating care ○ Parents valued providers that they had an ongoing relationship with; relational continuity included the concepts of emotional connection and familiarity with the child and family. ○ Trust was very important to parents: parents particularly valued providers who demonstrated that they trusted parents to know the child's needs.	*"We have had both of them involved in my child's care since the diagnosis. It was actually the metabolic doctor that had reached out to us to let us know about the diagnosis, and back at the beginning, we saw them on a weekly basis for probably 2 or 3 years, so there is quite a relationship that has been built up there over time. We are down to only seeing them about 3 times a year now, but there is still interaction over email as well."* (Participant #10, Mother of child with an IMD in Group 1 and 'large and sparse' network).

2.3.4 Providers as part of a dynamic network and care coordination

2.3.4.1 Providers as part of a dynamic network and care coordination: Quantitative findings

For each provider type, we calculated degree centrality, which refers to the number of connections that a provider was perceived to have within a care network. When children had multiple providers of the same type, we used the highest degree centrality for that provider type in our analysis. Table 7 summarizes the degree centrality of the most common provider types included in children's care networks, based on connections perceived by parents. The highest median degree centrality across all provider types is three, meaning that on average, parents did not perceive any provider type to have more than 3 connections to other providers in the network. The provider types with a median degree centrality of three included the metabolic doctor (IQR 2-4) and the dietitian (IQR 2-4). Family doctors had a median degree centrality of 0 (IQR 0-1) while pediatricians had a median degree centrality of one (IQR 0-3). Among key providers, metabolic doctors and dietitians who were designated as key providers also had a median degree centrality of three (IQR 2-4), family doctors who were designated as key providers had a median degree centrality of one (IQR 0-3), and pediatricians designated as key providers had a median degree centrality of one (IQR 0-1) (Table 7).

Another provider type-level measure was the provider's share, which we defined as the proportion of pairwise connections in a network that involved that provider. Again, for children with multiple providers of the same type, we used the highest share for that provider type in our analysis. The metabolic doctor had the highest share across the different provider types with a median share of 0.19 (IQR 0.13 – 0.25), indicating that when children had a metabolic doctor in their care network, on average, 19% of the pairwise connections that parents observed were connections involving the metabolic doctor. This was followed by dietitians, with a median share

of 0.17 (IQR 0.08 – 0.25) in networks with that provider type. Metabolic doctors designated as key providers had a median share of 0.20 (IQR 0.13 – 0.25) and dietitians designated as key providers had a median share of 0.20 (IQR 0.11- 0.25).

Table 7. Degree centrality and share of providers and supporters involved in care networks of children with IMDs, from parents' perspectives

Provider	Median (IQR) degree centrality [a,b]	Median (IQR) share [a,c]	Median (IQR) degree centrality for key providers [d,e]	Median (IQR) share for key providers [d,f]
Metabolic doctor	3 (2-4)	0.19 (0.13-0.25)	3 (2-4)	0.20 (0.13-0.25)
Lab technicians	1 (0-3)	0.07 (0 - 0.15)	1 (0-3)	0.09 (0 - 0.15)
Dietitian	3 (2-4)	0.17 (0.08 -0.25)	3 (2-4)	0.20 (0.11-0.25)
Family doctor	0 (0-1)	0 (0-0.10)	1 (0-3)	0.06 (0-0.17)
Nurse	2 (1-4)	0.13 (0.06 -0.18)	3 (3-4)	0.17 (0.15-0.20)
Friends	0	0	//	//
Pediatrician	1 (0- 3)	0.06 (0- 0.13)	1 (0-1)	0.03 (0-0.06)
Extended family	0	0	//	//
Social worker	1 (0-3)	0.06 (0 -0.16)	//	//
Daycare staff	0	0	//	//
Dentist	0 (0-1)	0 (0 - 0.04)	0 (0-1)	0 (0-0.02)
Occupational therapist	0 (0-1)	0 (0-0.05)	//	//
Physiotherapist	0 (0-1)	0 (0-0.02)	0	0
Teacher	0	0	//	//
Pharmacist	0 (0-1)	0 (0-0.02)	//	//
Ophthalmologist	0 (0-1)	0.005 (0.0 -0.03)	//	//
Neurologist	1 (0-4)	0.03 (0.0-0.13)	2 (0-5)	0.04 (0.02-0.09)
Cardiologist	1 (0-2)	0 (0-0.07)	//	//
Audiologist	0 (0-2)	0 (0-0.01)	//	//
Genetic counsellor	3 (2-3)	0.14 (0.10 – 0.17)	//	//
ER staff (Doctors, nurses)	1 (0-1)	0.03 (0-0.25)	//	//
Geneticist	3 (2-5)	0.12 (0.04- 0.17)	5 (3-6)	0.11 (0.14-0.27)

[a] Among children with at least one of this provider type in their care network
[b] For children with more than one of this provider type, we used the highest degree centrality for this provider
[c] For children with more than one of this provider type, we used the highest share value for this provider
[d] Among children with at least one key provider of this provider type in their care network
[e] For children with more than one key provider of this provider type, we used the highest degree centrality for this key provider
[f] For children with more than one key provider of this provider type, we used the highest share value for this key provider

Care coordination. Parents rated each key health care provider on their child's care map according to how well they were perceived to coordinate care with other providers. A large proportion of participants who identified a metabolic doctor (85%) or dietitian (91%) as a key provider rated at least one of these providers types as coordinating care fairly well or very well (Figure 6). A majority of participants who identified a pediatrician (71%) as a key provider also rated at least one of pediatrician in their child's network as coordinating fairly well or very well.

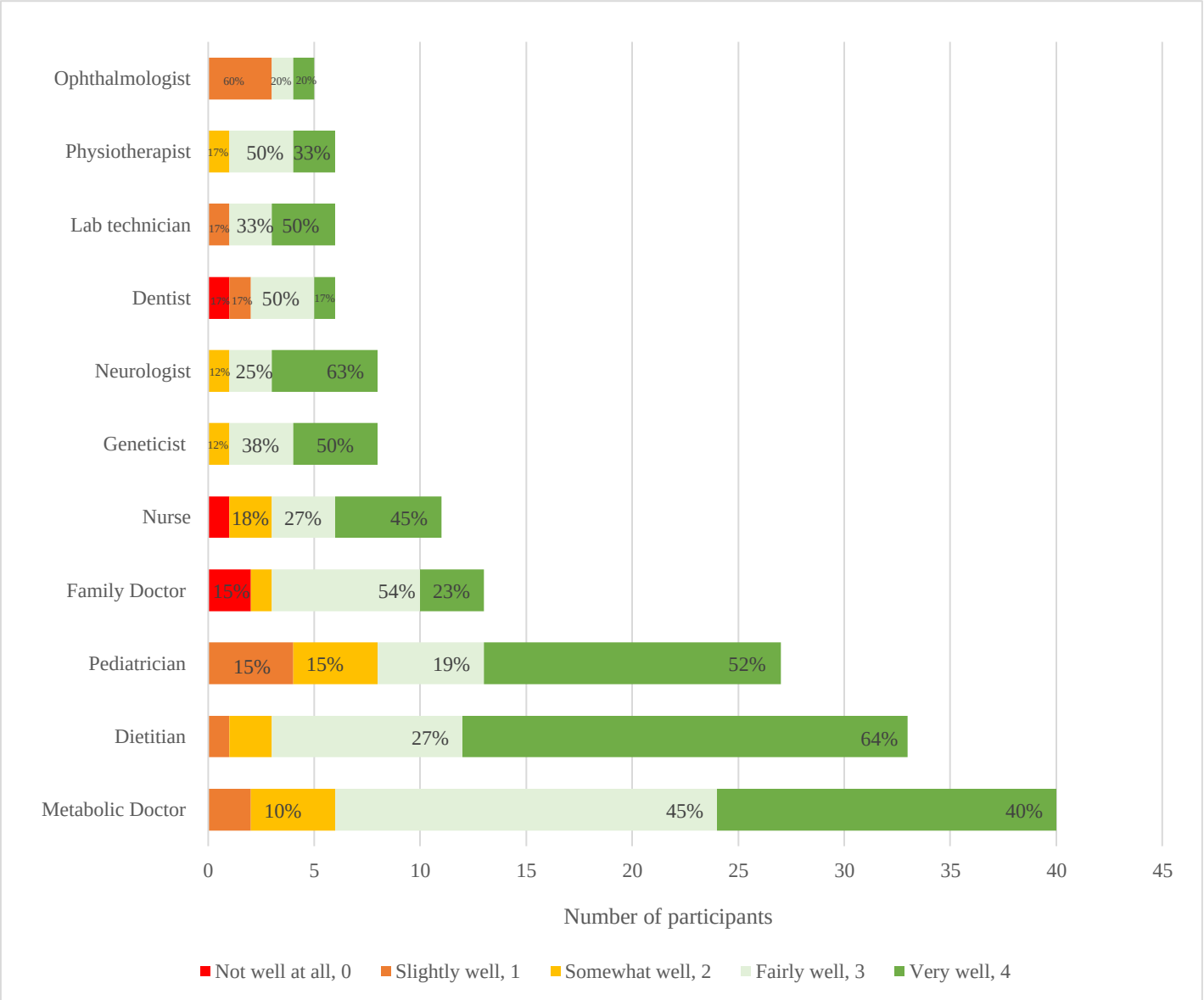

Figure 6. Care coordination ratings for key health care providers identified on care maps by at least 5 participants

2.3.4.2 Providers as part of a dynamic network and care coordination: Qualitative findings

Ways of coordinating care. As reported above from the quantitative analysis, no provider type had a median of more than three parent-perceived pairwise connections to other providers in children's care networks. During the interviews, parents elaborated further on the nature of the connections that they perceived between providers. We learned that parents viewed care coordination as one or more of these three main activities:

1) Providers can actively work as a team to organize care, for example by meeting the family together, coordinating appointments, or working to schedule multiple procedures on the same day:

   "*Like, for example, the hematologist and the ophthalmologist, they both have a procedure that they want to do. So, the pediatrician will be the one that's going to try and coordinate them to both do it at the same time, at the same hospital, so we only have to put her under once. So, she would be the one to kind of coordinate all that.*" (Participant #2, Mother of child with an IMD in Group 1 and 'large and relatively connected' network)

2) One provider can provide access to another (gatekeeper) through referrals or consultation:

   "The metabolic doctor is one that has been doing all the referrals for us to see specialists. For example, the ear, nose, and throat specialist, the pediatric respirologist." (Participant #6, Mother of child with an IMD in Group 3 and 'large and sparse' network)

3) Providers can share information with one another. This can occur "passively", for example, when providers share the same electronic medical record system and can see one another's notes; or it can occur "actively" when providers communicate directly with one another:

   "So, the cardiologist, although is the only specialty that is not in Hospital-Based Services #1, the referral was put to that particular cardiologist, and that cardiologist sends his findings to that Metabolic doctor, and her and her nurse are responsible for setting up the Home Care for the Home Care nurse. The Home Care nurse sends weekly reports to them." (Participant #8, Mother of child with an IMD in Group 3 and 'large and relatively connected' network)

Some parents noted that they valued passive communication sharing because there was less risk that a provider would be excluded from receiving important information or that the responsibility would fall to the parent to share information between providers. At the same time, when providers were sharing information with each other, there was a risk that the family would be left out of the communication and may thus feel external to the health care team:

> "*I definitely think that [online communication system], bringing that it was a really great resource because it saves me from explaining all of the times, that we have seen which pediatrician, which doctors, which specialists, what happened here. And you know, when they tell me what happened at the meeting, I only get like, maybe a snippet of what has actually happened. I don't get the full picture all of the time. So, for example, the audiologist will tell me something, and I would say, "Okay, that's great", but when I go see somebody else, they will tell me things like, "Oh, I read the audiologist's letter, [child]'s hearing got worse", and I would say, "What? That's not what she said to me", and "What did the letter say?"* (Participant #6, Mother of child with an IMD in Group 3 and 'large and sparse' network)

Need for a network "go-to" person. We found that for most parents we interviewed, there was at least one person in their network who they identified as key due to their availability and timely responsiveness to family concerns/questions: a "go-to person". Building on the earlier themes of availability and trust as provider characteristics that were valued by parents, the "go-to person" was typically a provider who had demonstrated that they trusted the parents and who either had the capacity to directly respond to the child's needs or to provide access to other providers who could respond. This was a "network-level" concept because we found that while many providers were considered key and described favourably for the reasons outlined above, parents frequently identified one single "go-to person" and sometimes described working to find a provider who could be in this role in the network:

> *"Um, usually she's the one that we would, I would phone or, she would phone me, and if I had a question, she would find it out for me. If she didn't know she'd phone the doctor that did, or she would have someone contact me, it seems. Like, um, they have been very helpful that way. Or, when I have needed the appointment, she was who I was letting know I was*

> *coming that day, so then she made sure that everybody else kind of had got a hold of me to schedule an appointment."* (Participant #5, Mother of child with an IMD in Group 1 and 'small' network)

Impact of poor care coordination. When care coordination was perceived as inadequate, parents described having to work as the 'middle person' to fill the gap, particularly in relaying information between providers. Parents identified this an important source of stress; some noted that they did not feel equipped with the necessary medical knowledge or that they worried that they were missing important information:

> *"At some point, I felt like they were going through me to get to each other, and I finally said, "Can you guys just talk? You guys just talk, keep me in the loop."" (*Participant #9, Mother of child with an IMD in Group 3 and 'large and relatively connected' network)

2.3.4.3 Providers as part of a dynamic network and care coordination: Mixed methods integration

We integrated the quantitative and qualitative findings regarding the concepts of providers as part of a dynamic network and care coordination, from the perspective of parents (Table 8). This revealed that on average, no provider type in these care networks had more than 3 connections to other providers in the network. The provider types with a median degree centrality of three included the metabolic doctor (IQR 2-4) and the dietitian (IQR 2-4). The dietitians and metabolic doctors who were designated as key providers were perceived to coordinate care fairly well or very well. Parents viewed and considered care coordination to entail either providers actively working as a team to organize care; referrals or consultation; or providers sharing information with one another.

Table 8. Joint display: Integrated summary of providers as part of a dynamic network and care coordination

Summary	*Quantitative Findings*	*Qualitative Findings*	*Quotation*
Parents generally did not perceive that providers across the network were well connected; this was particularly true for non-metabolic specialists, including primary care providers. In turn, when care coordination was perceived as inadequate, parents described having to work as the 'middle person' to fill the gap, particularly in relaying information between providers.	○ Median degree centrality (number of connections to other providers in the network): highest for metabolic doctors (median degree centrality of 3) and dietitians (median of 3); lower for family doctors (median of 0) and pediatricians (median of 1). ○ A high proportion of participants who identified a metabolic doctor (85%) or dietitian (91%) as a key provider also rated at least one of these provider types as coordinating care fairly well or very well.	○ Parents viewed care coordination as: Providers actively working as a team to organize care; referrals or consultation; or sharing information with one another. ○ One strategy parents used to fill in care coordination gaps was to identify a "go-to person": a provider who they felt would trust them as parents and who could either directly respond to the child's needs or provide access to other providers who could respond.	*"Basically, the nurse is the person that I talk to the most. Usually, I would send an email to the nurse. The nurse would relay all that information to the metabolic doctor, and then she would just send back a brief overview of what she told the doctor and what needs to happen, or what she has done, or what not. So, she's basically the main person that I connect with."* (Participant #6, Mother of child with an IMD in Group 3 and 'large and sparse' network)

2.3.5 Summary of parent recommendations for care network improvements

Most of the parents we interviewed described that they were generally satisfied with their children's health care networks, however, as noted above, many identified themselves as having a critical role in reaching the point of having a satisfactory care network, through their work to identify providers and services and manage and maintain relationships over time. Parents often noted that there were areas in need of improvement. When we explicitly asked how things could be better, many of the suggestions parents made to improve care networks for children with IMDs could be grouped into three broad categories: 1) improving knowledge and capacity among non-

specialist health care providers; 2) working on personal connections and relationships; and 3) better care coordination.

1) Parents valued providers who were knowledgeable about their child's IMD. Due to the rarity of these conditions, families identified that they frequently encountered primary health care providers with low knowledge of the child's condition who were thus not able to provide necessary advice and medical management:

> *"I wish our family doctor was a little bit more helpful. He has like, really, since [child's name] was a baby, really taken like, a hands-off approach, because he doesn't understand his condition (laughs), or know anything about it. So, yeah, we have really seen him for immunizations. We saw him for well-baby checks when he was a baby, but even then, like, there was no follow-up even when he was, like, not meeting his milestones, because the metabolic doctor, had already, was already on it, and ahead of it. (…) We've had to, I think, because there is that lack of support, I think that's why the metabolic doctor sort of got put into the role for us as like our go-to."* (Participant #4, Mother of child with an IMD in Group 2 and 'large and sparse' network)

2) As noted earlier, parents described the importance and need of personal connections with their child's health care providers. Children of the parents we interviewed frequently had large care networks and frequent encounters and parents identified the value of relationships with providers who took the time to understand and connect with the child and the family:

> *"Again, it was that personal connection that we see them face-to-face, and we would be sharing information about the condition in our lives, and how we are dealing with it. I guess that emotional personal connection, as well as you know, the more clinical (laughs) connection, yes"* (Participant #10, Mother of child with an IMD in Group 1 and 'large and sparse' network)

3) Many parents we interviewed discussed their concerns about care coordination. Having someone on the team coordinating the network would mean that the family would not have to direct their children's care and constantly re-tell their IMD story or relay information between different providers. This was identified as important not only to reduce the workload for

parents but also because parents perceived it as sometimes necessary for access to some services and products (e.g., medications, referrals, etc.):

> *"I don't know, maybe meds, medication-wise could be better flowing. I don't know why that, to me, it just feels like there's a bit of a, like, just the. They don't do that like they used to, like, where, you know what I mean, you are leaving an appointment, and "Where are you headed to? What office, and we will send that in ahead of time". Like, I just don't feel like that happens as much as it used to. Right? So, I go with the script. Right, like, that kind of, that? So, that's why it kind of stands alone, right? It should be, I would feel like that should be connected to all of them"* (Participant #1, Mother of child with an IMD in Group 3 and 'large and sparse' network)

2.4 Discussion

Our findings highlight that the health care networks of children with IMDs are large (parents identified a median of 14.5 providers) and heterogeneous, composed of a variety of health care providers, education providers, and informal supports. Our finding that parents reported a large number of services and providers to support the medical and social needs of their children with IMDs is aligned with the results of studies of children with a range of diagnoses leading to medical complexity.[37–39] We found variation among IMDs, with older children and those with diseases that were multi-system and progressive in nature having the largest care networks. Through a post-hoc exploratory analysis, we classified networks as 1) small networks (11 or fewer providers based on an obvious split in the distribution of network size in the sample); 2) large but sparse networks (>11 providers and lower or equal to the median for either density or centralization among large care maps); and 3) large *relatively* connected networks (>11 providers and higher than the median for both density and centralization among large care maps). Children with small networks tended to be relatively younger, spent less time traveling to the metabolic centre, and were diagnosed with a disease in IMD Groups 1 and 2 relative to those with large networks. Those with large and sparse networks were comparable to those with sparse and relatively more connected networks with regards to age, travel time to metabolic centre and IMD clinical trajectory

group. Overall, based on parent reports of pairwise connections between providers, children's care networks were neither highly connected (median density=0.08) nor highly centralized (median centralization=0.23), meaning that providers were not perceived to have much contact/connection with one another and there was not a single provider who would be well-placed to act as a coordinator for the full network. This was corroborated by the qualitative interviews which revealed that parents tended to act as 'managers' of their children's networks and felt responsible for establishing, adapting, and organizing their child's care network, across all network archetypes and IMD groups. The parent 'manager' role was reported to sometimes be overwhelming. A majority of parent participants in our study sample were university graduates with total household incomes of $100,000 CAD or higher; the difficulty of managing these networks would likely be even more pronounced for those with fewer resources, who may not have had the time or ability to participate in our study. Being able to participate in English was an eligibility requirement for our study; those with language barriers to participating may also face language barriers when managing their children's health care, further underscoring that the degree to which study participants felt overwhelmed may underestimate these challenges among parents of children with IMDs in the population.

The majority of participants identified a metabolic doctor (67%) and a dietitian (55%) as key providers in their children's care networks while 45% identified a pediatrician as a key provider. There were a variety of characteristics that parents we interviewed reported as criteria they used to designate someone a key provider, including disease expertise, availability, coordination/central role in network and provider demeanour. Many interviewed parents highlighted the importance of providers who trust the parents/family to know their child's needs and those who in turn can be trusted by families.

Many parents identified better care coordination as a needed improvement in their children's care networks. Having a coordinated network would mean that the family would not have to constantly re-tell their child's IMD story or relay information between different providers. Additionally, parents underscored the importance of relational continuity for their children's care, expanding that relational continuity goes beyond having the same provider over an extended period of time and entails having a provider that knows their child and family well, is familiar with the kind of services the child is receiving, and knows up-to date information about the medical history. Care coordination and relational continuity are connected: in order to achieve the experience of relational continuity described by parents, a network needs to have robust care coordination that enables providers to stay connected and updated on important information relating to the needs of the child.

Care coordination is an essential component to the delivery of high-quality family-centred health care for children with complex medical needs. Parents in this study described feeling an intense degree of responsibility to establish, adapt and monitor their children's networks of care to meet social and medical needs. This role was assumed by parents to compensate for gaps in family-centred care, including care coordination, in these care networks. Parents worked as unofficial managers and care coordinators in their children's care networks. A few studies examining the role of parents in the care of children with medical complexity have similarly found that parents assumed responsibility for their children's care coordination due to a desire to secure the necessary services to meet their child's needs.[40–42]

Despite the critical role that parents play in the management of care networks, we found that they often were not able to access information and resources they needed for this work, as they were not formally part of the health care team. For example, parents often cannot directly

access care without a referral and may not be included on communications between providers. Parents frequently described adopting an advocacy role and leveraging resources available to them to improve access to care for their child. One strategy parents commonly reported was identifying at least one provider who trusted the parents' knowledge of their child's needs and was available in a timely way, to act as their "go-to" provider in times of need for responses or assistance relating to their child's IMD. Family-centred care relies on the partnership and collaboration of families and patients with health care providers in decision-making.[43] Parents having to resort to these strategies to ensure that they have the best care for their child points to a lack of implementation of patient-centred care at a system level to support such partnerships and collaboration. Parents of children with medical complexity have reported that while they expect a collegial relationship with health care providers, providers often fail to acknowledge their expertise.[42] The responsibility of care coordination is shifted to the parents without an acknowledgement of their role, and access to the necessary resources and information to facilitate the management of these care networks.

Parents expressed relief when care was coordinated properly, and they were able to be in the role of recipients of care as opposed to coordinators of care. A high proportion of participants who identified a metabolic doctor, dietitian, or pediatrician as a key provider indicated that at least one of these provider types in their child's network knew their child and family very well or fairly well and rated at least one of those provider types as coordinating fairly well or very well. The high proportion of participants who identified and rated dietitians and metabolic doctors well on family-centred care elements is consistent with previous work with families of children with IMDs that has shown that caregivers tend to consider care received in the metabolic clinic to be family-centred.[6,44] This implies that there are some elements of the metabolic clinic that correspond to the family-centred care concept of a 'medical home' for children with IMDs. However, designating

the metabolic clinic as a medical home has recognized limitations as it might not be suitable for coordinating care in the community and is often less accessible relative to primary care settings, particularly for families residing outside of major cities where metabolic clinics are located.[44] A co-management model, where the care primarily resides with the metabolic clinic and partnerships are established with primary care providers, may enable the implementation of the medical home concept,[45,46] and could be explored. This corresponds with one of the key suggestions of parents we interviewed, that is, to increase the capacity of primary care providers to be involved in their children's care.

Strengths and Limitations

This study has important strengths. To the authors' best knowledge, this is one of the few studies to use social network analysis to study the care networks of children with medical complexities. By using quantitative and qualitative methods, we were able to gain a nuanced understanding of the care networks of children with IMDs. Not only were we able to understand the structures of these networks, including the types of services and providers in these networks, the connections between the different providers, and the overall cohesiveness of these networks but we also gained insight into how these networks are experienced by parents and families and what parents want out of their interactions and experiences in these care networks. In terms of data collection, our study relied on network visualization methods for data collection (the parent-drawn care maps). Using the network visualization method, as opposed to for example, name-generator lists, has important benefits in social network analysis: i) this process is efficient and intuitive for participants; ii) visual representation during data collection provided our team with opportunities for follow-up questions on interesting patterns of connection in network (i.e., care map interviews);

and iii) using visual representation is a practical option for obtaining data on the structure of large egocentric networks without sampling a subset of alters (i.e., care providers in this study).[27]

This study also has several limitations. First, to be eligible to participate in the ongoing cohort study, participants had to be proficient in English. This requirement could potentially limit the generalizability of the study findings as it will exclude a potentially more vulnerable population of children and families, who require access to translators or other additional supports. Also related to participant selection, during the ongoing enrollment of participants, we prioritized the selection of participants who expected that their child would have at least one health care encounter per month, to accommodate the needs of the prospective cohort study. As a result, the sample was likely over-representative of parents of children with IMDs who were more frequent health care users and, correspondingly, those with larger care networks. Finally, the care map data reflected parental perceptions about their children's health care and about connections between providers. These parental perceptions might be different from provider perceptions, for example if providers were sharing information to coordinate care but the parent was unaware of that sharing. However, our findings were intended to reflect the ways in which parents experienced health care and coordination of care across children's health care networks.

Conclusion and Future Directions

By using care maps and qualitative interviews, we were able to better understand the complexity of health care for children with IMDs and learn about them from a network and provider level, as experienced and reported by parents. Our findings point to care networks of varying size with a sparsity in coordination, leading parents to assume responsibility for many aspects of care, including care coordination. Providers such as dietitians and metabolic doctors were common across children's care networks and may be well-positioned to collaborate with

primary care providers to support the implementation of co-management family-centred care models.

References

1. Staniszewska S, Brett J, Simera I, et al. GRIPP2 reporting checklists: Tools to improve reporting of patient and public involvement in research. *BMJ*. 2017;358. doi:10.1136/bmj.j3453
2. Ferreira CR, Karnebeek CDM Van, Vockley J, Blau N, Centre AM. A proposed nosology of inborn errors of metabolism. *Genet Med*. 2019;21(1):102-106. doi:10.1038/s41436-018-0022-8.A
3. Pampols T. Inherited Metabolic Rare Disease. In: *Advances in Experimental Medicine and Biology*. ; 2010:397-431. doi:10.1007/978-90-481-9485-8_23
4. Ellaway C, Wilcken B, Christodoulou J. Clinical approach to inborn errors of metabolism presenting in the newborn period. *J Paediatr Child Health*. 2002;38(5):511-517. doi:10.1046/j.1440-1754.2002.00047.x
5. Khangura SD, Tingley K, Chakraborty P, et al. Child and family experiences with inborn errors of metabolism: a qualitative interview study with representatives of patient groups. *J Inherit Metab Dis*. 2016;39(1):139-147. doi:10.1007/s10545-015-9881-1
6. Siddiq S, Wilson BJ, Graham ID, et al. Experiences of caregivers of children with inherited metabolic diseases: a qualitative study. *Orphanet J Rare Dis*. 2016;11(1):168. doi:10.1186/s13023-016-0548-2
7. Baird J, Rehm RS, Hinds PS, Baggott C, Davies B. Do You Know My Child? Continuity of Nursing Care in the Pediatric Intensive Care Unit. *Nurs Res*. 2016;65(2):142-150. doi:10.1097/NNR.0000000000000135
8. Miller AR, Condin CJ, McKellin WH, Shaw N, Klassen AF, Sheps S. Continuity of care for children with complex chronic health conditions: parents' perspectives. *BMC Health Serv Res*. 2009;9(1):242. doi:10.1186/1472-6963-9-242
9. Carter B, Cummings J, Cooper L. An exploration of best practice in multi-agency working and the experiences of families of children with complex health needs. What works well and what needs to be done to improve practice for the future? *J Clin Nurs*. 2007;16(3):527-539. doi:10.1111/j.1365-2702.2006.01554.x
10. Espezel HJE, Canam CJ. Parent-nurse interactions: care of hospitalized children. *J Adv Nurs*. 2003;44(1):34-41. doi:10.1046/j.1365-2648.2003.02765.x
11. Fisher MJ, Broome ME. Parent–Provider Communication During Hospitalization. *J Pediatr Nurs*. 2011;26(1):58-69. doi:10.1016/j.pedn.2009.12.071
12. Turchi RM, Antonelli RC, Norwood KW, et al. Patient- and Family-Centered Care Coordination: A Framework for Integrating Care for Children and Youth Across Multiple Systems. *Pediatrics*. 2014;133(5):e1451-e1460. doi:10.1542/peds.2014-0318
13. Berwick DM, Nolan TW, Whittington J. The Triple Aim : Care , Health , And Cost Reproduced with permission of the copyright owner . Further reproduction prohibited without permission . *Health Aff*. 2008;27(3):759-769.
14. Adams S, Nicholas D, Mahant S, et al. Care maps for children with medical complexity. *Dev Med Child Neurol*. 2017;59(12):1299-1306. doi:10.1111/dmcn.13576
15. Stille C, Turchi RM, Antonelli R, et al. The Family-Centered Medical Home: Specific Considerations for Child Health Research and Policy. *Acad Pediatr*. 2010;10(4):211-217. doi:10.1016/j.acap.2010.05.002
16. Fetters MD, Curry LA, Creswell JW. Achieving Integration in Mixed Methods Designs-Principles and Practices. *Health Serv Res*. 2013;48(6pt2):2134-2156. doi:10.1111/1475-

6773.12117
17. Robinson O. Sampling in Interview-Based Qualitative Research: A Theoretical and Practical Guide. *Qual Res Psychol*. 2014;11(1):25-41. https://search.ebscohost.com/login.aspx?direct=true&db=a9h&AN=92017423&site=ehost-live
18. Chow AJ, Iverson R, Lamoureux M, et al. Families' healthcare experiences for children with inherited metabolic diseases: protocol for a mixed methods cohort study. *BMJ Open*. 2022;12(2):e055664. doi:10.1136/bmjopen-2021-055664
19. Sibley LM, Weiner JP. An evaluation of access to health care services along the rural-urban continuum in Canada. *BMC Health Serv Res*. 2011;11. doi:10.1186/1472-6963-11-20
20. Rajasekar P, Gannavarapu S, Napier M, et al. Parental psychosocial aspects and stressors involved in the management of inborn errors of metabolism. *Mol Genet Metab Reports*. 2020;25(October). doi:10.1016/j.ymgmr.2020.100654
21. Chella Krishnan K, Mehrabian M, Lusis AJ. Sex differences in metabolism and cardiometabolic disorders. *Curr Opin Lipidol*. 2018;29(5):404-410. doi:10.1097/MOL.0000000000000536
22. Clegg DJ, Mauvais-Jarvis F. An integrated view of sex differences in metabolic physiology and disease. *Mol Metab*. 2018;15:1-2. doi:10.1016/j.molmet.2018.06.011
23. Antonelli RC, Lind C. Care Mapping : A How-To Guide for Patients and Families Available: http://www.childrenshospital.org/-/media/ Care-Coordination/CareMappingforfamilies21813.ashx?la=en&hash=D8C02FCA893C9A29C939613532334E07127BF9E6. [Accessed May 10, 2021]
24. Harris PA, Taylor R, Thielke R, Payne J, Gonzalez N, Conde JG. Research electronic data capture (REDCap)-A metadata-driven methodology and workflow process for providing translational research informatics support. *J Biomed Inform*. Published online 2009. doi:10.1016/j.jbi.2008.08.010
25. Harris PA, Taylor R, Minor BL, et al. The REDCap consortium: Building an international community of software platform partners. *J Biomed Inform*. Published online 2019. doi:10.1016/j.jbi.2019.103208
26. Chung KKS, Hossain L, Davis J. Exploring sociocentric and egocentric approaches for social network analysis. *Proc 2nd …*. 2005;(September 2015):1-8. http://www.it.usyd.edu.au/~info4990/2006/papers/HossainKMAP.pdf
27. Perry BL, Pescosolido BA, Borgatti SP. *Egocentric Network Analysis: Foundations, Methods, and Models*. Cambridge: Cambridge University Press; 2018. doi:10.1017/9781316443255
28. Borgatti SP, Everett MG. and Freeman LC. Ucinet for Windows: Software for Social Network Analysis.e. Published online 2002.
29. Francis JJ, Johnston M, Robertson C, et al. What is an adequate sample size? Operationalising data saturation for theory-based interview studies. *Psychol Health*. 2010;25(10):1229-1245. doi:10.1080/08870440903194015
30. Guest G, Bunce A, Johnson L. How Many Interviews Are Enough? *Field methods*. 2006;18(1):59-82. doi:10.1177/1525822X05279903
31. Hennink MM, Kaiser BN, Marconi VC. Code Saturation Versus Meaning Saturation. *Qual Health Res*. 2017;27(4):591-608. doi:10.1177/1049732316665344

32. Jarrett N, Porter K, Davis C, et al. The networks of care surrounding cancer palliative care patients. *BMJ Support Palliat Care*. 2015;5(4):435-442. doi:10.1136/bmjspcare-2014-000782
33. Cheong LHM, Armour CL, Bosnic-Anticevich SZ. Primary health care teams and the patient perspective: A social network analysis. *Res Soc Adm Pharm*. 2013;9(6):741-757. doi:10.1016/j.sapharm.2012.12.003
34. Thorne S, Kirkham SR, O'Flynn-Magee K. The Analytic Challenge in Interpretive Description. *Int J Qual Methods*. 2004;3(1):1-11. doi:10.1177/160940690400300101
35. Braun V, Clarke V. Using thematic analysis in psychology. *Qual Res Psychol*. 2006;3(2):77-101. doi:10.1191/1478088706qp063oa
36. Lincoln Y, Guba EG. *Naturalistic Inquiry*. SAGE Publications; 1985.
37. Cohen E, Berry JG, Camacho X, Anderson G, Wodchis W, Guttmann A. Patterns and Costs of Health Care Use of Children With Medical Complexity. *Pediatrics*. 2012;130(6):e1463-e1470. doi:10.1542/peds.2012-0175
38. Adams S, Nicholas D, Mahant S, et al. Care maps and care plans for children with medical complexity. *Child Care Health Dev*. 2019;45(1):104-110. doi:10.1111/cch.12632
39. Dewan T, Cohen E. Children with medical complexity in Canada. *Paediatr Child Health*. 2013;18(10):518-522. doi:10.1093/pch/18.10.518
40. Cady R, Bushaw A, Davis H, Mills J, Thomasson D. Care coordination for children with medical complexity. *Nurse Pract*. 2020;45(6):11-17. doi:10.1097/01.NPR.0000666172.10978.4f
41. Quigley L, Lacombe-Duncan A, Adams S, Moore Hepburn C, Cohen E. A qualitative analysis of information sharing for children with medical complexity within and across health care organizations. *BMC Health Serv Res*. 2014;14(1). doi:10.1186/1472-6963-14-283
42. Smith J, Cheater F, Bekker H. Parents' experiences of living with a child with a long-term condition: A rapid structured review of the literature. *Heal Expect*. 2015;18(4):452-474. doi:10.1111/hex.12040
43. Kuo DZ, Houtrow AJ, Arango P, Kuhlthau KA, Simmons JM, Neff JM. Family-Centered Care: Current Applications and Future Directions in Pediatric Health Care. *Matern Child Health J*. 2012;16(2):297-305. doi:10.1007/s10995-011-0751-7
44. Chow AJ, Pugliese M, Tessier LA, et al. Family Experiences with Care for Children with Inherited Metabolic Diseases in Canada: A Cross-Sectional Survey. *Patient*. 2022;15(2):171-185. doi:10.1007/s40271-021-00538-8
45. Cohen E, Lacombe-Duncan A, Spalding K, et al. Integrated complex care coordination for children with medical complexity: A mixed-methods evaluation of tertiary care-community collaboration. *BMC Health Serv Res*. 2012;12(1). doi:10.1186/1472-6963-12-366
46. Gordon JB, Colby HH, Bartelt T, Jablonski D, Krauthoefer ML, Havens P. A tertiary care-primary care partnership model for medically complex and fragile children and youth with special health care needs. *Arch Pediatr Adolesc Med*. 2007;161(10):937-944. doi:10.1001/archpedi.161.10.937

Chapter 3 – General Discussion

This thesis project is embedded within a broader research program that aims to develop interventions tailored to the needs of children with IMDs that address challenges in family-centred care. The overall aim of this thesis was to gain a thorough understanding of parents' perceptions of the structure of care networks for young children with IMDs and how those care networks are experienced.

We used an explanatory sequential mixed-methods design (QUANT → QUAL). For the quantitative component, we conducted a secondary analysis that relied on a care map dataset from a prospective cohort study where participants were parents or legal guardians ("parents") of children 12 years of age or younger diagnosed with an IMD. Specifically, we analyzed these care maps using social network analysis to determine network characteristics such as size, cohesiveness, and centralization of these networks and at the provider type level, we reported on degree-centrality (number of connections that a provider was perceived to have within a care network) and share (percentage of connections in a network that involve that provider). We also descriptively reported on the frequency with which each provider type was included in the care maps and the frequency of identification of each provider type as a key health care provider. Finally, we analyzed care map questionnaires to determine, for each of the most common key health care providers, parental perceptions of relational continuity and care coordination. For the qualitative component, we purposively selected a subsample of participants to participate in a semi-structured interview to elaborate on their child's care maps. Through the care map interviews, we were able to better understand: (i) how parents experienced the process of drawing the care map, (ii) how they selected providers to include on the care map, (iii) characteristics of a 'key' health care provider and the role they play in the child's IMD care, (iv) the nature of the

connections between providers and the impact of those connections on the family, and (v) the adequacy of the care network in meeting the child and family needs, including parental perceptions of the need for improvements in the care network. Finally, the mixed-methods integration occurred at different stages at the level of the design, methods, and interpretation and reporting.

Summary of Findings

Participant, Child, and Household Characteristics

Our study sample consisted of 60 parents or stepparents of children with IMDs, and most (88%) were mothers. A majority of parents participants in our sample were university graduates with total household incomes of $100,000 CAD or higher. Close to half of the children with IMDs enrolled in the study had an IMD in Group 2 (acute and episodic disease).

Parent Perceptions of the Characteristics of Children's Care Networks

Quantitative findings. Our findings highlighted that care networks for children with IMDs are large with a median network size of 14.5 (interquartile range [IQR] 10-20). We found variation among IMDs, with older children and those with diseases that were multi-system and progressive in nature having the largest care networks. Further, these networks were heterogeneous in terms of providers and settings. Relating to the cohesiveness of networks of children with IMDs, we reported a median density of 0.08 (IQR 0.05-0.11), meaning that approximately 8% of all possible pairwise connections between providers were identified as observed connections by parents. Parents reported observing a higher proportion of connections among providers who could be connected in the care networks of children with diagnoses in IMD Groups 1 (median density 0.10) and 2 (median density 0.09) relative to Group 3 (median density 0.05). Finally, we also reported on the median centralization for children's care networks, which, based on parent

perceptions of connections among providers, was 0.23 (IQR 0.12-0. 31). Parents reported more centralized care networks for children who lived more than three hours (median centralization 0.30) from a metabolic centre relative to children whose travel time was an hour or less (median centralization 0.28) or one to three hours (median centralization 0.16).

We also used post-hoc visualization of the distributions of network size, density and centralization and discussion among the team to identify three care map archetypes: 1) small networks (11 or fewer providers based on an obvious split in the distribution of network size in the sample); 2) large but sparse networks (>11 providers and lower or equal to the median for either density or centralization or both among large care maps); and 3) large *relatively* connected networks (>11 providers and higher than the median for both density and centralization among large care maps).

Qualitative findings. Parents often described some level of predictability in terms of a schedule of follow-up appointments with regular providers such as a metabolic physician, specialist, or pediatrician. However, when discussing these care routines and frequency of interactions with different providers, parents also frequently added that their child's IMD care is dynamic, and they need to constantly adapt to their child's changing needs and health system changes. A key finding was that parents often perceived themselves to be in an intensive 'manager' role, responsible for establishing and maintaining their children's care networks and for compensating for the lack of formal care coordination.

Key care providers and relational continuity

Quantitative findings. The majority of participants identified a metabolic doctor (n=40, 78%) and a dietitian (n=33, 55%) as key providers. Most parents (80%) reported having at least one healthcare provider in the network that knew their child and family fairly well or very well. A

high proportion of participants who identified a dietitian (94%) or a metabolic doctor (75%) as a key provider indicated that at least one of these provider types knew their child and family very well or fairly well.

Qualitative findings. Parents identified several provider characteristics that were important to their designation as a key provider, such as expertise in an area of care impactful to the child's daily life or disease progression, availability, central coordination role, and affect. Across providers involved in the care of children with IMDs, parents highlighted trust as an important requirement for developing positive relationships with their child's care providers. This included valuing providers who trusted the parents and family to know their child's needs, and providers who could be trusted to provide appropriate care.

Providers as part of a dynamic network

Quantitative findings. Our findings also highlighted parent-perceived connections between providers and parent perceptions of care coordination. The highest median degree centrality across all provider types was three, meaning that on average, parents did not perceive any provider type to have more than 3 connections to other providers in the network. A high proportion of parents who identified a metabolic doctor (85%) or dietitian (91%) as a key provider rated at least one of these provider types in their child's network as coordinating fairly well or very well.

Qualitative findings. Parents viewed and considered care coordination to entail providers actively working as a team to organize care; referrals or consultation; or providers sharing information with one another. One strategy parents commonly reported was identifying at least one provider who trusted the parents' knowledge of their child's needs and was available in a timely way, to act as their "go-to" provider in times of need for care and access to services.

Integration of Findings and Recommendations

Parents of children with IMDs described health care networks that were large and heterogeneous. While there was variation across networks in the numbers and types of connections between providers, parents generally did not perceive that providers across the network were well connected; this was particularly true for non-metabolic specialists, including primary care providers. Our findings echo the growing body of research about the challenges parents face navigating services for children with complex health needs which often point to a lack of formal care coordination and communication between health care providers as areas of concern.[1–4]

Rather than solely being recipients of care, parents tended to act as 'managers' of their children's networks and felt responsible for establishing, adapting, and organizing their child's care network to fill unmet care coordination needs. Particularly, parents spent considerable effort in relaying information between providers. The parent 'manager' role was sometimes reported to be overwhelming. Similarly, other studies report that caregivers often felt a need to fill in as care coordinators to ensure that their child's care needs were met[1,4–6]; this added stressful demands on caregivers.[1,4,6] Despite the critical role that parents play in the management of care networks, we found that they often experienced challenges in facilitating this role, such as not being able to access information and resources they needed for this work, as they were not formally part of the health care team. Other studies have similarly noted that a lack of electronic and informational tools made it difficult for parents to facilitate care coordination.[7]

In our study, parents frequently described adopting an advocacy role and leveraging resources available to them to improve access to care for their child. One strategy parents used to fill gaps in care coordination was to identify a "go-to person": a provider who they felt would trust them as parents and who could either directly respond to the child's needs or provide access to

other providers who could respond. Parents having to resort to these strategies to ensure that they have the best care for their child points to a lack of implementation of family-centred care coordination at a system level that supports partnerships and collaboration between parents and providers.[8] Parents of children with medical complexity have reported that while they expect a collegial relationship with health care providers, providers often fail to acknowledge their expertise.[9] Our study, in line with previous work,[1,4] highlights that the responsibility of care coordination is often shifted to the parents without an acknowledgement of their role, and access to the necessary resources and information to facilitate the management of their children's care networks.

The lack of formal inclusion of parents contradicts the "team-based" care coordination conceptualized by Antonelli et al in 2009[10] and reiterated in the policy statement by the American Academy of Pediatrics[11], which suggests that that there multiple people required to support care coordination, including parents and/or caregivers, primary care providers, a designated care coordinator, specialists, and others to meet the individual needs of each child and family. A care coordination process that can include these aforementioned providers and allows for the negotiating of goals and priorities between caregivers and health care providers may help address some of the concerns that caregivers face with regards to care coordination. It has been recommended that members in these multidisciplinary and family-inclusive teams should have specific, defined roles and dedicated time and effort specific to care planning.[12]

Some of the strategies that have been proposed to help facilitate coordinated care includes care mapping[13–15], care plans[15–17] and electronic health records and patient portals.[18] Care mapping can be a useful approach to actively engage families and highlight the 'big picture' about the lived experiences, resources and needs of each family.[19] In our study, parents found that the care map

was a useful visual representation of the numerous providers and services involved in their child's care. It may also serve as an important tool to highlight the central role of parents in care coordination as well as to identify important gaps in communication between providers. For children with medical complexity, care maps can be used in clinical care, alongside care plans.[14,15] A comprehensive care plan created by health care providers and families, which contains medical information, an updated list of medications, care providers, appointments, and goals of care has been advocated as a tool to help support care coordination efforts while also enhancing the reciprocal exchange of information and strengthening relationships between parents and providers.[16,17] Parents in our study emphasized that care for their children was in 'flux' and required constant adaptation. Thus, implementation of care maps and care plans would require families and health care providers across organizations to collaboratively manage and update these documents over time.[6] An electronic health record can support the process of sharing and updating care maps and care plans and also enable the sharing of other health information with providers. Some parents noted that they valued such a system because there was less risk that a provider would be excluded from receiving important information or that the responsibility would fall to the parent to share information between providers. At the same time, when providers were sharing information with each other through electronic health records, there was a risk that the family would be left out of the communication and may thus feel external to the health care team. To ensure that "team-based" care coordination is implemented, patient portals have been recommended so that parents have access to the necessary information and continue to be updated similar to all other members in the network of care.[18,20]

Care maps, care plans, and the sharing of medical information on electronic health records and patient portals are all approaches that have been proposed and/or demonstrated to support an

environment where providers across different settings and families collaborate to meet the changing needs of children. Aside from their utility in enabling care coordination, these tools may aid in actualizing relational continuity with providers and building mutual trust, which parents in our study underscored as important for their children's care. Parents explained that relational continuity goes beyond having the same provider over an extended period of time and entails having a provider that knows their child and family well, is familiar with the kind of services the child is receiving and knows up-to date information about the child's medical history. With respect to mutual trust, parents described it in terms of providers who saw "the whole child", those whose knowledge and expertise were trusted by the family, and those who reliably followed through on commitments. Care plans have the potential to foster relationships between providers and families and allow for more reciprocal information sharing,[15,16] and in some studies, parents of children with medical complexity perceived the integration of care maps into clinical practice as a way to have their voices heard and ensure that their preferences and needs were addressed.[14,15] Thus, these tools do have the potential to facilitate providers' understanding of families' needs and perspectives and thus promote better patient and family-centred partnerships.[14,21]

However, while care maps, care plans and electronic health records have been shown to be important tools to support care coordination, a 'medical home' may still be important for children with IMDs given its emphasis in recommendations for improving family-centred care. Our findings show that a high proportion of participants who identified a metabolic doctor, or dietitian as a key provider in their child's care network indicated that at least one of these provider types in their child's network knew their child and family very well or fairly well and rated at least one of those provider types as coordinating fairly well or very well. This finding is consistent with previous studies of families of children with IMDs that have shown that caregivers tend to consider

care received in the metabolic clinic to be family-centred.[22,23] As discussed in chapter 2, this implies that there are some elements of the metabolic clinic that correspond to the family-centred care concept of a 'medical home' for children with IMDs.

However, designating the metabolic clinic as a medical home has recognized limitations relating to coordinating care in the community and accessibility, particularly for families residing outside of major cities were metabolic clinics are located.[22] Instead, a co-management model, where the care primarily resides with the metabolic clinic and partnerships are established with primary care providers, may enable the implementation of the medical home concept for children with IMDs.[24,25] Emerging evidence shows that primary care physicians, especially pediatricians, are comfortable with a co-management model.[26] A randomized controlled trial examining the role of a care coordinator in supporting the efforts of primary care physicians to provide comprehensive care for children with special health care needs through the medical home found that it improved parental satisfaction with health care services. In particular, the study pointed to increased number of children with individualized care plans, increased parental satisfaction with care coordination and high provider acceptance of the program.[27] Additional work has similarly pointed to improved family-centred outcomes with a co-management model.[24,25] To address the priorities of parents in our study, a co-management model would need to be inclusive of family caregivers as core to the care team. However, processes and systems (e.g., electronic health records, care plans) to ensure communication and coordination between providers are critically important to alleviate the need for families to fill in the care coordination gaps.[1] This corresponds with the key suggestions of parents we interviewed, that is, to increase the capacity of primary care providers to be involved in their children's care and improve care coordination efforts so that families are not required to fill in the gaps as unpaid 'managers'.

Implications for future research

This study is part of a bigger research program that aims to co-develop, with families of children with IMDs and health care providers, interventions that are tailored to support family-centred care for children and families with IMDs. Parents of children with IMDs with high socioeconomic status, as reflected by the high reported education levels and household income, comprised a majority of the study sample. It is important to situate this work with a reflection of the impact of the socioeconomic status of those participants on the nature of their experiences as it might underscore some of the further challenges that those with fewer resources face while navigating the healthcare system; especially given that we found that parents tend to play a crucial role in managing and coordinating their children's health care networks without formal recognition and resources. The parental responsibility to establish, maintain, coordinate and manage their children's care network is concerning for a number of reasons but one of the most important is that it likely contributes to inequitable access to family-centred care. Parents who have resources such as time, social capital, medical knowledge, high levels of English communication skills and who haven't historically faced systemic barriers to being able to access services and supports are likely much better positioned to advocate, navigate systems, and negotiate, including standing up to providers and systems or being persistent about what they feel their child needs. This would make it easier for them to ensure that their children's care needs are met. Given that more parents in our study may have these aforementioned capabilities, our findings could be pointing to a 'best case scenario' in terms of navigating healthcare networks and the strategies used by the parents in the study to compensate for gaps in care may be less available to parents who face systemic barriers to care. This underscores the importance for future research that specifically studies equity-deserving populations to better understand their experiences with care for their children with

IMDs. Further, it also emphasizes that solutions to receiving high-quality care that are based on parents having a strong advocacy role are unlikely to be equitable. Future research could aim to further our understanding of parent preferences regarding the nature of their role and identify strategies for the supported integration of parents in these networks in a way that alleviates rather than exacerbates the stress of the responsibility they currently feel. It is also important for future research to incorporate provider perspectives to clearly understand what may be necessary to help support their efforts in care coordination and collaboration with family caregivers within the context of a co-management model.

This study contributes to the methodological literature regarding the study of health care networks. In particular, by using a mixed-methods design employing a network visualization method (i.e., parent drawn care maps) and qualitative interviews, we were able to learn about networks from four dimensions: i) structure, which refers to the presence and patterns of linkages between actors (achieved understanding of structure through the care maps); function, which refers to types of exchanges, services, or supports accessible through ties to alters (achieved understanding of function through care maps and interviews); strength, which captures intensity and duration of bonds between an ego and alters within the network (achieved understanding of strength through care map interviews); and content, which represents flow from or to each person in a network (achieved understand of content through care map interviews).[29] Essentially, the use of mixed methods to study the networks allowed us to gain an 'outsider' view of the structure of the network and an 'insider' perception of the network.[28] While there are calls to employ mixed methods designs in social network analysis, there are sparse examples of such studies. Future studies could expand on these methods by collecting data from provider perspectives to integrate into the care maps.

Strengths and Limitations

This study has important strengths. To the authors' best knowledge, this is one of the few studies to use social network analysis to study the care networks of children with medical complexities. By using mixed methods, we were able to gain a nuanced understanding of the structures of these care networks of children with IMDs and how these networks are experienced by parents and families. This study used the network visualization method to produce the care map, as opposed to for example, name-generator lists, which has important benefits in social network analysis: i) this process is efficient and intuitive for participants; ii) visual representation during data collection provided our team opportunities for follow-up questions on interesting patterns of connection in network (i.e., care map interviews); and iii) using visual representation is a practical option for obtaining data on the structure of large egocentric networks without sampling a subset of alters (i.e., care providers in this study).[29] To ensure the accuracy of care maps, the cohort study team had multiple rounds of communication with the study participants. During data entry of care maps, ZA and a second team member employed a double-data entry process to ensure accuracy of entered data. To ensure qualitative rigor, we implemented several processes during different phases of the study that follow the criteria of credibility, transferability, dependability and confirmability.[30] Specifically, we employed verbatim transcription as a strategy to ensure rigor; we engaged with the study data for a prolonged period of time and reviewed the data and analysis with experts in qualitative research; we created thick descriptions of the themes; and we maintained an audit trail by keeping records of the transcripts, and notes. We also operationalized reflexivity throughout the research process. To do so, ZA used memoing and reflection. Immediately following each interview, ZA wrote memos about any part of the interview that invoked opinions or thoughts based on her personal and professional experiences. Finally, our

study engaged patient partners at all stages of the study and we will be using reporting guidelines (specifically GRIPP2)[31] to document patient partner involvement.

This study also has several limitations. First, the requirement that study participants speak English may limit the generalizability of the study findings as it excluded a potentially more vulnerable population of children and families, who require access to translators or other additional supports. Also related to participant selection, this study may be affected by selection bias as during the ongoing enrollment of participants, we prioritized the selection of participants who expected that their child would have at least one health care encounter per month, to accommodate the needs of the prospective cohort study. As a result, the sample was likely over-representative of parents of children with IMDs who were more frequent health care users and, correspondingly, those with larger care networks, potentially impacting the generalizability of the quantitative results. Finally, the care map data reflected parental perceptions about their children's health care and about connections between providers. These parental perceptions might be different from provider perceptions, for example if providers were sharing information to coordinate care but the parent was unaware of that sharing. However, our findings were intended to reflect the ways in which parents' experiences health care and coordination of care across children's health care networks.

Conclusions

A mixed-methods study design allowed us to develop a thorough understanding of the complexity of health care for children with IMDs and learn about care networks, as experienced and reported by parents. Our findings point to care networks of varying size with a sparsity in coordination, leading parents to assume responsibility for many aspects of care, including care coordination. Integrating care maps and care plans within electronic health records and patient

portals may allow parents' roles to be better recognized and to foster partnerships with providers. Simultaneously, providers such as dietitians and metabolic doctors, who were common across children's care networks, may be well-positioned to collaborate with primary care providers using tools such as care maps, care plans and electronic health records to support the implementation of co-management family-centred care models.

References

1. Golden SL, Nageswaran S. Caregiver voices: Coordinating care for children with complex chronic conditions. *Clin Pediatr (Phila)*. 2012;51(8):723-729. doi:10.1177/0009922812445920
2. Bourke-Taylor H, Howie L, Law M. Impact of caring for a school-aged child with a disability: understanding mothers' perspectives. *Aust Occup Ther J*. 2010;57(2):127-136. doi:10.1111/j.1440-1630.2009.00817.x
3. Page BF, Hinton L, Harrop E, Vincent C. The challenges of caring for children who require complex medical care at home: 'The go between for everyone is the parent and as the parent that's an awful lot of responsibility.' *Heal Expect*. 2020;23(5):1144-1154. doi:10.1111/hex.13092
4. Cady RG, Belew JL. Parent perspective on care coordination services for their child with medical complexity. *Children*. 2017;4(6):1-14. doi:10.3390/children4060045
5. Stille CJ, Primack WA, McLaughlin TJ, Wasserman RC. Parents as information intermediaries between primary care and specialty physicians. *Pediatrics*. 2007;120(6):1238-1246. doi:10.1542/peds.2007-1112
6. Quigley L, Lacombe-Duncan A, Adams S, Moore Hepburn C, Cohen E. A qualitative analysis of information sharing for children with medical complexity within and across health care organizations. *BMC Health Serv Res*. 2014;14(1). doi:10.1186/1472-6963-14-283
7. Horsky J, Morgan SJ, Ramelson HZ. Coordination of care for complex pediatric patients: perspectives from providers and parents. *AMIA Annu Symp Proc*. 2014;2014:681-690.
8. Kuo DZ, Houtrow AJ, Arango P, Kuhlthau KA, Simmons JM, Neff JM. Family-Centered Care: Current Applications and Future Directions in Pediatric Health Care. *Matern Child Health J*. 2012;16(2):297-305. doi:10.1007/s10995-011-0751-7
9. Smith J, Cheater F, Bekker H. Parents' experiences of living with a child with a long-term condition: A rapid structured review of the literature. *Heal Expect*. 2015;18(4):452-474. doi:10.1111/hex.12040
10. Antonelli, R, McAllister, J, Popp J. Making Care Coordination a Critical Part of the Pediatric Health System. 2009;(1277). http://www.commonwealthfund.org/publications/fund-reports/2009/may/making-care-coordination-a-critical-component-of-the-pediatric-health-system
11. Turchi RM, Antonelli RC, Norwood KW, et al. Patient- and Family-Centered Care Coordination: A Framework for Integrating Care for Children and Youth Across Multiple Systems. *Pediatrics*. 2014;133(5):e1451-e1460. doi:10.1542/peds.2014-0318
12. Stille C, Turchi RM, Antonelli R, et al. The Family-Centered Medical Home: Specific Considerations for Child Health Research and Policy. *Acad Pediatr*. 2010;10(4):211-217. doi:10.1016/j.acap.2010.05.002
13. Cady R, Bushaw A, Davis H, Mills J, Thomasson D. Care coordination for children with medical complexity. *Nurse Pract*. 2020;45(6):11-17. doi:10.1097/01.NPR.0000666172.10978.4f
14. Adams S, Nicholas D, Mahant S, et al. Care maps for children with medical complexity. *Dev Med Child Neurol*. 2017;59(12):1299-1306. doi:10.1111/dmcn.13576
15. Adams S, Nicholas D, Mahant S, et al. Care maps and care plans for children with medical complexity. *Child Care Health Dev*. 2019;45(1):104-110. doi:10.1111/cch.12632

16. Adams S, Cohen E, Mahant S, Friedman JN, MacCulloch R, Nicholas DB. Exploring the usefulness of comprehensive care plans for children with medical complexity (CMC): a qualitative study. *BMC Pediatr*. 2013;13(1):10. doi:10.1186/1471-2431-13-10
17. Cady R, Bushaw A, Davis H, Mills J, Thomasson D. Care coordination for children with medical complexity: Whose Care is it Anyways? *Nurse Pract*. 2020;45(6):11-17. doi:10.1097/01.NPR.0000666172.10978.4f
18. Kuo DZ, McAllister JW, Rossignol L, Turchi RM, Stille CJ. Care Coordination for Children With Medical Complexity: Whose Care Is It, Anyway? *Pediatrics*. 2018;141(Supplement_3):S224-S232. doi:10.1542/peds.2017-1284G
19. Antonelli RC, Lind C. Care Mapping : A How-To Guide for Patients and Families Available: http://www.childrenshospital.org/-/media/ Care-Coordination/CareMappingforfamilies21813.ashx?la= en&hash=D8C02FCA893C9A29C939613532334E07127BF9E6. [Accessed May 10, 2021]
20. Berry J, Agrawal R. The landscape of medical care for children with medical complexity. *Child Hosp …*. 2013;(June):1-16. http://www.columbia.edu/itc/hs/medical/residency/peds/new_compeds_site/pdfs_new/PL3 new readings/Special_Report_The_Landscape_of_Medical_Care_for_Children_with_Medical _Complexity.pdf
21. Batalden M, Batalden P, Margolis P, et al. Coproduction of healthcare service. *BMJ Qual Saf*. 2016;25(7):509-517. doi:10.1136/bmjqs-2015-004315
22. Chow AJ, Pugliese M, Tessier LA, et al. Family Experiences with Care for Children with Inherited Metabolic Diseases in Canada: A Cross-Sectional Survey. *Patient*. 2022;15(2):171-185. doi:10.1007/s40271-021-00538-8
23. Siddiq S, Wilson BJ, Graham ID, et al. Experiences of caregivers of children with inherited metabolic diseases: a qualitative study. *Orphanet J Rare Dis*. 2016;11(1):168. doi:10.1186/s13023-016-0548-2
24. Cohen E, Lacombe-Duncan A, Spalding K, et al. Integrated complex care coordination for children with medical complexity: A mixed-methods evaluation of tertiary care-community collaboration. *BMC Health Serv Res*. 2012;12(1). doi:10.1186/1472-6963-12-366
25. Gordon JB, Colby HH, Bartelt T, Jablonski D, Krauthoefer ML, Havens P. A tertiary care-primary care partnership model for medically complex and fragile children and youth with special health care needs. *Arch Pediatr Adolesc Med*. 2007;161(10):937-944. doi:10.1001/archpedi.161.10.937
26. McClain MR, Cooley WC, Keirns T, Smith A. A survey of the preferences of primary care physicians regarding the comanagement with specialists of children with rare or complex conditions. *Clin Pediatr (Phila)*. 2014;53(6):566-570. doi:10.1177/0009922814528035
27. Farmer JE, Clark MJ, Drewel EH, Swenson TM, Ge B. Consultative Care Coordination Through the Medical Home for CSHCN: A Randomized Controlled Trial. *Matern Child Health J*. 2011;15(7):1110-1118. doi:10.1007/s10995-010-0658-8
28. Chiu LF, West RM. Health intervention in social context: Understanding social networks and neighbourhood. *Soc Sci Med*. 2007;65(9):1915-1927. doi:10.1016/j.socscimed.2007.05.035

29. Perry BL, Pescosolido BA, Borgatti SP. *Egocentric Network Analysis: Foundations, Methods, and Models*. Cambridge: Cambridge University Press; 2018. doi:10.1017/9781316443255
30. Lincoln Y, Guba EG. *Naturalistic Inquiry*. SAGE Publications; 1985.
31. Staniszewska S, Brett J, Simera I, et al. GRIPP2 reporting checklists: Tools to improve reporting of patient and public involvement in research. *BMJ*. 2017;358. doi:10.1136/bmj.j3453

Thesis Appendices

Appendix A: Ethics certificate

22/12/2021

Université d'Ottawa
Bureau d'éthique et d'intégrité de la recherche

University of Ottawa
Office of Research Ethics and Integrity

Lettre d'approbation administrative | Letter of administrative approval

Numéro de dossier / Ethics File Number	H-04-20-5757
Titre du projet / Project Title	Family-centred care for children with inherited metabolic diseases
Type de projet / Project Type	Recherche de professeur / Professor's research project
CÉR primaire / Primary REB	CHEO / CHEO
Statut du projet / Project Status	Renouvelé / Renewed
Date d'approbation (jj/mm/aaaa) / Approval Date (dd/mm/yyyy)	24/04/2020
Date d'expiration (jj/mm/aaaa) / Expiry Date (dd/mm/yyyy)	15/01/2023

Équipe de recherche / Research Team

Chercheur / Researcher	Affiliation	Role
Elizabeth POTTER	Département d'épidémiologie et santé publique / Department of Epidemiology and Public Health	Chercheur Principal / Principal Investigator
Monica LAMOUREUX	Children's Hospital of Eastern Ontario	Coordonnateur de recherche / Research Coordinator
Andrea CHOW	Département d'épidémiologie et santé publique / Department of Epidemiology and Public Health	Coordonnateur de recherche / Research Coordinator
Zobaida AL-BALDAWI	École interdisciplinaire des sciences de la santé / Interdisciplinary School of Health Sciences	Étudiant-chercheur / Student-researcher

Conditions spéciales ou commentaires / Special conditions or comments:

The uOttawa expiry date is set in accordance with the one from the CHEO-REB.

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22/12/2021

Université d'Ottawa
Bureau d'éthique et d'intégrité de la recherche

University of Ottawa
Office of Research Ethics and Integrity

L'Université d'Ottawa a signé une Entente, conforme aux exigences de la plus récente version de l'EPTC et tout autre règlement ou législation applicable, permettant au CÉR ci-haut nommé d'être désigné comme CÉR primaire pour les projets de recherche où

1) les activités principales de recherche sont menées sous l'autorité ou sous les auspices de l'établissement lié au CÉR primaire et

2) Une partie du projet est également réalisé sous l'autorité ou sous les auspices de l'Université d'Ottawa.

Cette lettre confirme que l'Université d'Ottawa a autorisé que le CÉR primaire soit le CÉR officiel pour l'évaluation et la supervision de ce projet de recherche. Ceci n'est pas une approbation éthique.

Afin de nous aider à garder votre dossier à jour, veuillez soumettre une copie de toutes demandes de modification, renouvellement d'approbation éthique etc. soumis à et approuvé par le CÉR primaire dès qu'elles sont disponibles.

Cette approbation administrative est valide pour la durée indiquée ci-haut et est sujette aux conditions énumérées dans la section intitulée « Conditions spéciales ou commentaires ».

The University of Ottawa has signed an Agreement, compliant with current TCPS guidelines and any other applicable guidelines or legislation regarding multisite review, allowing the REB named above to serve as Board of Record (BoR) for research projects where

1) the main research activities are conducted within the auspices or jurisdiction of the BoR's institution and

2) parts of the project are also conducted under the jurisdiction or auspices of the University of Ottawa.

This letter confirms that the University of Ottawa has authorized the REB named above to serve as Board of Record for the review and oversight of this research project. This is not an REB approval.

In order to help us keep your file up to date, please submit a copy of all amendment requests, project renewals or any other changes submitted to and approved by the BoR, as they become available.

Administrative approval is valid for the period indicated above and is subject to the conditions listed in the section entitled «Special conditions or comments».

Safaa LAMHOUJEB
Coordonnateur de l'éthique / Ethics Coordinator
Pour/For **Daniel LAGAREC** Président(e) du/ Chair of the **Comité d'éthique de la recherche en sciences de la santé et sciences / Health Sciences and Sciences Research Ethics Board**

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Appendix B: IMD Eligibility

Clinical trajectory	Chronic and generally non-progressive (Group 1)	Acute episodes of severe illness with or without accompanying chronic multi-system sequelae (Group 2)	Progressive multi-system disease (Group 3)
	Amino acid disorders: Phenylalanine hydroxylase deficiency; homocystinuria; tyrosinemia type I; guanidinoacetate methyltransferase deficiency; galactosemia; pyridoxine-dependent epilepsy	**Amino acid disorders:** Maple syrup urine disease	Diseases in this category are not specified. Instead, **IMDs should meet the following criteria:** ·Disease involves 3 or more organ systems ·Chronic complications of the disease become progressively worse over time, even with available treatment **Examples:** MPS Type I, MPS Type III, Hunters disease, Farber disease
	Urea cycle disorders: Citrin deficiency	**Urea cycle disorders:** Arginase deficiency; Argininosuccinic academia; Carbamyl phosphate synthetase deficiency; Citrin deficiency; Citrullinemia; Hyperornithinemia-Hyperammonemia-Homocitrullinuria syndrome; N-acetylglutamate synthetase deficiency; Ornithine transcarbamylase deficiency	
	Fatty acid oxidation disorders: Carnitine uptake defect	**Fatty acid oxidation disorders:** MCAD deficiency; VLCAD deficiency; Carnitine uptake defect; Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency; Trifunctional protein deficiency	
	Organic acid disorders: Glutaric acidemia type I	**Organic acid disorders:** ß-Ketothiolase deficiency; Glutaric acidemia type I; HMG-CoA lyase Deficiency; Isovaleric acidemia; Methylmalonic acidemias; Propionic acidemia	
	Other disorders: Multiple carboxylase/ biotinidase deficiency	**Other disorders:** Glycogen storage disease type 1; Multiple carboxylase/ biotinidase deficiency	

Appendix C: Care map instructions provided to participants

Making Your Child's Care Map

What is a care map?

A care map shows the people involved in your child's health care and how each person is connected to your child and to each other. An **example** is on **page 3.**

How to make your child's care map

The care map should reflect **how you see** your child's care, who's involved and how they're connected. There is no one way to create a care map. You can draw your own or use the template on page 4. It's up to you. Don't worry about getting it 100% right. If you would like, your child can help you draw the care map.

Things to remember

1. **Please do not put your child or other names on the care map.** Use "child," "family" and job titles instead.

2. When adding people or organizations that are part of your child's health care team. Group them together in a way that makes sense to you. See page 5 for examples of people and organizations that you could include. But there can be others!

3. **Try to include the people involved in your child's health care, not just organizations** (e.g., add teacher, Education Assistant, etc. instead of just "school").

What if my child sees 2 people with the same job title in the same clinic?

1) Label them Job Title #1, Job Title #2, etc.

2) Decide whether you consider one of them to be the **main "job title".** If yes, put a star next to Job Title #1. Example: if your child sees 2 metabolic physicians at the metabolic clinic, Dr. Chan, the one your child usually sees, and Dr. Singh, the one you see if Dr. Chan is away, label as follows: "Metabolic Physician #1★" and "Metabolic Physician #2". See Figure 1 below.

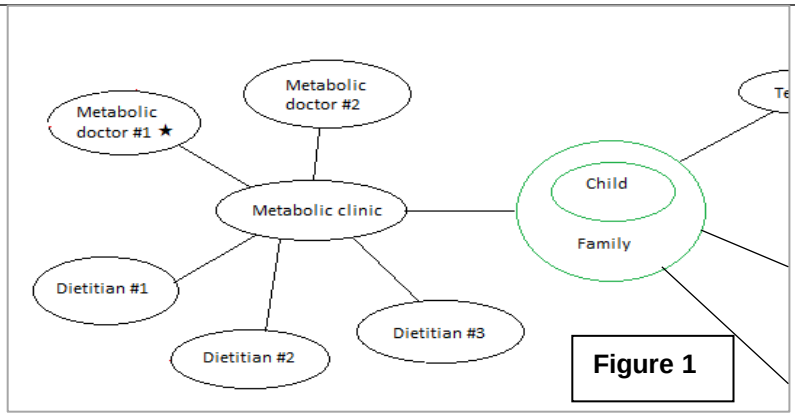


Figure 1

4. **Connect providers:** Add lines to connect people or groups who work together for your child's health care, for example, by sharing information, providing, or receiving referrals. People can be connected to others in same group or organization or at different groups. (See example, Page 3.)

> What if I don't know if 2 people work together or not?
>
> That's OK. Just draw the connections that you know about.

5. **IMPORTANT: Identify up to 10 key providers:** On the Care Map, put the letters "**KP**" next to that person's job title. **Key provider** = someone you think is key to your child's health care. If you do not think any of your child's caregivers is a key provider, just write "No key providers." (See example, Page 3.)

Once you are finished the care map

1. **Take a picture** of the care map or **save as a PDF** file. Make sure it is readable in the image.

2. To **upload the picture,** follow the steps in the email we sent you with this document. Please do not email the picture to the study team.

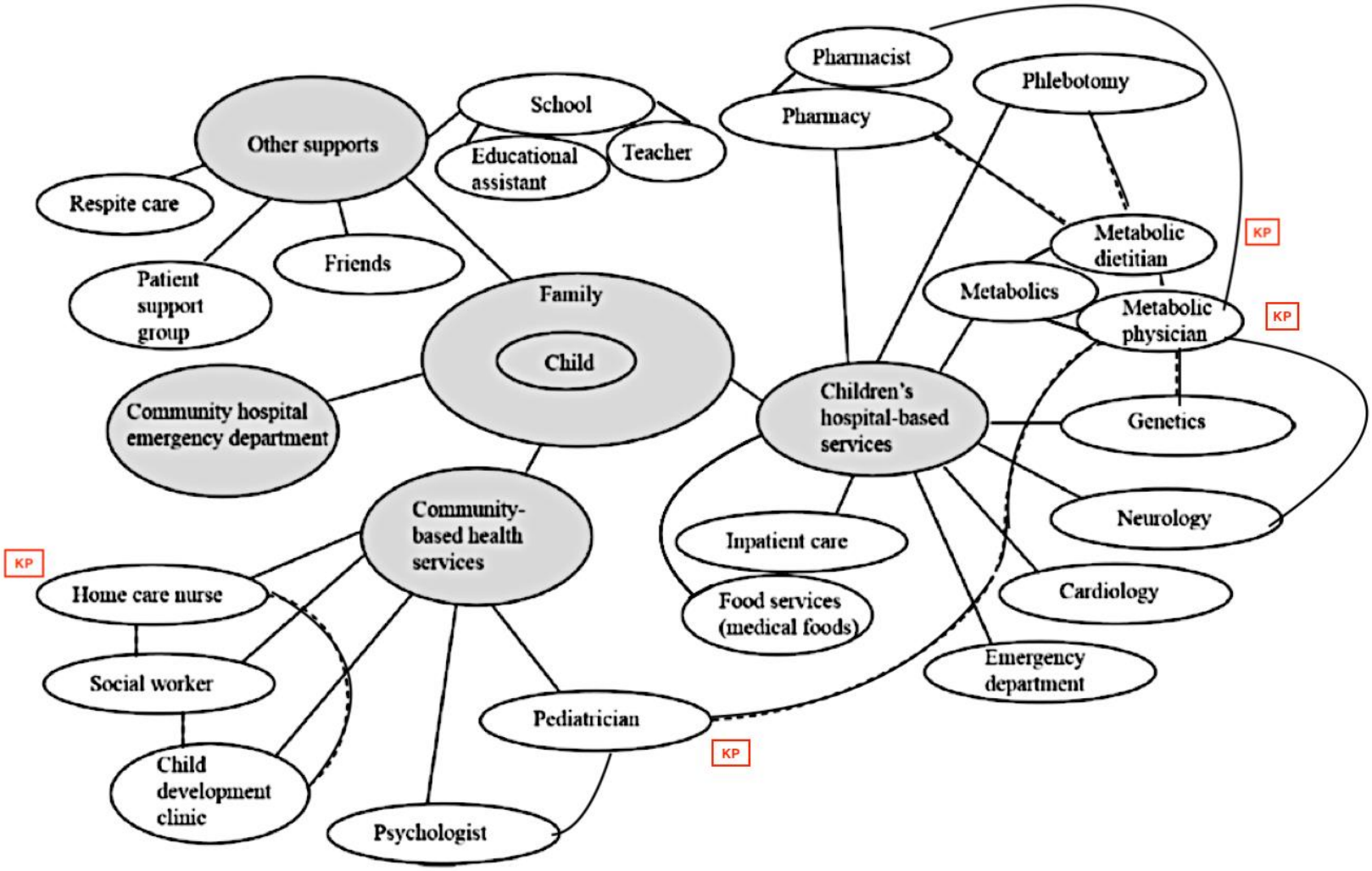
3. We will make a digital version of your care map. We will send you a link to view it and make sure that it is correct.

Questions?

If you have any questions while creating or uploading your care map, please **contact Andrea Chow**, study coordinator, at **613-562-5800 x4353**, or by email at achow@uottawa.ca**.**

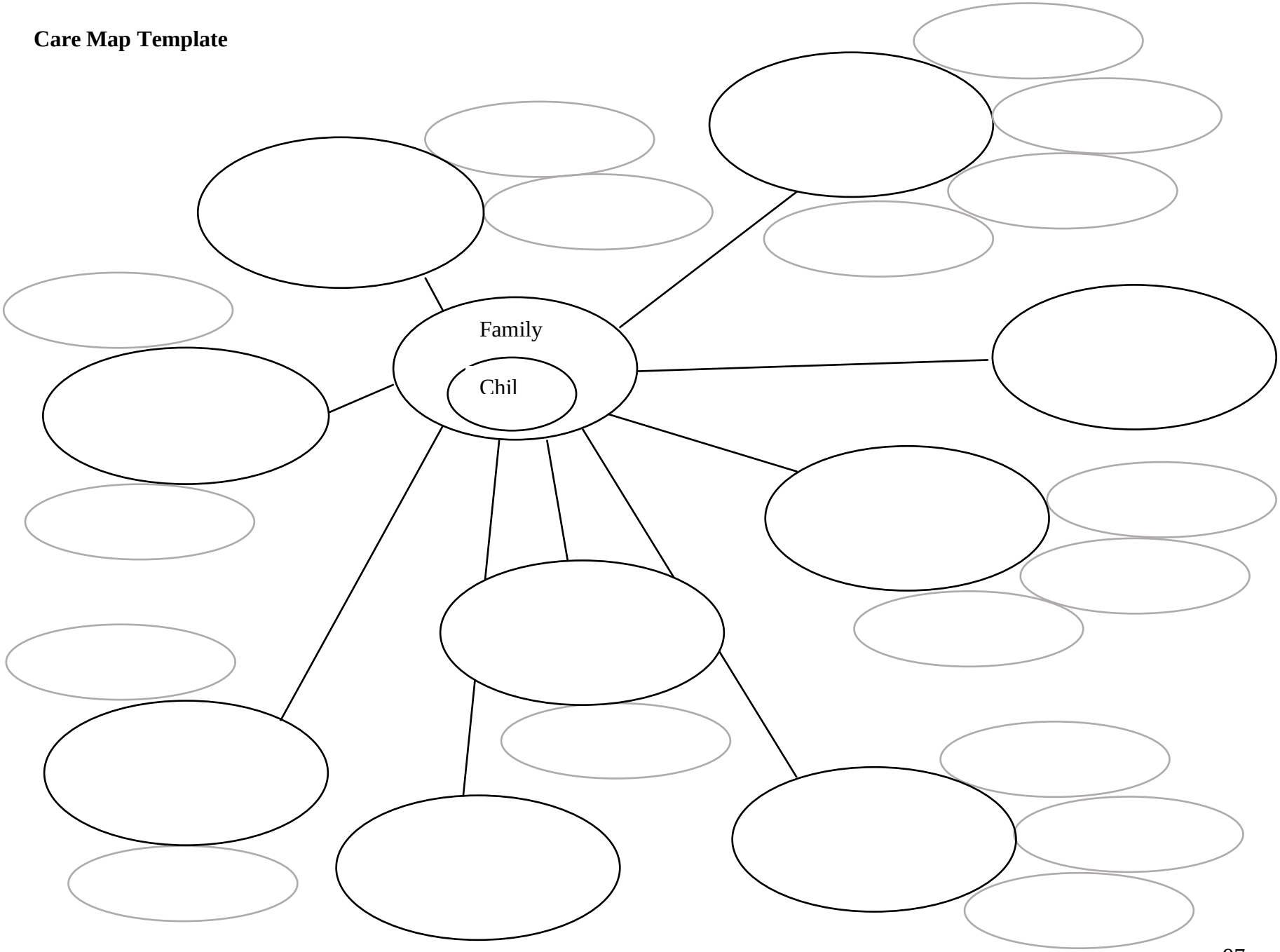
Instructions adapted from: Antonelli, RC and Lind, C. Care Mapping: A How-To Guide for Patients and Families. http://www.childrenshospital.org/-/media/Care-Coordination/CareMappingforfamilies21813.ashx?la=en&hash=D8C02FCA893C9A29C939613532334E07127BF9E6. Accessed September 8, 2017.

Care Map Example



Adapted from Dewan & Cohen. PaediatrChild Child. 2013,18,518; Miller et al. BMC Health Serv Res. 2009,9,242; Jarrett et al. BMJ upport Palliat Care. 2015.

Care Map Template



EXAMPLES

SETTINGS – IN HOSPITAL

Specialty Clinics
Audiology or Speech Therapy Clinic
Cardiology Clinic
Complex Care Clinic
Dental Clinic
Dermatology Clinic
Ear Nose Throat Clinic
Endocrinology Clinic
Gastroenterology Clinic
Hematology Clinic
Metabolic Clinic
Nephrology Clinic
Neurology Clinic
Optometry / Ophthalmology Clinic
Orthodontics Clinic
Orthopedic Clinic
Pediatrician's Clinic
Physiotherapy Clinic
Psychology / Psychiatry Clinic
Rehabilitation Clinic
Respirology Clinic
Rheumatology Clinic
Urology Clinic

Other
Ambulatory or Day Unit
Emergency Department
Feeding or Nutrition Clinic
Genetics Unit
ICU
Inpatient Unit
Laboratory
Mental Health / Counselling Services
NICU
Palliative Care Unit

SETTINGS – IN HOSPITAL CONTINUED

Sleep Clinic
Social Work Unit

SETTINGS - IN COMMUNITY

Blood Lab
Clinic
Community Centre
Daycare
Diagnostic Imaging or other Laboratory
Hospice
Primary Health Care Clinic
School
Sleep Clinic
Walk-in or Urgent Care Clinic
Your Home

JOB TITLES

Acupuncturist
Audiologist
Behavioural therapist
Cardiologist
Care coordinator
Chiropractor
Complex care doctor
Counsellor
Critical care doctor
Dentist
Dermatologist
Dietitian
Doctor
Ear nose throat doctor
Educational Assistant
Endocrinologist
Family doctor

JOB TITLES CONTINUED

Gastroenterologist
Genetic counsellor
Geneticist
Hematologist
Homeopath
Lab technician
Massage therapist
Mental health professional
Metabolic doctor
Naturopathic doctor
Nephrologist
Neurologist
Nurse
Nurse practitioner
Occupational therapist
Ophthalmologist
Optometrist
Orthodontist
Orthopaedic doctor
Palliative care doctor
Paramedic
Personal support worker
Pediatrician
Pharmacist
Pharmacy assistant
Pharmacy technician
Physical therapist
Physiotherapist
Psychiatrist
Psychologist
Respirologist
Rheumatologist
Social worker
Speech therapist
Surgeon
Therapist
Urologist
Radiologt

Appendix D: Guidelines for the study team for creating an electronic care map

Decision Guide for Creating an Electronic Care Map
The care map process begins when a participant submits a paper care map. In creating an electronic care map, the study team goes through rounds of communication on the secure REDCap platform with participants to ensure that details about settings, hubs of care, providers and connections between providers are accurate.

Care Map REDCap Codes
CT – Please select all providers that your child sees at this setting
CP – Please clarify this organization or place
CCa and CCb – Please clarify if these two people are supposed to be connected to each other
CF – Please clarify if this person is supposed to be connected to your family
CM – Please clarify if this is a star to indicate a main provider
CKP – Please clarify if this person is one of your child's key health care providers
CX – We have made an assumption. Is this correct?

Information about Hubs

Participants will typically group their providers by hubs – for example: community-based, hospital-based, etc. It is important for us to gather as much information as possible about those hubs. Some common things we have been noting/checking with participants are:
→ If a participant lists speciality clinics such as 'metabolic clinic' and 'hospital-based services' as separate hubs, **clarify where the 'hospital-based services' is located**
→ Can use information in the baseline questionnaire (i.e., distance to clinic) to help aid in phrasing questions; if participant lives close to center, can frame questions with the assumption that the hospital-based services is located at children's hospital. If a participant lives far from center, can frame question with the assumption that the hospital-based services is located at a local hospital. Questions can be framed in two ways, using either one of these REDCap codes:
→ Can use a CP code with this question: "CP: Is the 'hospital-based services based at the children's hospital'?
→ Can use a CX code with this question: In the hospital-based services bubble, insert either 'children's hospital.'
→ Ensure that connections within larger hubs (e.g., 'Hospital-Based Services' and 'Community-Based Services') are understood **(We will not be making the assumption that everyone in those hubs is connected).**
→ Ask about providers in hubs such as 'blood lab', 'pharmacy.'
→ If family is listed in 'other supports,' label as 'extended family.'

Information about Providers

→ Ensure that we know about providers NOT only settings (e.g., if cardiology clinic is listed → ask if cardiologist or other providers are seen there).
→ Our aim is to gather information about specific providers at each setting. If a participant lists a setting with no provider attached to it, there are a few options:
→ If it is **safe to make an assumption about the provider titles, you can do so with a CX code**. For example, if a participant lists 'cardiology clinic', it is safe to assume that

the participant sees a ‘cardiologist’ here. Keep the setting bubble and add the ‘provider’ bubble attached to it with a CX code.

→ If a **setting is more ambiguous/less common/could have the potential for numerous providers** (e.g., sleep clinic, blood lab, pharmacy), you can **use CT code**.

→ Provider titles at clinics should be harmonized where applicable (e.g., dietitian, metabolic doctors).

Information about Connections

→ Another one of our aims is to ensure that we understand connections between providers and settings correctly

If **a participant draws a connect between one provider and a setting** (e.g., pediatrician and hospital-based services hub), we need to know whether they mean the provider is connected to everyone at the setting

→ Can ask about this with an open-ended question such as this one: “‘Does Provider X works with everyone Location Y?’ If yes, there is nothing to do! If no, select ‘yes’ to the question ‘do you want the study team to make any changes to your child’s care map?’ use the textbox to tell us who they work. “

→ Can also add a CX code and adapt the wording of the question as needed

→ Ensure that connections within larger hubs (e.g., ‘Hospital-Based Services’ and ‘Community-Based Services’) are understood **(We will not be making the assumption that everyone in those hubs is connected).** Sometimes, participants will draw ambiguous lines in these big hubs. We have to make sure that we explicitly know whether everyone in those hubs is connected.

Appendix E: Guidelines for the study team for care map data entry

Electronic Care Map – Decision Guide for Data Entry

We recognize that participants conceptualize their child's network of care in terms of both settings and individual providers.

Sometimes a setting is small and cohesive enough that we are assuming that the providers at that setting are connected (e.g., a doctor and nurse in the same clinic). For some types of settings like this, participants actually seem to connect the care to the setting itself instead of the person (e.g., the blood lab or emergency department, where it can be hard to distinguish the roles of different providers). At all of this type of small and/or cohesive settings, we link all providers.

At other times, participants use hubs on their care maps simply as a way of organizing providers, either by geographic location (e.g., different clinics within the same hospital) or by category of care (e.g., community services, or friends and family). In these cases, we do not assume that the providers are linked.

<u>Checklist for Data Entry</u>

Assumptions about connections

- If a line connects two providers, list them as connected.
- Assume all providers in a small, cohesive hub (e.g., metabolic clinic, primary care clinic, blood lab, emergency room) are connected.
- At other times, participants use hubs on their care maps simply as a way of organizing providers, either by geographic location (e.g., 'Hospital-Based Services', 'Community Based Services') or by category of care (e.g., 'Other Supports'). In these cases, we DO NOT assume that the providers are linked.
- Assume that if a line connects a service (e.g., Blood lab) and another service (e.g., Primary Care Clinic) that everyone in those services is connected to each other. E.g., if a phlebotomist (located at blood lab) is connected to a primary care (with a doctor and nurse in it) then the phlebotomist is connected to both the doctor and the nurse.
- If a participant draws a LINE specifically connecting ONE provider in a hub to provider in another hub, we will honour that line (e.g., if there is a metabolic hub with a doctor and a dietitian, and a connection ONLY between the metabolic doctor and another provider, will honour that line and WON'T assume that the dietitian is connected to that other provider).

Genericizing the Provider Titles

- When a map has a bubble 'doctor' in 'clinic', enter provider as 'speciality title'. For example, if a map has a bubble titled 'cardiology' with an adjacent 'doctor' bubble, enter it as 'cardiologist' NOT as doctor.
- If a participant adds 'private' in front of a specific provider type, leave out the 'private' part.

Appendix F: Care map questionnaire

For each key provider identified on the Care Map, the following two questions are asked:

Question	Response options
How well does each of your child's key Health Care Providers know your child?	5-point Likert type scale
How well do you think your child's key health care providers coordinate your child's care with other providers?	5-point Likert type scale

Appendix G: Qualitative Interview Guide

Aims: (i) To get a more in-depth understanding of parental perceptions of children's care networks; (ii) To identify providers viewed as most important by parents and the role they play in the child's care; (iii) To identify how providers and services are connected.

Introduction

1. Share aims of the interview
2. Reminder of confidentiality: None of the information you share now will be shared outside the study team; will not be shared with the hospital or health care providers.
3. The Research Ethics board requires us to let you know that the only exception to confidentiality is in cases of suspected abuse or harm to children, which must be reported.
4. Reminder not to share names of care providers. If shared, will not be recorded in the transcripts
5. Voluntary participation – do not have to answer any questions that you do not want to answer
6. Can stop or take a break at any time
7. Reminder of audio and video recording and transcription of audio recording. Explanation of deletion of audio and video files immediately after transcription.
8. Remind participant of note taking during interview.
9. Remind participant that the submitted care map will be the topic of interview.

Questions

[This is a semi-structured interview. These are the main questions that we plan to pose to all participants but responses to each question may be probed further on a case-by-case basis.]

To start, could you tell me about is required to manage your child's care?

[Alternative] On a typical week, walk me through some of the things you do to manage your child's care?

Overall Network of Care

1. Can you please walk me through your child's network of care?
    - *Probe for specific aspects related to:*
        - Validation of listed providers and connections – is the network accurate as it is, or would you like to make any changes to it?
        - The process of drawing the network of care- how did you decide who to include in the network?

Identification of Key Providers

2. You identified [provider X] as a key provider. What are the factors that make them a 'key provider' for (kid's name)?
    - *Probe for specific aspects related to:*
        - From the care map questionnaire, I noticed that you indicated that this provider knows your child very well. What does that look like to you? (How do you know?)

- How often does (kid's name) interact with the provider?
- What is the provider's role in the child's care?

Care Coordination

3. You identified that [provider X] and [Provider Y] are connected. Can you tell me about that connection?
    - *Probe for specific aspects related to:*
        - How do provider X and provider Y work together, in your view?
        - What is the nature of the connection?
        - What is the impact of the connection on the family? How can you tell?
4. On the care map questionnaire, you told us that [provider X] coordinates with other providers "very well."
    - How does provider x work with other providers (e.g., shares information, makes referrals)? Can you tell me about factors that influenced your positive rating?

Adequacy of Network of Care

5. How well does this network of care meet your child's needs? How does this network of care meet your needs?
    - *Probe for specific aspects related to*
        - How did you get to this point where you like this network? Was it always this way? Has it always worked really well?
        - What do you believe your role has been in setting up this health care network?
        - Are there parts of the network that work better than others? What parts work better? In what ways?
        - What can be improved in this network of care? How could the network be improved to better meet (kid's) needs?
        - Are there people who should be key providers, but they are not listed as such? Who and how come?
        - Are there providers who should be connected on your care map but who are not currently connected? Which providers do you think should be connected? How would this help?
        - Is there a provider that you consider as the person who brings all providers together?

Is there anything you'd like to add about your care map that I didn't ask you about?

Appendix H: Network size distribution

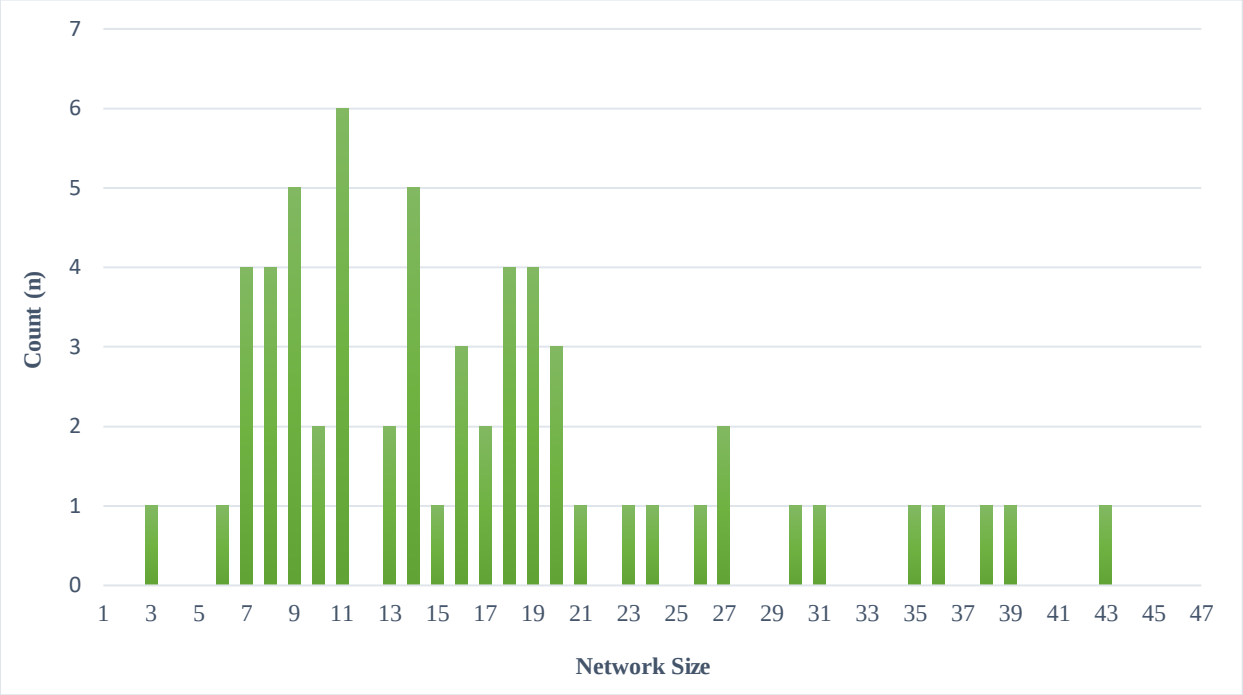


Figure 1. Network size distribution across sixty care networks for children with IMDs.

Appendix I: Full list of provider types identified on care networks for children with IMDs

Table 1. Providers and supporters involved in care networks of children with IMDs, from parents' perspectives

Provider	Children with this provider in network N (%)	Range per participant [a]	Median degree centrality [a,b]	Median share[a,c]	Children with this provider as a key provider in network n (%)	Key provider range per participant [d]	Median degree centrality for key provider [d,e]	Median share for key provider[d,f]
Metabolic doctor	55 (92%)	0-2	3 (2-4)	0.19 (0.13-0.25)	40 (78%)	0-1	3 (2-4)	0.20 (0.13-0.25)
Lab technician	55 (92%)	0-1	1 (0-3)	0.07 (0 - 0.15)	6 (10%)	0-1	1 (0-3)	0.09 (0 - 0.15)
Dietitian	45 (75%)	0-2	3 (2-4)	0.17 (0.08 – 0.25)	33 (55%)	0-2	3 (2-4)	0.20 (0.11-0.25)
Family doctor	41 (68%)	0-3	0 (0-1)	0 (0-0.10)	14 (23%)	0-2	1 (0-3)	0.06 (0-0.17)
Nurse	40 (67%)	0-5	2 (1-4)	0.13 (0.06 – 0.18)	11 (18%)	0-1	3 (3-4)	0.17 (0.15-0.20)
Friends	40 (67%)	0-1	0	0	0	//	//	//
Pediatrician	39 (65%)	0-3	1 (0- 3)	0.06 (0-0.13)	27 (45%)	0-1	1 (0-1)	0.03 (0-0.06)
Extended family	23 (38%)	0-1	0	0	2 (3%)	//	//	//
Social worker	22 (37%)	0-3	1 (0-3)	0.06 (0 – 0.16)	4 (7%)	//	//	//
Daycare staff	21 (35%)	0-1	0	0	2 (3%)	//	//	//
Dentist	20 (33%)	0-2	0 (0-1)	0 (0 – 0.04)	6 (10%)	0-1	0 (0-1)	0 (0-0.02)
Occupational therapist	19 (32%)	0-4	0 (0-1)	0 (0-0.05)	4 (7%)	//	//	//
Physiotherapist	18 (30%)	0-3	0 (0-1)	0 (0-0.02)	6 (11%)	0-1	0	0
Teacher	18 (30%)	0-4	0	0	3 (5%)	//	//	//
Pharmacist	17 (28%)	0-2	0 (0-1)	0 (0-0.02)	4 (15%)	//	//	//
Ophthalmologist	16 (27%)	0-1	0 (0-1)	0.005 (0.0 - 0.03)	5 (8%)	//	//	//
Neurologist	16 (27%)	0-1	1 (0-4)	0.03(0.0-0.13)	8 (13%)	0-1	2 (0-5)	0.04 (0.02-0.09)
Cardiologist	14 (23%)	0-1	1 (0-2)	0 (0-0.07)	3 (5%)	//	//	//
Audiologist	13 (22%)	0-2	0 (0-2)	0 (0-0.01)	3 (5%)	//	//	//
Genetic counsellor	13 (22%)	0-1	3 (2-3)	0.14 (0.10 – 0.17)	2 (3%)	//	//	//
ER staff (Doctors, nurses)	13 (22%)	0-3	1 (0-1)	0.03 (0-0.25)	2 (7%)	//	//	//

Geneticist	12 (20%)	0-1	3 (2-5)	0.12 (0.04-0.17)	8 (13%)	0-1	5 (3-6)	0.11 (0.14-0.27)
Imaging Services Staff	11 (18%)	0-1	0	0	0	//	//	
Educational assistant	10 (17%)	0-1	0 (0-1)	0 (0-0.04)	2 (3%)	//	//	//
Orthopedic doctor	10 (17%)	0-3	1 (0-2)	0.02 (0.00 - 0.05)	4 (7%)	//	//	//
Optometrist	9 (15%)	0-1	0 (0-1)	0 (0-0.04)	4 (7%)	//	//	//
Ears, Nose and Throat Doctor	8 (13%)	0-1	0 (0-1)	0 (0-0.04)	2 (3%)	//	//	//
Speech therapist	8 (13%)	0-1			1 (4%)			
School Principal	8 (13%)	0-1	0 (0-2)	0 (0-0.07)	0	//	//	//
Nurse Practitioner	7 (12%)	0-4	2 (0-5)	0.10 (0-0.12)	4 (7%)	//	//	//
Endocrinologist	7 (12%)	0-1	0 (0-2)	0 (0-0.07)	1 (4%)	//	//	//
Respirologist	7 (12%)	0-1	0 (0-2)	0 (0-0.07)	1 (4%)	//	//	//
Clinic technician/assistant	7 (12%)	0-2	0 (0-2)	0 (0-0.07)	0	//	//	//
Respite staff	7 (12%)	0-1	0	0	0	//	//	//
Grandparents	6 (10%)	0-1	0	0	0	//	//	//
Gastroenterologist	6 (10%)	0-1	1 (0-1)	0 (0-0.04)	1 (4%)	//	//	
Care coordinator	6 (10%)	0-1	5 (0-10)	0.07 (0-0.09)	3 (5%)	//	//	//
Counsellor	6 (10%)	0-1	0	0	0	//	//	//
Surgeon	6 (10%)	0-1	1 (0-4)	0.005 (0-0.11)	3 (7%)	//	//	//
Families with IMD social media support group	6 (10%)	0-1	0	0	0	//	//	//

Remaining types of providers on networks include: Speech language pathologist, dental hygienist, hematologist, nephrologist, orthodontist, psychologist, resource teacher, special education teacher, pediatric ward team, behavioural therapist, feeding assistant, massage therapist, naturopathic doctor, rare disease patient organization caseworkers, on-call metabolic doctor, school staff, therapist, chiropractor, orthopedics equipment supplier, orthoptist, palliative care doctor, paramedic, psychiatrist, venous access team, cooks, student doctor, equine therapy staff, child life specialist, physical therapist, critical care doctor, financial assistance caseworker, hard of hearing teacher, oncologist, pharmacy assistant, urologist, vice principal, porter, drug program coordinator, speciality food shop staff, bus driver, hepatologist, food services staff, speech language assistant, neuromuscular doctor, babysitter, neighbors, church and complex care doctor.

Remaining types of key providers on networks include: optometrist, orthopaedic doctor, nurse practitioner, social worker, cardiologist, care coordinator, audiologist, behavioural therapist, chiropractor, dental hygienist, massage therapist, psychologist, special education teacher, therapist, critical care doctor, hematologist, naturopathic doctor, orthodontist, pharmacy assistant, venous access team,

speciality food shop staff, bus driver, feeding assistant, neuromuscular doctor, food services staff, babysitter, speech language pathologist, complex care doctor, physical therapist, and speech language assistant

Appendix J: Full list of settings identified on care networks for children with IMDs

Setting	Number of children with this setting on care map
Metabolic Clinic - Hospital	58 (97%)
Clinic - in community	57 (95%)
Blood Lab - in community or hospital	55 (92%)
School	33 (55%)
Optometry / Ophthalmology Clinic - Hospital	15 (25%)
Cardiology Clinic - Hospital	10 (17%)
Audiology or Speech Therapy Clinic - Hospital	9 (15%)
Neurology Clinic - Hospital	9 (15%)
Orthopedic Clinic - Hospital	9 (15%)
Emergency Department - Hospital	8 (13%)
Pharmacy - in community	8 (13%)
Dental Clinic – in community	7 (12%)
Endocrinology Clinic - Hospital	6 (10%)
Gastroenterology Clinic - Hospital	6 (10%)
Ear Nose Throat Clinic - Hospital	5 (8%)
Nephrology Clinic - Hospital	5 (8%)
Respirology Clinic - Hospital	5 (8%)
Social Work Unit - Hospital	5 (8%)
Inpatient unit - Hospital	4 (7%)
Pediatrician's Clinic - Hospital	4 (7%)
Radiology Unit - Hospital	4 (7%)
Sleep Clinic - Hospital	4 (7%)
Genetics Unit - Hospital	3 (5%)
Complex Care Clinic - Hospital	2 (3%)
Orthodontics Clinic - Hospital	2 (3%)
Hematology Clinic - Hospital	2 (3%)
Physiotherapy Clinic - Hospital	2 (3%)
Psychology / Psychiatry Clinic - Hospital	2 (3%)
Pharmacy - in Hospital	2 (3%)
Community Centre	2 (3%)
Dermatology Clinic - Hospital	1 (2%)
Feeding or Nutrition Clinic - Hospital	1 (2%)
NICU - Hospital	1 (2%)
Palliative Care Unit - Hospital	1 (2%)
Rehabilitation Clinic - Hospital	1 (2%)
Diagnostic Imaging Lab - in community	1 (2%)
Seating clinic- Hospital	1 (2%)
Feeding assessment clinic - Hospital	1 (2%)
Rapid Access clinic - Hospital	1 (2%)
Telehealth- Hospital	1 (2%)
At Infant and Preschool Assessment Service Clinic	1 (2%)
At Child learning and development non-profit organization	1 (2%)
At Family Support for Children with Disabilities	1 (2%)

Appendix K: Table of network archetypes according to child age, distance to the metabolic group and IMDs group.

Table 2. Post-hoc exploratory analysis of network archetypes by child age, IMDs trajectory group, and travel time to the metabolic centre

	Small networks (n=23)	Large and sparse networks (n=23)	Large and relatively connected networks (n=14)
Travel time to the metabolic centre			
1 hour or less	15 (65%)	13 (56%)	7 (50%)
More than 1 hour and up to 3 hours	2 (9%)	8 (35%)	2 (14%)
More than 3 hours	6 (26%)	2 (9%)	5 (36%)
Age group (years)			
≤1 – 3	16 (70%)	6 (26%)	4 (29%)
4 – 6	5 (21%)	7 (30%)	4 (29%)
7 – 12	2 (9%)	10 (43%)	6 (42%)
IMDs group			
IMDs Group I (chronic and non-progressive)	11 (48%)	4 (17%)	2 (14%)
IMDs Group II (acute and episodic)	12 (52%)	9 (39%)	7 (50%)
IMDs Group III (progressive and multi-system)	0	10 (44%)	5 (36%)

Appendix L: Additional illustrative quotations supporting all qualitative themes

From section 2.2.2 Characteristics of children's care networks
Theme: parental reflection on process of drawing care maps
"*I kind of like worked through what are the important things in [child's] life. Like, people who take care of him, people who see him."* (Participant #3, Mother of a child with an IMD in Group 2 and 'small' network)
"I keep a binder of all the people that we see, so I actually just kind of went, "Okay, all of these people, you know, are important to us, and all of these people. He needs to follow-up with them to make sure that everything is going on track". So, I just kind of actually went from that binder and went, "Okay, well it all starts here with the Children's Hospital", and it just kind of all fans out from there." (Participant #9, Mother of a child with an IMD in Group 3 and 'large and relatively connected' network)
"No, it was really eye-opening to see really who is involved in our lives. It was huge, like, especially for us, I feel like this is a big care map, and um, and just all the little pieces that fit and who goes where, and how important these all are." (Participant #1, Mother of a child with an IMD in Group 3 and 'large and sparse' network)
Theme: the dynamic nature of IMD care
"The metabolic doctor and dietitian, we usually see every six months." (Participant #4, Mother of child with an IMD in Group 2 and 'large and sparse' network)
"We are followed by the pediatrician here, and usually that happens about every 3 months" (Participant #2, Mother of a child with an IMD in Group 1 and 'large and relatively connected' network)
"All of the specialists are kind of every once in a while, like every 6 to 12 months unless there's something else going on" (Participant #6, Mother of a child with an IMD in Group 3 and 'large and sparse' network)
"*Sure, so, actually so presently, it's a little bit less than what it used to be, which is wonderful… Neurology, we see them a bit more, because that's more of a symptom we have right now that they are following, seizures. So, we see Neurology a bit more*." (Participant #1, Mother of a child with an IMD in Group 3 and 'large and sparse' network)
"*It [visits to pediatrician] changes in frequency as she is getting older, and she is getting more stable with, her different medical needs. I think this last time, she graduated her to every nine months, but I think it's been every six months for the past little while."* (Participant #2, Mother of child with an IMD in Group 1 and 'large and relatively connected' network)
Theme: Parental responsibility – parents as 'managers' of their children's care networks
"At the beginning we felt, we were in the learning phase, learning all about the condition, and what it means for our family, and once we had a better grasp of that and felt a little bit more on our feet with the information, then we started to ask more questions about how we could do things better. And that's how we ended up being connected to the pediatrician locally, so that we could do things like blood work without the drive. Those types of things, yes, I think we had

a role in that." (Participant #10, Mother of a child with an IMD in Group 1 and 'large and sparse' network)
"In the very beginning when [child] was diagnosed… we were starting to learn everything that we needed to learn to give him the best care, and you know, the doctor said to us, "What you need is [highly specialized therapy]"... So, we started to fundraise. We actually joined up with another family who have the same diagnosis..; and we started to fundraise, and started to sponsor studies to further research…they started the clinical trials and I ended up deciding to put him into the trial.." (Participant #9, Mother of a child with an IMD in Group 3 and 'large and relatively connected' network)
"And [care provider's] son plays hockey with [child], so we kind of have also like a social relationship, which is nice. It's only because of that. You are not always going to see the same doctor, and so they may not actually know, like, you know, they have to check the chart, they. It's even the same for adults, right? Like, I go to the doctor, they have to read my chart, and they don't know me either. So, it's just because of the way it works. Yeah." (Participant #3, Mother of a child with an IMD in Group 2 and 'small' network).
"I guess the only other issue would be a little bit more communication with possibly the ophthalmologist and the metabolic clinic. We run into issues where, even though they work in the same hospital, they don't use the same charting software. So, the ophthalmologist being the way that they are, where we don't get a whole lot of like, information from them, it is then up to us to pass on the two sentences that we did get (laughs) to the metabolic doctor when we see them. So, it would be nice if our metabolic doctor had access to a little bit more information from the ophthalmologist, for sure." (Participant #4, Mother of a child with an IMD in Group 2 and 'large and sparse' network)
<u>From section 2.2.3 Key providers and relational continuity</u>
Theme: the characteristics of key providers
"The ophthalmologist, because of her vision, it's a daily thing, it's a daily struggle with her vision. I don't know how much you know about her, but she had [type of surgery] surgery, so she wears contact lenses and glasses, and then she is going to have a [second] surgery … so the ophthalmologist we see on a regular basis, because we are constantly watching her eyes, and so they are a pretty key provider as well" (Participant #2, Mother of a child with an IMD in Group 1 and 'large and relatively connected' network)
"Um, but yeah, his metabolic doctor has always, is just a kind person. He's always been able to, when we have weird questions about something he is like, doing, or like, the way he's behaving, or whatever, whatever we see, he's always been really quick to get back to us if we are concerned" (Participant #2, Mother of a child with an IMD in Group 1 and 'large and relatively connected' network)
"I think accessibility has to be a big key. Being accessible, like, I can call them and get an appointment within a day if I am very concerned." (Participant #6, Mother of a child with an IMD in Group 3 and 'large and sparse' network).
"Our Metabolic doctor is also the lead investigator in the clinical trial that my son is participating in, at Hospital-Based Services #1. Whereas a typical [IMD] patient would only see their Metabolic doctor once every six months, we see her once a month. That's why we don't see our family doctor at all. I am lucky enough to have her in front of me once a month,

so I have all of my questions answered, and we have referrals put in place for different clinics if there is a need, like, more often than once every six months, which is where we would normally have that person involved. [Doctor] is available even when I don't see them every once a month. I can call them, or email them, and they will look into different clinical trials for us. They will answer any questions that I might have." (Participant #8, Mother of a child with an IMD in Group 3 and 'large and relatively connected' network)
"Responsiveness. When you have a question, obviously you have reached out because it is urgent and it's important. For one example, also related to the medication, we had just come home with our new supply, and it needs to be refrigerated, and my husband wasn't thinking, and put it in the freezer instead of the fridge, and we didn't know what impact that would have on the drug, because we didn't discover it until the next day. We reached out to the geneticist, and within an hour, I had an answer (laughs). You know, just that sort of responsiveness and knowing that when we need support in some way, that they are there." (Participant #9, Mother of a child with an IMD in Group 3 and 'large and relatively connected' network)
"So… another example, my daughter reacted to amoxicillin in the hospital, and you know, the hospital dealt with it, and we dealt with it when we were in the emergency room, but then we went back to the pediatrician a day or two later, and she went and got us the appointment for an allergist to get all that testing done. So, she's kind of the main person that we would go to, to get referred out to other places, to get any community supports that are not one of these clinics, which are not people we would see regularly; for example, the allergist, which we have only seen once, or she got referred to for early intervention for speech, and things like that. So, she's definitely the one that keeps track of all the blood tests and results, and she has a folder, probably like six inches thick on all of her blood tests, and ultrasounds and everything, so she is definitely the main provider for just general needs." (Participant #2, Mother of a child with an IMD in Group 1 and 'large and relatively connected' network)
"That doctor is the one, like, if he goes to the hospital, that's who they call, as opposed to like, a family doctor or whatever. So, yeah, he's always just been sort of at the centre of everything. Like, when we go to like, the pediatrician, or the ophthalmologist, or the neurologist, they all go back to him. If they are unsure of something, because it's such a unique condition that most, I don't think any other provider we have met has ever heard of it before." (Participant #4, Mother of a child with an IMD in Group 2 and 'large and sparse' network)
"So, she plays a big role there to make sure that we are on track with all of the follow-up, and she's kind of the go-to. When I have a question or concern, or even a little thing like, um, for example, should he get the COVID vaccine? I need to ask somebody that question, and she is the go-to, yeah, so she's very important, yes. She would be a #1." (Participant #9, Mother of a child with an IMD in Group 3 and 'large and relatively connected' network)
"And it just, there is a he human side to him, right? Like, he's not just the doctor, but he is a man who cares, I think genuinely about [son]." (Participant #1, Mother of a child with an IMD in Group 3 and 'large and sparse' network)
Theme: Relational continuity.
"We have had both of them involved in my child's care since the diagnosis. It was actually the metabolic doctor that had reached out to us to let us know about the diagnosis, and back at the beginning, we saw them on a weekly basis for probably 2 or 3 years, so there is quite a relationship that has been built up there over time. We are down to only seeing them about 3

times a year now, but there is still interaction over email as well." (Participant #10, Mother of a child with an IMD in Group 1 and 'large and sparse' network).
"You know, what it is, it's when it's the, whomever is on-call on your, on the 10th floor, which is your Peds floor, whoever, say, is the on-call doctor, like, starts over again with all of, like, "Oh, I see this" and it's probably them just trying to get a really good feel, but that, I think that's more of a, more, more, like, that creates a lot of just, anxiety, like a bit of anxiousness when you are there, because you are like, "Oh, that we have to start all over again" (laughs). Like, here's the symptoms, here's this. I think that's my biggest thing, is when, when they are admitted, right?" (Participant #1, Mother of a child with an IMD in Group 3 and 'large and sparse' network)
Theme: Mutual trust
"Just by knowing my child history. By encompassing, I guess seeing everything that he has been going through. A lot of the things that the metabolic doctor has been advising me to do is just, like, if I have any reports on anything, just to send it to the metabolic doctor, so that they would have a very complete and thorough look at what is happening with him. For example, his school gives him an [special education plan] every year, just like a plan of the things of how he is progressing. That's been sent to the metabolic doctor and put in his file so that all of his specialists can understand and look through that and see where [child] is at in terms of, you know, following instructions, or how he's doing in school, and may be there is something related to school that might be affecting his sleep or whatever, you know." (Participant #6, Mother of a child with an IMD in Group 3 and 'large and sparse' network).
<u>From section 2.2.4 Providers as part of a dynamic network and care coordination</u>
Theme: Ways of coordinating care
"So, because [son] disease, you can not um regularly, um, uh, sedate, give him sedation, and put them under…we debated for a long time whether or not (schedule an orthopedic surgery)… and so, when we finally did, I, we said, like, he also has dental work that does need to be taken care of as well, so, in one shot he would be sedated, and … that doctor (who performed the orthopedic surgery), and the dentist, of course had to do a lot of communication beforehand, and usually the Anesthesiologist, you know, comes in, into that. So, the communication, you know, timing. The Dentist actually, you know, didn't work at the hospital, so she had to coordinate her time to come in at the same time as, like, our Orthopedics appointment, and (laughs). So, important to us, but I knew in the end that both were completely, um, I have very much trust for both of them. This Dentist actually had already previously, years before, did work, did connected work with the Ears, Nose, Throat doctor. Um, we coordinated that at one time. So, so that was the connection between those two." (Participant #1, Mother of a child with an IMD in Group 3 and 'large and sparse' network)
"If I find it's getting too long, which I've done in the past, I just call them, or I just tell the pediatrician, and she just puts in the referral herself to the gastroenterology team to get us seen, so, it just depends." (Participant #2, Mother of a child with an IMD in Group 1 and 'large and relatively connected' network)

"Um, well Dr. #1 and Dr. #2, I think they, they are the ones who referred us to the dietician to see how [child] is doing in terms of her growth and stuff like that." (Participant #7, Father of a child with an IMD in Group 1 and 'small' network)
"So, usually in our appointments the dietitian will come in first, and she will sort of like, take like, a food history log, review medications, and that kind of stuff, and then she leaves, presents it to the doctor, they both come back. So, she sometimes, if he is busy (laughs) the doctor, or maybe like, misunderstands something we've said, she's always there to clarify, or like, "Oh, they were", or like, if we ask. (Sorry, I'm not like being coherent right now). If he like, presents something to her, like, "Oh, we are worried about this level in his blood work", um, she'll bring it up to him." (Participant #4, Mother of a child with an IMD in Group 2 and 'large and sparse' network)
"Usually when we see a specialist, the specialist will send that directly to the physician. In [province], we have what is called [online healthcare communication system], so if all of the physicians are on [online healthcare communication system], they can see everything related to my child." (Participant #6, Mother of a child with an IMD in Group 3 and 'large and sparse' network)
Theme: Need for a network "go-to" person
"Basically, the nurse is the person that I talk to the most. Usually, I would send an email to the nurse. The nurse would relay all that information to the metabolic doctor, and then she would just send back a brief overview of what she told the doctor and what needs to happen, or what she has done, or what not. So, she's basically the main person that I connect with." (Participant #6, Mother of a child with an IMD in Group 3 and 'large and sparse' network)
"What I do sometimes, if I can't find the Metabolic doctor, I can go to that nurse and she will find the doctor and have the doctor call me if I need anything. That happened recently, where I needed to know something right away. I didn't feel comfortable calling the doctor on her cell phone, because it wasn't an emergency, but I just needed to know something right away, so I know that nurse is actually in the same office as the Metabolic doctor, and that if the doctor wasn't sitting right there, she would know where the doctor was, and so she did, and she coordinated that for me. She found the doctor for me and let her know that I needed to speak with her, and then the doctor called as soon as she could." (Participant #8, Mother of a child with an IMD in Group 3 and 'large and relatively connected' network)
Theme: Impact of poor care coordination
(no additional quotations)
<u>From section 2.2.5: Summary of parent recommendations for care network improvements</u>
"I would say the pediatrician, although the pediatrician does look after my son's physical well-being and physical health, the pediatrician maybe was not as aware of all the things that we needed to look for. My son's condition is the first time that my pediatrician has ever seen this condition. It was new to both of us, and with that being said, she wasn't fully aware, even though you know, she was seeing him since he was an infant, wasn't fully aware of what signs and what were the things that we should be looking for. I guess the pediatrician had really

relied on the metabolic doctor to kind of look at things that we should be looking at." (Participant #6, Mother of a child with an IMD in Group 3 and 'large and sparse' network)
"I think it's the relationships that make them different. I don't get warm and fuzzy feelings with the man trying to do the, to do [son's] hearing aids. Um, the Eye Clinic, they, they're a bit cold too, right? Important, but we are just a person, we are just a number." (Participant #1, Mother of a child with an IMD in Group 3 and 'large and sparse' network)
"I definitely think that [online healthcare communication system], bringing that it was a really great resource because it saves me from explaining all of the times, that we have seen which pediatrician, which doctors, which specialists, what happened here. And you know, when they tell me what happened at the meeting, I only get like, maybe a snippet of what has actually happened. I don't get the full picture all of the time." (Participant #6, Mother of a child with an IMD in Group 3 and 'large and sparse' network)