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Families' healthcare experiences for children with inherited metabolic diseases: Protocol for a mixed methods cohort study

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ABSTRACT

Introduction Children with inherited metabolic diseases (IMDs) often have complex and intensive healthcare needs and their families face challenges in receiving high-quality, family centred health services. Improvement in care requires complex interventions involving multiple components and stakeholders, customised to specific care contexts. This study aims to comprehensively understand the healthcare experiences of children with IMDs and their families across Canada.

Methods and analysis A two-stage explanatory sequential mixed methods design will be used. *Stage 1:* quantitative data on healthcare networks and encounter experiences will be collected from 100 parent/guardians through a care map, 2 baseline questionnaires and 17 weekly diaries over 5–7 months. Care networks will be analysed using social network analysis. Relationships between demographic or clinical variables and ratings of healthcare experiences across a range of family centred care dimensions will be analysed using generalised linear regression. Other quantitative data related to family experiences and healthcare experiences will be summarised descriptively. Ongoing analysis of quantitative data and purposive, maximum variation sampling will inform sample selection for *stage 2:* a subset of stage 1 participants will participate in one-on-one videoconference interviews to elaborate on the quantitative data regarding care networks and healthcare experiences. Interview data will be analysed thematically. Qualitative and quantitative data will be merged during analysis to arrive at an enhanced understanding of care experiences. Quantitative and qualitative data will be combined and presented narratively using a weaving approach (jointly on a theme-by-theme basis) and visually in a side-by-side joint display.

Ethics and dissemination The study protocol and procedures were approved by the Children's Hospital of Eastern Ontario's Research Ethics Board, the University of Ottawa Research Ethics Board and the research ethics boards of each participating study centre. Findings will be published in peer-reviewed journals and presented at scientific conferences.

Strengths and limitations of this study

- This study will ascertain family perspectives on healthcare networks and positive and negative care experiences for children with high care needs, such as those with inherited metabolic disease, forming a comprehensive understanding of current care, including gaps in family centred care that will form the foundation for successful development of complex interventions to improve healthcare experiences for this understudied population.
- We expect this study to contribute to the methodological literature on assessment of healthcare experiences by using a novel combination of approaches, including care maps, diaries and interviews.
- This study exemplifies partnership with patients and their families in co-designing research towards improved healthcare.
- A limitation of this study is the requirement of English proficiency for study participation, which will exclude a potentially more vulnerable population of children and families who, for example, require language supports for their healthcare.

BACKGROUND

Inherited metabolic diseases (IMDs) are individually rare genetic conditions, often diagnosed in early childhood, that have a collective estimated global prevalence of 50.9 in 100 000 live births.[1] Many children with IMDs have complex and intensive healthcare needs.[2 3] Due in part to health service inequities related to infrastructure and funding, they and their families face multiple challenges in receiving high-quality care[4] and, in common with children with medical complexities generally, may not receive optimal interdisciplinary family centred services.[5 6]

Patient experience is a key pillar of a high performing health system.[7–9] Assessments of patient experience frequently address established principles of patient-centred care,[10] including access, coordination, continuity and communication.[8 11 12] In paediatrics, these principles extend to family centred care, emphasising children's developmental needs and recognising the central role of family members in disease management.[13 14] Families are often experts about the care needs of their children with rare diseases such as IMDs, underscoring the importance of their perspectives and their engagement in both healthcare and research.[5]

Several studies have focused on the quality of life and caregiving experiences of families of children with IMDs[15–25]; a smaller proportion have identified challenges or needs associated with providing and accessing care.[15–19] To begin to understand the healthcare experiences of this potentially underserved population, we completed two qualitative studies: first with representatives of relevant patient groups, then with caregivers of children with IMDs enrolled in a Canadian cohort study.[26 27] Overarching themes included a lack of familiarity with IMD care among many care providers outside of the metabolic clinic and poor suitability of some care systems to meet the needs of frequent and complex users. These studies expose a need for interventions that improve healthcare experiences of children with IMDs and their families. An Australian study found that families of children with IMDs experienced improved healthcare if care was accessed through a coordinating centre.[28] Guidance about family centred care for children with chronic conditions more generally suggests additional potential strategies for addressing some of these challenges, for example, co-developed care plans, receipt of care within a 'medical home', relational continuity with a key provider, improved collaboration between providers and increased family involvement.[5 13 14 29 30] These potential strategies reflect *complex interventions*: each single strategy would require multiple interacting components, targeting multiple individuals or systems and customisation to specific contexts of care, with potential impacts on a range of outcomes.[31] Guided by the UK Medical Research Council (UKMRC) Complex Interventions Framework,[32 33] we have planned a rigorous, four-phase research programme (online supplemental material 1) to develop complex interventions to improve family experiences with care. This protocol outlines our plans for 'phase I', the first study in our research programme, in which we seek to build on our previous qualitative studies to more fully understand and describe the 'problem'[26 27 33]: the nature, frequency, heterogeneity and impact of positive and negative healthcare experiences of children with IMDs and their families. Such a purpose requires both quantitative data that can be generalised to a larger population and qualitative data to understand the nuances of individual experiences and is thus well-suited to a mixed methods design.[34] Mixed methods designs have been used in several studies of patient or family experiences in paediatric healthcare.[35–41]

Objectives

The overall aim of this study is to comprehensively understand the healthcare experiences of children with IMDs and their families across Canada.

Quantitative objectives

- To identify and describe the providers and services included in children's care networks and how they are connected to both the family and to one another, from parents' perspectives.
- To prospectively measure the frequency, heterogeneity and satisfaction with healthcare encounters of children and their families.
- To identify the family characteristics and circumstances that form the context in which families experience healthcare, and their association with healthcare encounter satisfaction.

Qualitative objectives

- To explain and enhance our understanding of:
  a. parents' perceptions and assessments of their children's care networks;
  b. how families experience positive and negative healthcare encounters.

Mixed methods objectives

To merge the quantitative and qualitative findings to arrive at an enhanced understanding of:

- the nature of children's care networks and how they are experienced and assessed by parents;
- the family centred elements and processes related to parent perceptions of positive and negative healthcare encounters.

Pursuit of these objectives will be foundational to understanding how to develop complex, family centred care interventions. For example, identifying the constellation of providers and services and their roles and connections in children's care networks may enable us to identify key providers for healthcare coordination interventions (quant, qual). Knowing the most frequently used services will help with the prioritisation of intervention development and implementation (quant). Understanding which aspects of care contribute to negative and positive experiences will help inform the creation of responsive interventions (quant, qual). An understanding of family characteristics and situations will shape interventions that

account for the challenges and realities faced by families managing their child's care at home (quant).

The COVID-19 pandemic has exacerbated existing challenges related to access to care, and is expected to continue to affect how healthcare is delivered in the future. Therefore, we will collect data to understand the current context of healthcare delivery across Canada during the pandemic. In particular, we will aim to understand family experiences with virtual care, since this delivery modality has become more common due to pandemic response measures and the increase in its use is likely to influence healthcare delivery in a postpandemic environment.

METHODS

Study design

The UKMRC Complex Interventions Framework, a phased approach to the design, evaluation and implementation of complex interventions, guided this study's design.[32 33] Following previous studies of healthcare experiences,[42–46] we will also use the Picker Principles of Patient-Centred Care to provide a framework to guide data collection and analysis regarding key aspects of family centred care.[12]

We will conduct a mixed methods study, following a two-stage explanatory sequential design (figure 1).[34] *Stage 1*: quantitative data will be collected on parent perceptions of children's healthcare networks (the people involved in a child's healthcare and how they are connected) and on healthcare encounters (frequency, context, experiences with care). These data will be analysed on an ongoing basis to inform the sample selection for *stage 2*: two subsets of participants from stage 1 will participate in qualitative data collection (interviews) about (i) the participant's perception of the child's care network and/or (ii) the factors that contributed to a strongly positive or negative healthcare experience. At the individual level data collection will be sequential: the quantitative collection of data related to the child's care network and experiences will precede the qualitative collection of data related to the network or to a specific healthcare experience. Data from both stages will be integrated during analysis. We will

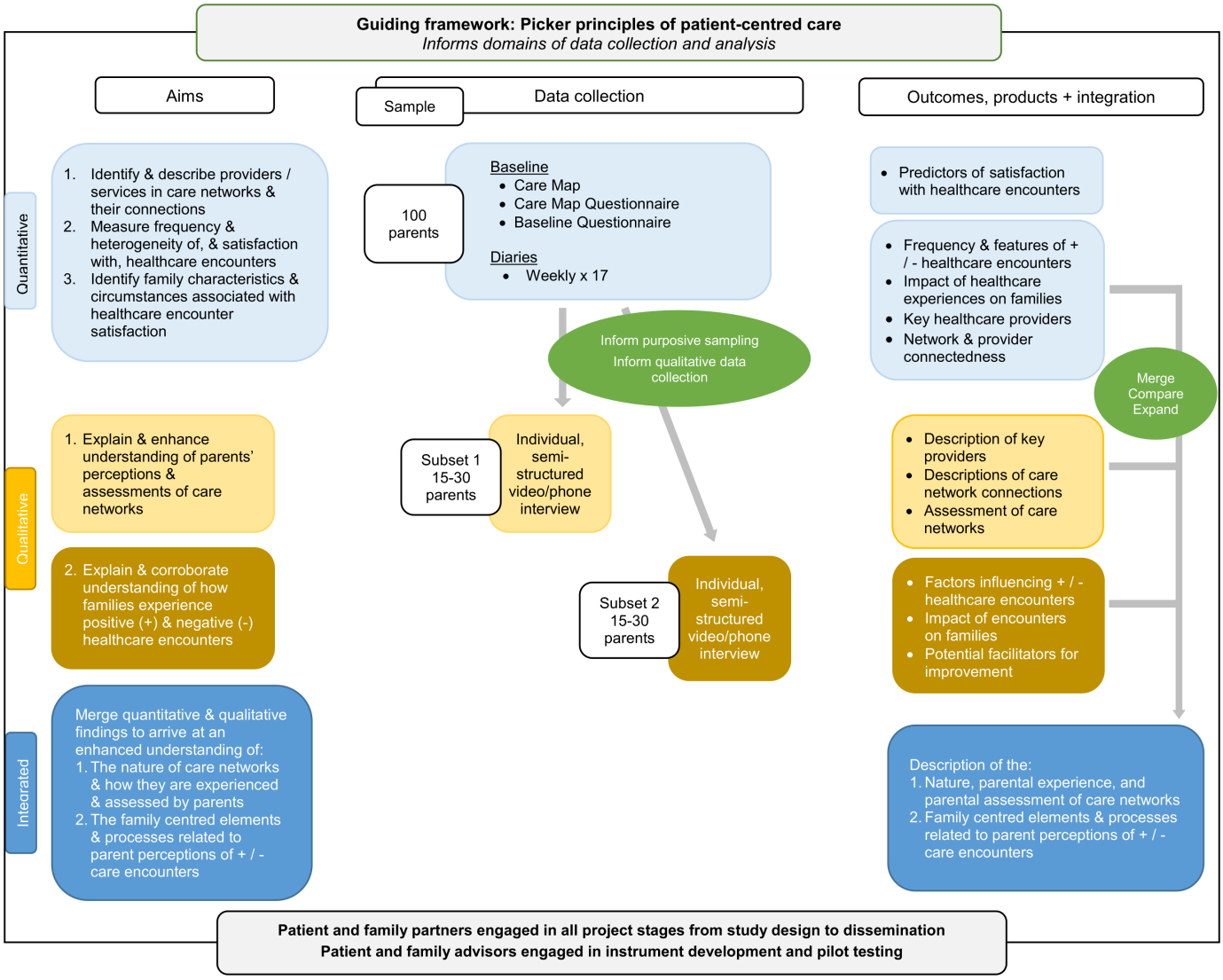

Figure 1 Study design overview: mixed methods explanatory sequential design.

use the STrengthening the Reporting of OBservational studies in Epidemiology guideline[47] to report the study (online supplemental material 2).

Patient and public involvement

The interventions informed by this study will be complex, involving diverse systems, providers and families, and aim to be family centred. This underscores a need to engage families and providers,[48 49] especially in the context of rare disease where families become experts in their children's care needs.[50] Parents of children with IMDs and adults living with IMDs are engaged in this study to provide expertise on the family/patient experience. Three family/patient partners (IJ, NP, MS) are study co-investigators, leading the family engagement strategy, advising and providing expertise, and sharing in decision-making at all study stages, from conceptualisation to dissemination. The study also engaged 11 patient/family advisors, recruited through IMD family advocacy and support organisations, to provide advice and feedback during study instrument development; 6 of them also pilot tested the data collection instruments.

Quantitative sample

Participants will be parents or legal guardians ('parents') of children diagnosed with an IMD. Although children's self-report of experiences is important, we seek to understand the experiences of healthcare for younger children (≤12 years). Parents are the family members most actively involved in seeking and managing healthcare for their children and thus are likely the best informants to provide comprehensive information on healthcare for this age group. For each participating family, one parent will be identified by the family as the 'designated parent' to provide data regarding one child in their family with an IMD ('designated child').

Eligibility criteria are described in table 1. Child age will be restricted to ≤12 years as adolescents with chronic conditions have different healthcare and clinical treatment needs.[51 52] With respect to eligibility of IMD diagnoses, >1000 IMDs have been identified.[53] IMDs typically follow one of three broad clinical course trajectories, with different implications for healthcare usage and experiences: (a) chronic and generally non-progressive; (b) acute episodes of severe illness with or without accompanying chronic multisystem sequelae and (c) progressive multisystem disease. Children with any of 30 priority IMDs included in an existing Canadian paediatric cohort study that will serve as one potential recruitment source[54 55] are eligible for this study (table 1). Few of the IMDs included in that cohort study, however, are characterised as following trajectory (c). Thus, children will also be eligible for this study if they have an IMD that meets clinical criteria associated with trajectory (c) (table 1), to be evaluated by clinician investigators on a case-by-case basis.

In order to collect data on healthcare experiences from a diverse sample of families, we will use a purposive, maximum variation sampling approach[56–58] to identify and recruit participants. We will aim for maximum variation on six selection variables on which experiences with care are anticipated to vary: study centre, travel time from home to study centre, child's sex, child's age (years), IMD type and IMD typical clinical course trajectory. Treatment protocols and healthcare service availability and practice vary by IMD, clinical course classification, study centre and/or distance to specialists.[27 59] Healthcare encounters tend to be more frequent in the first years following an IMD diagnosis (usually in infancy) and parents characterise this time as uncertain and stressful.[27] Sex differences can affect metabolism, resulting in different care experiences for girls and boys.[60 61] We will prioritise the selection of participants who expect the designated child to have ≥1 healthcare encounter per month during the study to collect sufficient data for analysis.

Quantitative procedures

Participants will be recruited from the existing cohort study and/or from the study centres across Canada. Eligible parents will be notified of the study by the study team (by telephone) or by their associated study centre (by telephone or at a clinic visit). For those notified by telephone, up to three contact attempts will be made. Participants will be enrolled on a rolling basis and the sample continually assessed for diversity on study selection variables to identify characteristics desired for further recruitment. Based on our previous experiences conducting studies with this population, we estimate a 50% response rate. Recruitment commenced in November 2020 and will be concluded when 100 families are enrolled. Interested parents will receive via email a postcard with study information and a link to the online Eligibility and Pre-Screening Questionnaire (5–10 min).

Data collection procedures are outlined in figure 1. All questionnaires will be web-based. Study data will be collected and managed using Research Electronic Data Capture hosted at the Children's Hospital of Eastern Ontario (CHEO).[62 63] The participant, if they desire, may consult other family members, including the designated child, to complete the data collection tools. Children will continue to access healthcare normally. Participants will be reminded up to two times to complete each questionnaire.

Quantitative data elements and instruments

Data collection instruments are described in table 2. Care map instructions, sample survey questions and measurements and interview guides are provided in online supplemental material 3. Instruments were developed with input from clinicians, methodological experts and family/patient partners and advisors and pilot tested.

Care maps

In this study, a 'care map' is a pictorial representation of the networks of healthcare providers around a child with

Table 1 Eligibility criteria

Inclusion	Exclusion
► The designated parent and designated child are Canadian residents ► The designated child is ≤12 years at prescreening ► The designated child is receiving healthcare from one of 11 participating paediatric metabolic clinics across Canada: Alberta's Children Hospital, British Columbia Children's Hospital, Children's Hospital of Eastern Ontario, Health Sciences Centre Winnipeg Children's Hospital, The Hospital for Sick Children, IWK Health Centre, Kingston General Hospital, London Health Sciences Centre, McMaster Children's Hospital, Montreal Children's Hospital, Stollery Children's Hospital ► The designated child has an IMD that is *either* 1. identified in the following list (these conditions were the focus of an existing cohort study; most have a typical clinical course that aligns with what we call trajectory a or trajectory b): – β-Ketothiolase deficiency – Arginase deficiency – Argininosuccinic aciduria – Carbamoyl phosphate synthetase deficiency – Carnitine uptake defect – Citrin deficiency – Citrullinemia – Farber disease – Galactosemia – Glycogen storage disease type 1 – Glutaric acidemia type I – Guanidinoacetate methyltransferase deficiency – HMG-CoA lyase deficiency – Homocystinuria – Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome – Isovaleric acidemia – Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency – Maple syrup urine disease – Medium chain acyl-CoA dehydrogenase deficiency – Methylmalonic acidemias – Mucopolysaccharidosis type I – Multiple carboxylase/biotinidase deficiency – N-acetylglutamate synthetase deficiency – Ornithine transcarbamylase deficiency – Phenylalanine hydroxylase deficiency – Propionic acidemia – Pyridoxine-dependent epilepsy – Trifunctional protein deficiency – Tyrosinemia type I – Very long-chain acyl-CoA dehydrogenase deficiency 2. *or* meets the following clinical criteria (included to expand the list of eligible conditions and to increase representation of IMDs with a typical clinical course that aligns with what call trajectory c): – involves at least three organ systems *and* – chronic complications of the disease get progressively worse over time, even with available treatment	Designated parents who cannot speak, write and read English comfortably

HMG-CoA, 3-hydroxy-3-methylglutaryl coenzyme A; IMD, inherited metabolic disease.

an IMD and their family, commonly used in research on children with complex or chronic health conditions.[5 64–66] Guided by a set of instructions,[67] care maps will be drawn by hand, photographed and uploaded to the study data collection database by the participant, and a digital version rendered by the study team.

Baseline questionnaires

Participants will be invited to complete three questionnaires: the Care Map Questionnaire, the Baseline Questionnaire and the Pre-Questionnaire for the Weekly Logs (content overview, table 2). The Baseline Questionnaire includes a number of validated instruments. Child health

Table 2 Data collection instruments

Data collection period Data instrument	Data type	Instrument completion time* (min)	Instrument and data details
Baseline			
Care map	Quantitative	40	Participant creation of a care map of their perceptions regarding their child's network of care providers, which providers are perceived to work together to coordinate their child's care, and which providers are considered 'key providers' (maximum 10).
Care Map Questionnaire	Quantitative	5	Participant perceptions about: ► Coordination of their child's care. ► Familiarity with their child by identified key healthcare providers.
Baseline Questionnaire	Quantitative	20–40	Demographics and potential predictors of healthcare encounter satisfaction ratings, for example, child health status, child and family characteristics, family resources in IMD management and effects of the COVID-19 pandemic on child health and healthcare since March 2020.
Pre-Questionnaire for the Weekly Logs	Quantitative	5–20	Data will be used to tailor the healthcare diaries, to reduce repetition of questions where responses are anticipated to remain constant over the study period.
Follow-up			
Healthcare diaries†	Quantitative, qualitative	5–60	Descriptive data on healthcare encounters including: the mode of interaction, the care setting if applicable, the healthcare providers involved, the date of the encounter, financial costs, time inputs and any parent-perceived effects of the COVID-19 pandemic (eg, on scheduling or delivery of care). Optional, open-ended questions for descriptions of participant perceptions of care in each Picker Principle domain, and for the overall encounter. The Experience Questionnaire will be tailored to each encounter's mode of interaction (in-person or virtual/remote), care setting and context (planned or urgent care; whether it is a 'frequent' care encounter, as identified on the Pre-Questionnaire for the Weekly Logs).
Interviews	Qualitative	(a) 30–60 (b) 30–45	(a) Map interviews: seek to understand and elaborate on the care map, including how the participant selected providers to include on the map, the roles and relationships with the family for the providers designated on the map as 'key providers', the meaning of connections drawn between providers and how the participant feels about the effectiveness of the care network, including what improvements they see as potentially important. (b) Encounter interviews: seek to clarify, interpret and deepen our understanding of information collected in the healthcare diaries, specifically: elements of a healthcare encounter that contributed to participants' high or low satisfaction with that encounter; the impact of these experiences, especially the challenges, on the child, parent, other family members and the context of general healthcare for their child (ie, comparison between this encounter and past similar encounters). Impact will be iteratively defined, depending on the information shared by participants, and may include psychosocial, health and/or economic impacts.

*Estimated.
†All elements are completed once except the healthcare diaries, which are completed weekly ×17 weeks.

status will be assessed using the Child Health Questionnaire - Parent Form 50[68] for children ≥5 years or the Infant and Toddler Quality of Life Questionnaire-47-item short-form[69] for children <5 years. Both are parent-reported measures and have good validity and reliability.[69–71] Parent-perceived quality of life related to caring for the designated child will be measured using the CarerQol instrument. The CarerQol has good psychometric properties[72–75] and has been used with parents of children with chronic conditions, including rare diseases.[76–79] We reformatted the measure for online use.

Healthcare diaries

The Healthcare Diary ('Diary') is composed of two parts: a Healthcare Log and Experience Questionnaire. Once per week, participants will record whether a child had any healthcare encounters in a given week on the Healthcare Log. If yes, they will complete an Experience Questionnaire for each of those encounters. Diary methods have been used in health studies to capture real-time information to reduce the recall errors associated with retrospective surveys,[80 81] with electronic diaries yielding higher quality data than paper diaries.[82 83] The definition

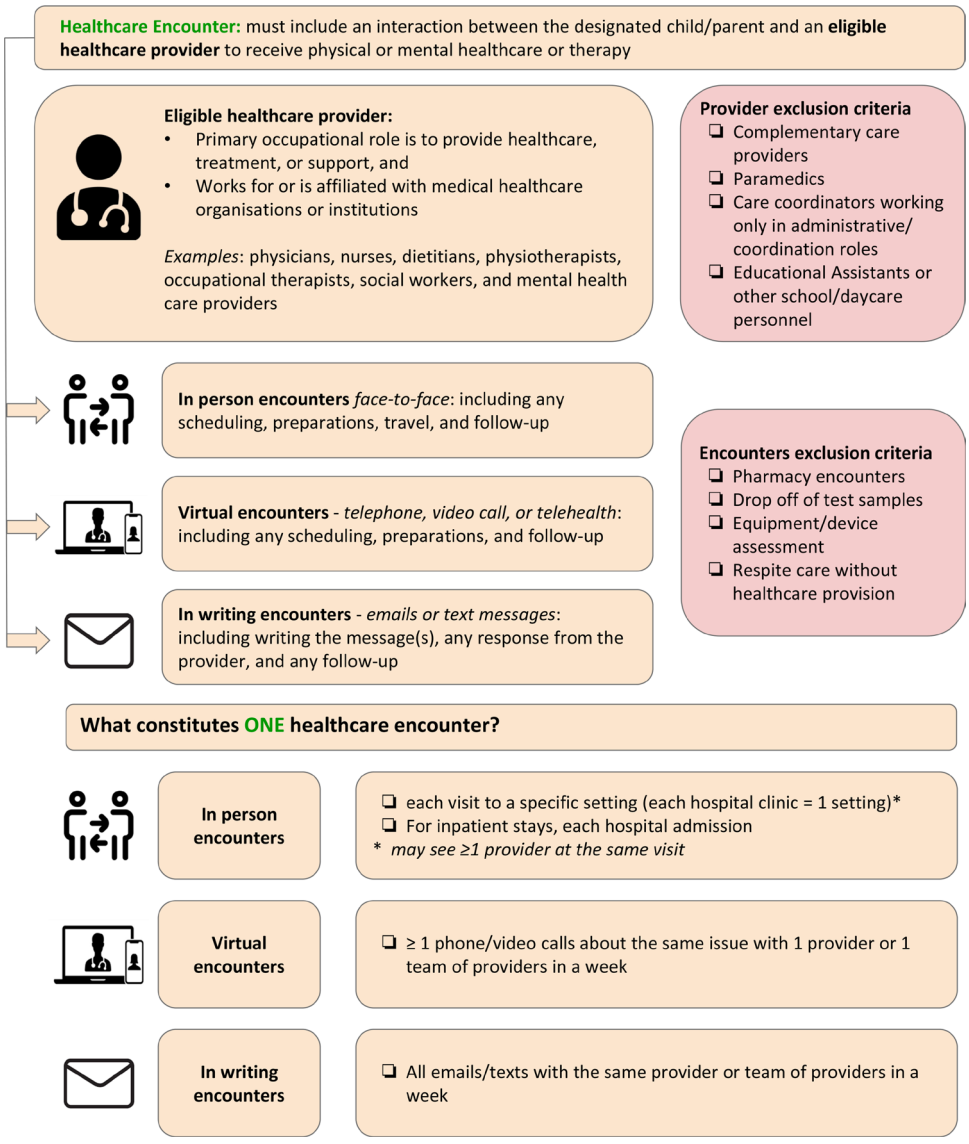

Figure 2 Healthcare encounter definitions/eligibility.

of a healthcare encounter is provided in figure 2. Evaluations will be made for the overall experience as well as in eight domains consistent with the Picker Principles of Patient-Centred Care where applicable[84]: access to care, information sharing/communication, care coordination, physical comfort, emotional support, family involvement, respect for the patient/family and continuity/follow-up. The Consumer Assessment of Healthcare Providers and Systems Child Hospital Survey,[85] Ontario Emergency Department Patient Experience of Care Survey,[86] Outpatient Survey (Christine Kouri, Manager for Patient Experience, CHEO, email communication, October 2017) and the Cost Utilisation Survey for Child Phenylketonuria[87] were used as resources for our diary instrument development; diary questions were either author-developed, informed by or adapted from these resources.

We will collect prospective data on blood draws done at home by the family, following the same family centred care domains. For many IMDs, blood draws are essential to the ongoing monitoring of a child's health status, and although sometimes conducted by the family, require an ongoing dialogue with healthcare providers to adjust a child's medication, diet or other treatment.

Qualitative sample

The two qualitative samples will be nested in the quantitative sample. Qualitative participants queried about their children's care networks must have completed the Care Map Questionnaire, and those queried about their positive or negative encounters must have completed at least four diaries. For the interview focused on the healthcare encounter ('encounter interview'), we will select participants who have had a healthcare encounter with which they reported they were 'extremely satisfied', 'extremely dissatisfied' or 'somewhat dissatisfied' overall or on at least one family centred care domain. We will use purposive, maximum variation sampling and extreme case sampling to separately sample participants for each interview set,[56–58] aiming for sample variation across the selection variables used for the quantitative sample and across

healthcare settings in the encounter interviews. For the encounter interviews, if the parent who accompanied the child to the encounter is not the designated parent, they will be invited but asked to provide informed consent before proceeding. Some participants in the quantitative sample may be invited to participate in both interviews.

Qualitative procedures and data elements

On a rolling basis, participants will be identified and invited by email to participate in a one-on-one, semi-structured interview held by videoconference or by audio-conference, according to participant preference. For the interview focused on care network ('map interviews'), participants may be sampled at any time after completing the Care Map Questionnaire. For the encounter interviews, participants will be sampled during and up to 3 weeks after completing week 17 of the diaries. Interviews will be audio-recorded with participant consent and transcribed. Up to three attempts to contact participants will be made to invite interview participation. Both interview sets will be semi-structured and informed by an interview guide.

Sample size

While we did not conduct a formal power calculation for the quantitative part of this study, given our largely descriptive purpose, we deemed a sample size of 100 families sufficiently large to support planned analyses across a heterogeneous sample, while maintaining feasibility for recruitment and study administration.

Because of the duration and intensity of study participation, we anticipate some dropout. Dropout rates may increase with longer study lengths.[88 89] To facilitate participant retention, we pilot tested the feasibility of study questionnaires. In addition, we will: (1) enrol a new participant to replace any participant withdrawn before completion of at least four diaries; (2) actively monitor completion of study instruments and follow-up with participants if necessary; (3) provide participants with financial compensation (up to CA$400 in gift cards) for their time and as a participation incentive[90]; (4) encourage the scheduling of time each week to complete the diaries; (5) allow for instrument completion over multiple sittings and (6) allow for flexibility of instrument completion.

A participant will be considered lost to follow-up on notification of withdrawal or non-completion of an instrument within prespecified timeframes; they will have the option to continue in the study if they proactively express a desire to do so. Data collected up to time of withdrawal will be included in the study.

The qualitative sample sizes will not be determined in advance; they will be assessed continuously and finalised during data collection. Information power is a methodological model for determining a qualitative sample size, and has five contributing dimensions related to: narrow versus broad qualitative objectives; the homogeneity of the sample on important characteristics; use of a theoretical framework; quality of interview data and planned analytic strategy (case vs cross-case analysis).[91] Based on this concept and previous qualitative studies with parents of children with chronic conditions,[26 92–94] we anticipate a sample size of approximately 15–30 participants for each interview set.

Analyses

Quantitative analyses

We will describe continuous variables using means and SD or medians and IQRs, and categorical variables using counts and proportions (%). Baseline data will be analysed to describe the characteristics of participating families, including child and parent demographic variables, quality of life, experiences with managing an IMD in the context of COVID-19 and experiences with managing an IMD in general, including time and cost impacts.

From the care maps, children's networks of care providers and their interactions will be analysed using an adapted form of social network analysis,[95 96] conducted using UCINET software.[97] We will describe who is in the network (nodes), identify the most common providers perceived as key providers and analyse connections among providers from parents' perspectives (social network analysis calculations of network size and density and the degree centrality of providers).

From the diaries, we will calculate the frequency (count and rate) of encounters by participant/child, accounting for follow-up time contributed. We will calculate counts and proportions to describe characteristics of healthcare encounters (eg, preplanned vs unplanned, the types of healthcare providers interacted with, care setting or mode of interaction), overall satisfaction ratings and satisfaction ratings by Picker Principles (access to care, communication, coordination of care, etc).

To explore the potential relationships between a range of explanatory variables and satisfaction with healthcare experiences, depending on data quantity and distribution, we propose to use generalised linear regression analysis. The unit of analysis will be the individual healthcare encounter with each child having potentially different numbers of encounters. Explanatory variables will include both time-fixed and time-varying factors, namely child, family and setting/provider characteristics (eg, child age, IMD clinical course trajectory, travel time from home to care setting, socioeconomic status), healthcare setting and mode of interaction. The five-point ordinal score for the overall experience of the healthcare encounter will be analysed using ordinal logistic regression. Correlation in repeated measures on the same child will be accommodated either by directly modelling the covariance matrix or through the addition of child-specific random effects. A similar approach will be used to analyse the experience ratings within the eight family centred care domains.

This study will minimise missing data by regularly monitoring completion of instruments and diary entries and following up with participants as necessary. Participants will have access to ongoing support from the study team. We will report on the number of missing values for

each variable of interest, the reasons for missing values (if known), characteristics of participants with missing versus non-missing values for key variables and missing data counts for each analysis. Our analytic strategy for managing missing data will depend on the extent of missingness of data for particular analyses and may rely on complete case analysis or multiple imputation. Withdrawn participants will be considered lost to follow-up at the date of their last completed baseline instrument or Healthcare Diary.

Qualitative analyses

Guided by principles of family centred care and incorporating an inductive approach, we will use thematic analysis[98] to guide the coding and analysis of qualitative data across participants, using the following recommended process: (1) review the interview transcripts and familiarise themselves with the data; (2) do an initial, systematic coding of the data; (3) identify themes of codes; (4) review the generated themes against both the initial codes and the original data; (5) refine the themes and (6) select and review extracts to illustrate the themes. We will repeatedly cycle through steps, particularly steps 3–4, to ensure the themes remain reflective of the original data.[98]

Mixed methods integration and analysis

The two types of data will be integrated at several points in the study. The quantitative data will be used to inform the qualitative sample as well as the interview questions and topics. We will compare the qualitative sample with the quantitative sample on the quantitative sampling selection variables. The quantitative and qualitative results will be merged in analysis and integrated to better understand the elements and processes related to healthcare networks and to positive or adverse healthcare encounters than would be gained from either data type alone.[34] In the final report, the qualitative and quantitative results will be integrated narratively using a weaving approach[99] (reported together, grouped by theme or concept) and presented visually in a side-by-side joint display with interpretations of the combined results and inferences about the meaning of the integrated data.[100]

DISCUSSION

This study will collect important information about parent perceptions about their families' experiences with healthcare for children with IMDs, a population with complex needs. Few paediatric studies have attempted to collect similarly comprehensive data on healthcare experiences.[40] Previous studies of children with IMDs and their families have focused on the impact of the IMD on the child, caregiver or family well-being[16–22 25] and/or on family experiences managing healthcare.[15 23 25] To our knowledge, this is the first broad study of healthcare experiences in paediatric IMDs. We have designed a study that draws on mixed methods that best suit the research objectives, enabling the collection of experiential information of both breadth and depth. Diaries are an innovative tool in health research with potential for collecting real-time quantitative and qualitative data simultaneously. Care maps provide useful insight on how participants conceive of the networks of care around their children.

The main findings of this study will inform future phases of our research programme, culminating in the co-development of family centred interventions to improve healthcare for children with IMDs and their families. Comprehensive, prospective information collected on individual healthcare experiences will help elucidate the elements of healthcare that contribute to caregivers' negative and positive experiences. This information will also enable an assessment of the degree to which healthcare experiences are family centred, ultimately helping to inform the creation of responsive interventions, especially for highly frequented services. Care map data will identify key providers and enable an understanding of how participants perceive providers to be connected to each other and to the family. This may help to identify key providers who may be able to lead a child's 'medical home', playing an active role in coordinating their healthcare. Knowledge about the time, financial costs and other inputs required to care for a child with an IMD is necessary to ensure that interventions are responsive to the realities of families for whom the interventions are designed to support. Data captured on healthcare experiences during the COVID-19 pandemic will contribute important information on the benefits and drawbacks of significant changes to healthcare delivery, such as virtual healthcare, which can improve the way that this care is delivered in the future. Through our larger research programme, the evidence generated in this study will have a direct, actionable impact on family centred healthcare for paediatric IMDs.

This study has limitations. All study data will be sought from parents. Their perceptions of their child's healthcare, for example, whether or not two providers work together to coordinate their child's care, may differ from providers' perceptions. However, healthcare providers will be interviewed about their perceived barriers to and facilitators of effective healthcare for children with IMDs in the next phase of the research programme. Requiring English proficiency for study participation will limit the generalisability of study findings and will exclude a potentially more vulnerable population of children and families who, for example, require access to translators and additional supports as part of their care.

This study may be affected by selection and information biases. We will prioritise the selection of participants who expect the designated child to have multiple healthcare encounters during the study; our quantitative sample will be over-representative of families who are frequent healthcare users. This characteristic of our anticipated sample will increase the number of prospective healthcare experiences reported; however, it may limit the generalisability of quantitative findings. Although Canada has a publicly funded healthcare system, access to all care and services is

not equitable.[101] A higher frequency of encounters may indicate greater access to care. Children with fewer expected encounters will still be enrolled in the study, and access to care (unavailable services, out-of-pocket expenses) will be analysed. Past positive or negative experiences with care may motivate parents to participate in a study that provides the opportunity to share those problems and experiences. Non-response bias has been associated with both high and low patient satisfaction.[102 103] Parents whose children are experiencing urgent or critical healthcare issues, whose children are newly diagnosed (often associated with younger age) or who experience significant financial and time costs may feel overwhelmed and be less likely to participate or remain in the study than parents whose children's health issues are relatively stable.[26 104] We will attempt to minimise the burden of study participation by employing web-based data collection and offering compensation for study participation. To ensure that lack of home internet access is not a barrier to study participation, participants may be loaned a study tablet with a mobile data plan to participate in the study. We anticipate that this may affect 10–15 participants.[105]

Factors such as recall and negativity bias may affect the reporting of all healthcare encounters. Our collection of prospective data via diaries, however, aims to capture experiences during all healthcare encounters, positive and negative, with a high frequency of reporting to mitigate errors associated with recall time.[81 106] The perspectives of the interviewer and data analysts may affect the collection and analysis of qualitative data. Interviewers will be trained by investigators with expertise in qualitative interviewing. Interviews will be transcribed as soon as possible after interviews and reviewed.

ETHICS AND DISSEMINATION

The study protocol and procedures were approved by associated research ethics boards (online supplemental material 4). Participants will provide informed consent. Study data will be analysed and stored securely.

Study findings will be published in peer-reviewed, open access journals and presented at relevant conferences. Additionally, a summary of study results will be shared with interested participants (opt-in). Study results will also inform future phases of our research to develop interventions to improve family centred healthcare for this population.

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