

Exploring the experiences of family caregivers with low income accessing health care services for children with inborn errors of metabolism

by

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Abstract

Inborn errors of metabolism (IEMs) have been shown to impact the wellbeing of family caregivers, with more challenges experienced by families with lower income. The main objective of this study was to gain understanding of the experiences of family caregivers with low income accessing diagnosis-related care for their children with IEMs in Manitoba. Secondary objectives included: (1) To explore the factors that facilitate or cause difficulties accessing IEM-related care, (2) To explore the perceived impact of the COVID-19 pandemic on family caregivers' experiences of accessing IEM-related care, and (3) To compare the experiences of accessing IEM-related care for urban and rural families. This mixed methods study involved the distribution of a survey to 98 family caregivers of children with IEMs who access care in Manitoba. Additionally, we conducted eight semi-structured individual interviews with survey respondents to further explore the experiences of these family caregivers. These qualitative interviews were transcribed, coded, and analyzed to identify themes and make recommendations to improve access to care. Twenty-five surveys were returned, and descriptive statistics are presented on the experiences of these study participants. Additionally, qualitative analysis revealed that participants found caregiving was overwhelming and consuming especially throughout the first year following a positive newborn screen. Participants expressed that they were lacking psychosocial support as well as assistance with navigating financial resources. Participants also recognized many costs of care, shared experiences outlining difficulties in accessing care, and described changes in care due to the COVID-19 pandemic. This research reveals the unmet needs of family caregivers of children with IEMs in Manitoba, especially in the psychosocial and financial domains. Study findings will enable genetic counsellors to advocate for equitable access to metabolic care in Manitoba and beyond.

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List of Abbreviations

COVID-19	Coronavirus disease 2019
IEM	Inborn error of metabolism
MCADD	Medium chain acyl-CoA dehydrogenase deficiency
NBS	Newborn screening
PKU	Phenylketonuria
RAC	Research Advisory Council

Chapter 1: Introduction

1.1 Inborn Errors of Metabolism

Inborn errors of metabolism (IEMs) are a diverse group of more than 1000 genetic disorders resulting in reduced activity of an enzyme, structural protein, or transporter molecule in a metabolic pathway (Tingley et al., 2020; Waters et al., 2018). Although these disorders can present at any age including in utero, they are often diagnosed in the neonatal period or infancy and often require complex and resource-intensive medical care as well as have intense home management and caregiving needs (Guerrero et al., 2018; Pugliese et al., 2020). Each specific disease is rare, but collectively IEMs are common, affecting up to 1 in 800 newborns (Hartley et al., 2011; Wang et al., 2014). More recently, Waters and colleagues (2018) estimated the global birth prevalence of all IEMs to be 50.9 per 100,000 live births and suggested that IEMs are an important cause of morbidity and mortality for children under the age of 5 years.

Most IEMs are caused by a block in a metabolic pathway due to reduced or absent activity of a specific enzyme resulting in insufficient or absent conversion of substrate to product (Guerrero et al., 2018; Vernon, 2015). This blocked metabolic step leads to a build-up of substrate, deficiency of product, and secondary metabolic abnormalities and thus can lead to an increase in the production and accumulation of potentially harmful metabolites (Guerrero et al., 2018). IEMs can be categorized into eight main classes based on which metabolic pathway is blocked: amino acid disorders, organic acid disorders, fatty acid disorders, lysosomal storage disorders, carbohydrate metabolism disorders, urea cycle disorders, mitochondrial disorders, and peroxisomal disorders (Waters et al., 2018). The two most common IEMs are phenylketonuria (PKU), an amino acid disorder with a global birth prevalence of 6.55 per 100,000 live births, and medium chain acyl-CoA dehydrogenase deficiency (MCADD), a fatty acid oxidation disorder

with a global birth prevalence of 5.78 per 100,000 live births (Lamoureux et al., 2015; Pugliese et al., 2020; Waters et al., 2018). Therapies for IEMs, such as reduction of toxic metabolites through dietary restriction of enzyme precursors, increased disposal of toxic metabolites, or enzyme replacement therapy, are targeted at correcting the biochemical abnormalities caused by an enzymatic block (Vernon, 2015).

IEMs have a wide variety of clinical presentations, ranging from childhood developmental delay to acute metabolic decompensation in the neonatal period, and can share broad, non-specific features, including gastrointestinal, neurological and endocrine problems, with other more common diseases (Arpaci et al., 2020; Guerrero et al., 2018; Waters et al., 2018). Some IEMs may be mild and require minimal to no intervention, whereas others are more severe (Potter et al., 2013). Most IEMs are treatable, however without early identification and treatment, IEMs can lead to severe infant outcomes including irreversible neurologic injury or death within days after birth (Potter et al., 2013; Wang et al., 2014; Waters et al., 2018). Possible triggers of metabolic decompensation in a newborn commonly include breastfeeding, extended periods of fasting, or an intercurrent illness (Dinchong, 2019). Advancements in newborn screening (NBS) has resulted in earlier identification of newborns with IEMs which has allowed medical interventions prior to symptom onset that can positively alter the course of the condition, increase life expectancy, and dramatically improve outcomes in many IEMs such as PKU and MCADD (Tingley et al., 2020; Vernon, 2015; Wang et al., 2014). However, some infants can experience severe and potentially fatal events in the early neonatal period prior to the receipt of NBS results (Karaceper et al., 2019). Additionally, rarer IEMs are often not included as part of routine NBS and thus rely on a high degree of clinical suspicion upon symptomatic presentation

or acute illness as well as up-to-date laboratory techniques to diagnose or they can be missed (Waters et al., 2018).

There are a number of consensus guidelines in the literature that outline recommendations for providing care to children with IEMs (Potter et al., 2013). While some of these conditions can be successfully managed with dietary changes, medication, and strict care plans, many of these conditions can still have serious complications despite appropriate treatment and may be life-threatening (Hartley et al., 2011). Frequent follow-up visits to speciality clinics, blood sample collection for biochemical monitoring, multiple hospital admissions, gastrostomy tube feeding, placement of central lines, and various other invasive procedures are commonly required (Hartley et al., 2011; Rajasekar et al., 2020; Zeltner et al., 2016). A study by Wang and colleagues in 2014 reported that approximately 41% of children with IEMs experienced emergency department visits and/or inpatient hospital admissions during their first 3 years of life. The hospital utilization rates for these children were found to be affected by social factors, including more frequent acute care hospital utilization for those who received government-subsidized healthcare such as Medicaid as well as for children of young mothers (Wang et al., 2014). This is consistent with other findings in the literature that highlight the influence of social factors on outcomes of IEMs, whereas associations with genotype and biochemical phenotype are often weaker than predicted (Potter et al., 2013). These interactions between social factors and outcomes can lead to challenges for clinicians' in predicting prognoses for children with IEMs (Potter et al., 2013).

1.2 Family Caregiving for Children with IEMs

Despite improved outcomes for children with IEMs due to advancements in NBS, many family caregivers caring for a child with an IEM report encountering substantial challenges for

the entire family (Cederbaum et al., 2001; Gramer et al., 2014; Piercy et al., 2017). These challenges can impact the wellbeing of the family, not only physically and psychologically but also with regard to financial status and relationships with their community (Cederbaum et al., 2001). Family caregivers must coordinate and manage the life-long demands of care for their child while coping with multiple sources of stress, including the threat of metabolic decompensation, financial burden, and lack of information available due to the rarity of the condition (Cederbaum et al., 2001; Packman et al., 2007). However, family caregivers may be reluctant to express this burden due to their close emotional attachment with their children (Arpaci et al., 2020). Ultimately, these challenges combined with a lack of support to meet psychosocial needs can hinder family caregivers' adaptation to their child's diagnosis which can lead to poorer outcomes for children with IEMs (Hassall et al., 2022).

1.2.1 Initial Experiences Following Positive NBS & Diagnosis

Previous studies have shown that family caregivers initially experience a variety of emotional reactions following positive NBS and their child's diagnosis of an IEM. These emotions commonly include shock, disbelief, fear, anxiety, guilt, and sadness (Carpenter et al., 2018; Hartley et al., 2011; Packman et al., 2007). Family caregivers were more likely to have intensified reactions especially if they were previously told by health care providers that their newborn was unlikely to have a positive NBS or if they had previous children who had negative NBS (Carpenter et al., 2018). Although NBS leads to improved outcomes of children with IEMs, family caregivers still report substantial burden from the condition and its management (Gramer et al., 2014). Additionally, family caregivers of children with IEMs have reported to be overwhelmed by the perceived pressure of health care professionals' expectations for them to immediately take responsibility for their child's management and achieve metabolic control,

despite their initial emotional shock and distress following diagnosis (Carpenter et al., 2018). Especially within the first few months following diagnosis, family caregivers are faced with uncertainty about their child's prognosis and the threat that IEMs can pose to their child's development, especially for those with neurological manifestations (Fidika et al., 2013; Khangura et al., 2016). Family caregivers may wonder whether their child's IEM will improve or worsen over time, even considering the possibility of their child's early mortality as often as daily (Cederbaum et al., 2001; Khangura et al., 2016; Packman et al., 2007). While family caregivers are provided education about management and psychosocial support by genetic counsellors at this time, the information is often lost due to these intense emotional states (Hartley et al., 2011).

Gramer and colleagues (2014) also found that family caregivers' initial perceptions of their child's IEM diagnosis were not necessarily associated with the severity of the IEM. For example, family caregivers who had undergone traumatic birth events or who had little time to address their initial feelings due to being thrust into learning how to manage their child's IEM experienced more intense grief-like reactions such as denial, anger, and depression (Carpenter et al., 2018). On the other hand, family caregivers who engaged in emotion-centred coping strategies following the diagnosis were more able to resolve their grief and begin accepting their child's IEM (Carpenter et al., 2018). Thus, Carpenter and colleagues (2018) suggest that family caregivers should be given time to process their emotions about their child's IEM by discussing with and having these emotions validated by professionals such as clinical psychologists, genetic counsellors, or specialist nurses.

1.2.2 Managing Diet & Coordinating IEM-Related Care

Family caregivers act as the primary carer and main point of contact when collaborating with the metabolic team to manage their child's IEM (Rajasekar et al., 2020). Even family caregivers who had enough time to process their emotions regarding their child's diagnosis face challenges and experience substantial stress when navigating health care systems and adjusting to the demands of complex disease management (Rajasekar et al., 2020; Siddiq et al., 2016). For family caregivers providing care to a child requiring specific and lifelong dietary management, they must constantly plan, monitor, and measure all daily nutritional intake of their child (Arpaci et al., 2019; Fidika et al., 2013; Shaji Thomas et al., 2021; Siddiq et al., 2016). Additionally, there may be additional management requirements when their child is ill or otherwise metabolically unstable (Cederbaum et al., 2001). Children must also be taken for numerous medical appointments, including frequent blood draws for biochemical monitoring and time-consuming follow up visits with the metabolic team, especially those at increased risk of metabolic decompensation (Arpaci et al., 2019; Cederbaum et al., 2001; Rajasekar et al., 2020; Shaji Thomas et al., 2021; Siddiq et al., 2016). In order to accommodate this immense level of care, some family caregivers take a leave of absence from their occupation and sacrifice career opportunities (Hassall et al., 2022; Siddiq et al., 2016). Rajasekar and colleagues (2020) report that while family caregivers gain expertise in managing their child's condition over time, they often feel unsupported and undervalued in these management roles. Often, this family caregiving role is primarily imposed on mothers rather than on fathers or other family members (Arpaci et al., 2019; Hassall et al., 2022).

Of the challenges associated with their child's IEM-related care, family caregivers often rank the management of their diet as the greatest source of stress (Abdelaziz et al., 2019).

Especially in infancy, family caregivers experience substantial stress associated with ensuring their child obtains adequate energy intake from regular feeds (Piercy et al., 2017). For example, for children with MCADD, the maximum length of fasting during the first four months of life is limited to four hours, even throughout the night (Piercy et al., 2017). Additionally, intervals must be adjusted during periods of illness to follow strict emergency feeding plans (Piercy et al., 2017). These intervals of fasting can be gradually increased with age, for example once children reach one year of age, they can go up to twelve hours without food, however parents often found these transitions to extended periods of fasting extremely anxiety-inducing and often continued night feeds longer than recommended (Piercy et al., 2017). This management of frequent care needs is both time-consuming and physically-exhausting, especially when family caregivers' sleep schedules are disturbed, and can often result in social losses due to these constraints (Hassall et al., 2022). As children with IEMs age and dietary management becomes routine, the anxiety associated with this management lessens (Piercy et al., 2017).

Another critical aspect of dietary management for children with IEMs when restricting certain types of foods is the supplementation of vital nutrients with medical foods (Berry et al., 2020). Medical foods are products that are designed to supplement the diets of those with limited abilities to metabolize all nutrients in ordinary foods (Berry et al., 2020). These products, which come in many forms including powders, drinks, or tablets, typically supply all essential nutrients except the restricted nutrient, such as amino acid formulas (Berry et al., 2020). Medical products may also come in the form of foods modified to be low in the restricted nutrients, such as pasta, baked goods, or other food substitutes (Berry et al., 2020). For some children with IEMs, lack of access to these medical foods can lead to growth failure and metabolic decompensation, and the reduced palatability of some options as well as time and financial restrictions may be barriers to

optimal dietary adherence (Berry et al., 2020; Lea et al., 2019). For example, Rose and colleagues (2019) reported that the average annual cost of medical foods for children with PKU was over \$4000 for medical formulas and almost \$2000 for low-protein food substitutes. Additionally, some family caregivers reportedly spend more than 300 hours per year purchasing medical foods and preparing special diet foods for children with IEMs (Rose et al., 2019).

As dietary therapy is highly restrictive and adherence is critical to preventing adverse outcomes, family caregivers often assume full responsibility for managing this care rather than delegating responsibility to others (Carpenter et al., 2018; Fidika et al., 2013). Family caregivers' were limited in the amount of support available due to fear of relinquishing control and allowing others to participate in dietary management (Carpenter et al., 2018). Family caregivers who do share responsibility in managing their child's diet with others, such as family members or other childcare providers, must continuously liaise with and inform them of appropriate dietary management (Shaji Thomas et al., 2021). This support can be both a source of relief and anxiety depending on the level of perceived competence others have to manage the child's care, especially as friends, family members, and the community at large frequently have a lack of knowledge and misconceptions about children with IEMs (Carpenter et al., 2018). In some cases, family caregivers receive assistance in educating others from health care providers such as dietitians, who are better suited to present information in a calm and balanced fashion (Piercy et al., 2017).

Family caregivers also experience stress when health care providers have a lack of knowledge and understanding of IEMs (Packman et al., 2007). Some family caregivers experience difficulties accessing phlebotomy services, including availability of highly trained professionals and inconvenient scheduling (Siddiq et al., 2016). Additional stress can result from

health care staff delaying treatment due to being entirely unfamiliar with the critical nature of these conditions as well as disregard of the family caregiver's expertise, especially in urgent situations when even emergency protocol letters may be ignored (Cederbaum et al., 2001; Siddiq et al., 2016). Culture, employment status, and access to interpreter services can also impact family caregivers' overall experiences accessing metabolic-related care (Rajasekar et al., 2020).

Despite these many challenges in coordinating IEM-related care and managing their child's diet, family caregivers eventually adjust and the complexities of care become a part of normal, daily life for the family (Siddiq et al., 2016).

1.2.3 Impact of IEMs on Family Caregivers' Wellbeing

Family caregivers have previously reported the impact of IEMs on the family's physical, emotional, social, and financial wellness (Gramer et al., 2014; Rajasekar et al., 2020; Siddiq et al., 2016; Weber et al., 2012; Zeltner et al., 2016). In fact, family caregivers of children with IEMs have not only been found to have lower quality of life compared to family caregivers of healthy children, but also compared to family caregivers of children with diabetes, leukemia, end-stage renal disease, and Duchenne muscular dystrophy (Fabre et al., 2013; Hassall et al., 2022; Hatzmann et al., 2009; Ten Hoedt et al., 2011). Numerous factors related to family caregivers themselves and their children with IEMs have been associated with decreased family caregiver quality of life. Predictors of increased family caregiver stress also include the child having a younger age, decreased developmental level, or severe neurocognitive impairment (Abdelaziz et al., 2019; Hatzmann et al., 2009; Shaji Thomas et al., 2021; Ten Hoedt et al., 2011). Furthermore, family caregivers caring for children with greater dietary constraints also report to experience a poorer quality of life (Shaji Thomas et al., 2021). Additional challenges have been associated with IEMs that require frequent emergency care despite treatment as well

as the financial burden they pose when compounded with lower monthly income (Gramer et al., 2014; Ten Hoedt et al., 2011). Other factors associated with decreased family caregiver quality of life include younger age, lower education level, and rural location of the family caregiver as well as increased family stress and lack of coping and social support (Abdelaziz et al., 2019; Hatzmann et al., 2009; Shaji Thomas et al., 2021). Psychosocial factors such as emotional support have been found to be the most important in predicting quality of life of family caregivers, with several studies demonstrating that lack of support has a negative impact on the adjustment process (Fidika et al., 2013; Hatzmann et al., 2009). Therefore, routine assessment of quality of life could benefit family caregivers by providing additional psychosocial support when needed (Shaji Thomas et al., 2017).

As managing their child's IEM requires constant vigilance, organization, and planning, family caregivers often experience consequences for their social lives such as engaging in fewer social gatherings and losing "the joy of spontaneity" (Carpenter et al., 2018; Fidika et al., 2013). Additionally, as food is often involved in social gatherings, many family caregivers feel anxiety over requiring cooperation from other individuals present to help their child adhere to dietary restrictions and are often concerned that their child's diet might lead to judgement (Carpenter et al., 2018; Fidika et al., 2013). Although extended family members may provide support with this management, their overall knowledge of the child's IEM can be limited (Rajasekar et al., 2020). As such, family caregivers value when others take initiative to learn about the condition and management as this increases the ease of communication, creates a wider support system, and normalizes life outside of the immediate family (Carpenter et al., 2018; Rajasekar et al., 2020). Still, extended family members' understanding of the experience of caring for children with IEMs is often lacking, leading to family caregivers' feeling isolated and yearning for social

connection with other family caregivers with similar experiences. This poses a challenge for family caregivers in finding sufficient social support, as the rarity of IEMs can limit opportunities to meet other families who have children with the same diagnosis (Hartley et al., 2011). Thus, many family caregivers fill this void by using social media and the internet to reduce geographical barriers to connecting with other families with experience caring for a child with the same IEM (Khangura et al., 2016; Siddiq et al., 2016).

Metabolic health care providers can improve wellness outcomes by assisting family caregivers in their experience of caring for a child with an IEM (Rajasekar et al., 2020). For example, the metabolic team can help to educate family caregivers and their extended families as well as others in their local communities, such as primary care providers and school systems (Packman et al., 2007). Many family caregivers desire professional guidance for their mental health and would benefit from assistance in coordinating the use of social support resources (Arpaci et al., 2020; Rajasekar et al., 2020). Multidisciplinary metabolic teams, which can include geneticists, genetic counsellors, dietitians, psychologists, and social workers, can address these mental and emotional needs by providing formal clinic-associated supports including sessions on family caregiver self-compassion, mindfulness, and resilience (Lamoureux et al., 2015; Packman et al., 2007; Rajasekar et al., 2020; Rose et al., 2019). Metabolic team members such as social workers can also facilitate access to financial assistance which may further improve the wellness outcomes of family caregivers (Arpaci et al., 2020; Packman et al., 2007).

Family caregivers of children with IEMs report substantial stress related to financial wellbeing, especially those who care for children requiring dietary management (Cederbaum et al., 2001; Gramer et al., 2014; Packman et al., 2007). Frequent medical visits incur both out-of-pocket and time costs and have an impact on family caregiver employment and career

progression, especially for family caregivers living in more remote geographical areas who must travel to access IEM-related care (Cederbaum et al., 2001; Lamoureux et al., 2015; Rajasekar et al., 2020; Rose et al., 2019). Additional financial concerns may arise due to changes in employment, such as loss of insurance (Cederbaum et al., 2001). Family caregivers report spending significant amounts of time attempting to obtain coverage or reimbursement for necessary management supplies such as medical foods, many of which are purchased out-of-pocket (Berry et al., 2020). For example, foods modified to be low-protein are typically ordered through a medical foods company, require shipping, and can be up to eight times more expensive than their unmodified counterparts (Berry et al., 2020). Additionally, coverage for these products as well as other supplements or pharmaceuticals may or may not be available depending on where the family resides (Lamoureux et al., 2015). The costs of these management supplies and frequent clinical monitoring disproportionately impact family caregivers with low socioeconomic status, especially those who have poor accommodation and in families without access to a personal vehicle (Arpaci et al., 2020; Cederbaum et al., 2001). Consequently, financial wellbeing may be an important predictor of social adjustment and adaptation to caregiving for a child with an IEM (Cederbaum et al., 2001).

All in all, the constant supervision and coordination of care required for children with IEMs presents family caregivers with immense challenges (Arpaci et al., 2020). Family caregivers who lack strong support systems or who have limited financial resources experience a lack of personal time and freedom which can contribute to reduced overall wellness (Arpaci et al., 2020; Cederbaum et al., 2001).

1.3 Impact of COVID-19 on Caregiving for IEMs

In March 2020, the coronavirus disease 2019 (COVID-19) pandemic was declared by the World Health Organization (Herle et al., 2021). Since then, COVID-19 has disrupted health care, transportation, and many other systems globally (Chen et al., 2021). According to recent literature, in some cases family caregivers have even delayed taking their children to the emergency department in circumstances when they typically would have prior to the pandemic (Baumbusch et al., 2022). In particular for health care systems, access to medical specialists and allied health care such as physiotherapy, occupational therapy, and speech and language therapy were disrupted as the model of care shifted from in-person to virtual platforms to accommodate public health measures (Baumbusch et al., 2022). While virtual care has been an effective substitute for in-person care for some individuals, this transition has exacerbated substantial health access disparities for others, for example family caregivers with lower household income and limited English proficiency (Chen et al., 2021; Qian et al., 2022). Families with lower household incomes are also more likely to encounter challenges related to unemployment, crowded living environments, and reduced internet access to attend virtual appointments (Jones et al., 2020). Additionally, for those who are still required to attend in-person medical visits, the pandemic presents new barriers such as changes in the availability of public transit (Chen et al., 2021). Overall, the COVID-19 pandemic required many patients with complex care to adapt with changes to management strategies and coordination of care, including family caregivers of children with IEMs. Furthermore, children with underlying chronic disease, including children with IEMs, are at increased risk of experiencing more severe outcomes due to COVID-19, especially those at risk of acute metabolic decompensation with intercurrent illness (Brunetti-Pierri et al., 2020; Elmonem et al., 2020; Jones et al., 2020).

Globally, IEM-related health care services were reduced up to 80% of normal in 2020 at the peak level of restrictions put in place to prevent the spread of COVID-19 (Herle et al., 2021). Metabolic services had to adapt to comply with public health measures by rearranging or reducing outpatient clinics and converting most consultations and follow up appointments into virtual care via phone calls or videoconferencing software (Brunetti-Pierri et al., 2020). At the same time, metabolic services such as NBS programs were maintained, although family caregivers were directed to comply with restrictions such as the limitation of only one family caregiver accompanying their newborn with positive NBS to the hospital for evaluation (Brunetti-Pierri et al., 2020). With these restrictions in place, children with IEMs had fewer in-person appointments and blood collection for IEM monitoring, although initial research appears to show that metabolic control was typically maintained when monitored by at-home dried blood spot cards during this period, at least for children with PKU (Herle et al., 2021; Rovelli, Zuvadelli, Ercoli, et al., 2021). Although studies exploring perspectives of family caregivers are limited, virtual care was highly appreciated among most patients with IEMs, with many citing reduced travel and wait times, decreased risk of infection, and fewer impacts on daily life such as school and employment (Rovelli, Zuvadelli, Paci, et al., 2021; Walkowiak et al., 2021). Overall, it appears there is a desire for virtual care to be implemented into support provision and as standard follow up on a long-term basis for patients with IEMs regardless of COVID-19 and public health measures (Rovelli, Zuvadelli, Paci, et al., 2021).

1.4 Rationale for Current Study

The experience of family caregivers with low-income accessing care for their children with IEMs has not been previously investigated in Manitoba. Previous studies have shown that IEMs can have a dramatic impact on the wellbeing of families. Understanding the patient and

family experience is crucial to providing person-centred care for IEMs (Khangura et al., 2016). Recent research has demonstrated that the COVID-19 pandemic has had a disproportionate effect on disadvantaged populations (Wright et al., 2020). This has increased the risk for people with lower socioeconomic status to have adverse experiences due to COVID-19, including those related to challenges accessing healthcare (Wright et al., 2020). This study is intended to promote health equity by highlighting the experiences of families with low-income caring for children with IEMs in Manitoba. Advocacy for families with low-income caring for children with IEMs is crucial as clinical interventions can only lead to improved outcomes if patients have appropriate access to them (Potter et al., 2013). It is our goal that the information gathered through this study will be used to improve patient care for families that are affected by IEMs in Manitoba and beyond and to advocate that all patients have equitable access to metabolic care services regardless of socioeconomic status.

Chapter 2: Study Overview

2.1 Overview of Program of Genetics & Metabolism

The Program of Genetics & Metabolism located at Health Sciences Centre in Winnipeg, Manitoba, Canada is the only genetics program responsible for providing both pediatric and adult metabolic services to the entire province of Manitoba as well as Northwest Ontario and Western Nunavut (Hartley et al., 2011). This geographic region encompasses more than three million square kilometers of land with a population of over 1.2 million people (Hartley et al., 2011). The Program of Genetics & Metabolism in Winnipeg is supported by a variety of staff members including administrative professionals, genetic assistants, genetic counsellors, and medical geneticists. Currently, the pediatric metabolic service, including positive NBS follow-up, diagnosis, and management of children with IEMs, is supported by two of the program's genetic assistants, two metabolic genetic counsellors, three of the program's medical geneticists, a few genetic residents, and a variety of administrative professionals. The metabolic service is limited to one dietitian and currently does not have a dedicated nurse, social worker, or psychologist as part of the metabolic team.

In Manitoba, NBS is facilitated by Cadham Provincial Laboratory following the collection of a dried blood spot sample within five days of birth (Dinchong, 2019). As consent for NBS is implied, parents must actively opt-out if they do not wish to participate (Dinchong, 2019). In the case of a positive NBS result, Cadham Provincial Laboratory notifies the collection site and the parents of the newborn are typically contacted by the on-call genetic counsellor or geneticist from the metabolic service in Winnipeg (Dinchong, 2019). In the most urgent cases, parents are instructed to bring their newborn to the emergency department immediately to treat or prevent a metabolic crisis (Dinchong, 2019). For some, this can require significant travel

especially if living rurally or remotely, for example with families even requiring MedEvac by air ambulance from Northern Manitoba, Northwestern Ontario, or Nunavut. At this time, relevant information on medical and family history is gathered and parents may be instructed to stop breastfeeding and start dietary management until an IEM is confirmed or ruled out, which can take several weeks to months (Dinchong, 2019). If the result is confirmed to be a true positive and the infant is diagnosed with an IEM, ongoing monitoring, education, and treatment is provided by a metabolic geneticist and genetic counsellor (Dinchong, 2019). Importantly, NBS programs vary by region and thus not all children with IEMs are diagnosed on NBS. Families may also opt-out of NBS, have a false negative result, or immigrate from a country that does not have a formal NBS program. In these cases, the metabolic service in Manitoba initiates biochemical and molecular testing as appropriate upon referral with clinical suspicion of an IEM.

Upon initial diagnosis, families are provided psychosocial support and education about the specific IEM via genetic counselling. However, families may not grasp all information provided in this first session due to the shock and distress of receiving a diagnosis or coping with acute management issues (Hartley et al., 2011). Families are provided a detailed written management protocol letter specific to their child which is also shared with their primary care provider and local emergency department (Hartley et al., 2011). Children with IEMs require frequent follow-up with the metabolic service, with some requiring check-ins as frequent as once per week to stabilize and maintain their metabolic control, while others are monitored once every 3 to 12 months depending on their age and stability of their condition (Hartley et al., 2011). This thesis explores the experiences of family caregivers who are accessing this ongoing care for children with IEMs in Manitoba.

2.2 Research Questions & Objectives

The main research question explored in this study was “What are the lived experiences of family caregivers with low-income specifically as it relates to accessing diagnosis-related care for their children with IEMs in Manitoba?” There were several additional objectives of this study. The main objective of this study was to gain understanding of the experiences of these family caregivers through the identification of themes across information shared by participants. This study also had three secondary objectives: (1) To explore the factors that facilitate accessing IEM-related care and those that cause difficulties or act as barriers to accessing IEM-related care; (2) To explore the perceived impact of the COVID-19 pandemic and related restrictions on family caregivers’ experiences of accessing IEM-related care; and (3) To compare the experiences of accessing IEM-related care for urban and rural families.

Chapter 3: Methodology

3.1 Overview of Research Design

This research used a mixed methods sequential explanatory design, collecting both quantitative and qualitative data in two consecutive phases (Ivankova et al., 2006). The first phase of this study employed a quantitative survey instrument to collect demographic and related data about family caregivers and children with IEMs, and emphasis was given to the second phase in which individual qualitative interviews were collected to further explore study participant's personal experiences providing and accessing care for their children with IEMs.

This study was approved by the University of Manitoba's Health Research Ethics Board under approval number HS24997 (H2021:233) and the Shared Health Research Impact Committee (SH2021:101).

3.2 Survey

3.2.1 Participants

Participants in this study are defined as family caregivers who were identified by a clinical geneticist and metabolic genetic counsellor querying our clinical database for a list of children who met inclusion criteria: children under the age of 17 years with IEMs who may require frequent emergency care due to metabolic decompensation, even with appropriate treatment. Family caregivers were broadly defined for the purpose of this study to include any family caregiver (e.g., biological parents, adoptive parents, foster parents, grandparents, aunts, uncles, siblings, etc.). Potential participants were also identified prospectively by clinicians as families were seen in the metabolic clinic for regular follow-up visits in an effort to increase response rate, and four potential participants enrolled in the study following clinician completion of the Consent to Contact form (Appendix A) during scheduled clinic visits. In total, 98 children

with a diagnosis of an IEM were identified, meaning that at least 98 family caregivers were eligible for this study.

3.2.2 Recruitment

Recruitment for this study began with the distribution of a study invitation letter (Appendix B) to family caregivers providing study details, a paper copy of the survey with return postage, as well as access to an online survey to a total of 98 family caregivers. The online survey was hosted at the University of Manitoba through REDCap, which is a secure, web-based software platform designed to support data capture for research studies (Harris et al., 2009). Clinicians mailed out the 98 study invitation packages to the family caregiver of each child diagnosed with an IEM who met inclusion criteria previously defined. Previous studies surveying populations with low income have had response rates between 35-64% depending on the user-friendliness and literacy level of the survey (Fredrickson et al., 2005). As there are numerous barriers known to impact the participation of low-income populations in research, this study was designed to mitigate some of these. Strategies employed to reduce barriers for low-income research participants include: (1) providing recruitment and study materials accessible to potential participants with lower literacy levels (2) being flexible in scheduling of phone interviews which allowed participants to contribute from home, offering availability in the evenings or weekends in addition to regular hours; and (3) providing an honorarium to interview participants to value their time and contribution and reduce the financial barriers to participating in research (Schnirer & Stack-Cutler, 2012). Additionally, the Children's Hospital Research Institute of Manitoba's Research Advisory Council (RAC) was engaged to ensure this survey was accessible, respectful, and appropriate for the study population. Ninety-four reminder packages, including a reminder letter (Appendix C) providing study details and access to the

secure online survey as well as a second paper copy of the survey with return postage, were mailed out four weeks following the initial survey distribution in an effort to increase response rate. Review of the Survey Consent Disclosure (Appendix D) and completion of this survey was considered to be participant consent to participate in the quantitative survey portion of this research project.

3.2.3 Data Collection

The survey was composed of two parts and is provided in Appendix E. The survey was expected to take approximately 15-20 minutes to complete, and data collection occurred from November 2021 to February 2022. Part A of the survey collected details about the child: their current age, sex assigned at birth, diagnosis, age at presentation, comorbidities, and level of required IEM-related care including frequency of clinic follow-up, emergency visits, hospital admissions, and treatment. IEM-related care was defined as outpatient follow-ups, obtaining relevant medications or formula, emergency visits, hospitalizations, and other specialist visits related to the child's diagnosis. Specific information related to diagnosis was excluded from any reported data to protect patient confidentiality.

In addition, Part B of the survey collected demographic data regarding the family caregiver: relationship to the child with an IEM, current age, gender, ethnicity, level of education, occupation(s), and average gross monthly or annual household income. The survey collected the first three digits of each participant's postal code which were used to determine whether each family caregiver lived urban or rurally and to approximate how far they must travel to receive care. Data was also collected on a pre-determined rating scale of severity of the child's diagnosis as perceived by each caregiver and whether COVID-19 and related restrictions had a positive impact, no impact, or a negative impact on access to IEM-related care. Data on out-of-

pocket expenses for IEM-related care was collected using survey questions adapted from the Statistics Canada General Social Survey [Cycle 32] Caregiving and Care Receiving Questionnaire (Statistics Canada, 2018).

Survey participants were also asked whether they would be interested in having the opportunity to participate in an interview to share their experiences in the second phase of the study. Due to time constraints of the study, it was clear that not all participants who were interested may be selected for the interview, and that information collected in the completed survey would help to determine eligibility for participation.

3.3 Interview

3.3.1 Participant Selection

Once survey responses were received, it was initially planned to conduct approximately 15 interviews. Purposive sampling for family caregivers with lower income and who were supporting a child with one of the several included metabolic disorders known to our centre was planned to select approximately 5 family caregivers with low income to participate in individual, semi-structured interviews of approximately 30-40 minutes in length. These participants would have been selected based on preliminary analysis of quantitative data, using Question 10 in Part B of the survey to offer interviews to respondents who reported the lowest income. If respondents had left this question blank, they would have been offered an interview if they responded “Yes” to Question 21 in Part B of the survey regarding whether they had experienced financial hardship due to their caregiving responsibilities. The remaining 10 family caregivers were planned to be selected for interviews following these first 5 interviews so that purposive sampling could be adjusted to achieve data saturation and to explore missing variables that had been provided in survey responses to Questions 11-13 in Part B of the survey regarding factors

that have facilitated or caused difficulties in accessing care but had not yet been discussed in previous completed interviews. However, only nine survey respondents indicated that they would be interested in being contacted for an individual interview and thus we offered an interview to all interested respondents, with a total of eight participants who were successfully contacted and who provided consent for the qualitative portion of the study via the Consent Form for Individual Interview (Appendix F) and had an individual interview completed. One participant could not be contacted despite multiple attempts to invite for an interview.

3.3.2 Data Collection

The interview guide, provided in Appendix G, was developed to capture data to reflect the study objectives and expand on the data collected in the survey by exploring the participants' overall experiences of accessing IEM-related care for their children, exploring factors that facilitated and factors that caused difficulties in accessing care, as well as exploring the impacts of COVID-19 and related restrictions and the financial impacts of caring for a child with an IEM. Each interview was left open at the end to allow the participant to speak freely about anything that they felt was relevant and important. The Children's Hospital Research Institute of Manitoba's RAC was engaged to ensure this interview guide was respectful and appropriate for the study population. All interviews were conducted by the student principal investigator over the phone and were recorded virtually via Zoom videoconferencing (Zoom Video Communications Inc., 2016). One interview participant had a family member present during the interview to help interpret in their language. All interviews were conducted between February 2022 and March 2022. The interviews were then transcribed either by the student principal investigator or by a third-party transcription service Transcript Heroes (*Transcript Heroes*

Transcription Services Inc., 2022). All audio recordings were reviewed by the student principal investigator to ensure accuracy of data transcription.

To ensure that family caregivers' had access to financial support and other resources, a plain language booklet outlining benefits and programs at the federal, provincial, and regional levels produced by Manitoba Centre for Health Policy was shared with the participant following each interview (Manitoba Centre for Health Policy, 2020). Additionally, a \$25 honorarium in the form of a pre-paid credit card was mailed to each participant or to a charitable organization of their choice following their interview as a token of appreciation for their participation.

3.3.3 Positionality & Reflexivity

Positionality is the process of self-examining one's social location, including their intersecting relationships with power and identity, to become aware of privileges that may have been unearned or taken-for-granted (Charmaz, 2017). I, the student principal investigator of this study, am a white cisgender female in my mid-20s with a bachelor's degree from a Westernized educational institution and am pursuing a career as an allied health professional. I recognize that although I am a first-generation university student and would not have been able to pursue graduate education without generous support from awards, government student loans and bursaries, I have still benefitted from socioeconomic privilege. I also recognize that I have not had personal experience as a parent or as a family caregiver and that this lack of experience likely affected the development of my research questions and study materials, including the survey and interview guide. I do have experience working and volunteering in a variety of settings that support family caregivers, including being co-facilitator of a support group for family caregivers of adults with dementia, and I hope that this experience has improved my ability to explore this research topic with openness and authenticity. Through these and other

personal experiences, I have grown passionate about supporting family caregivers, especially those caring for individuals with genetic conditions, so I developed this project in the hopes of benefitting family caregivers of children with IEMs. Finally, I acknowledge that this thesis project contributes to the fulfillment of my graduate program and that I ultimately benefit from this research as well.

Self-reflexivity is the practice of maintaining self-awareness and self-exposure throughout the research process to acknowledge one's role and maintain honesty within one's research and with one's audience (Tracy, 2010). Reflexivity was incorporated throughout this research project, beginning when the student principal investigator assessed their own motivations and qualifications to explore this research topic. The student principal investigator also wrote memos before and after each interview to note their own expectations, biases, thoughts and feelings (Charmaz, 2017).

3.4 Quantitative Data Analysis

Survey response data were entered into Microsoft Excel and IBM SPSS software to perform descriptive statistical analyses (IBM Corp, 2020). The small sample size in this study limited the amount of quantitative statistical analyses that could be performed. A University of Manitoba biostatistician was consulted to inform the bivariate data analysis, which was conducted using the one-tailed Fisher's exact test, a statistical test which is used to assess independence between two variables especially when working with small sample sizes (Kim, 2017). A *p*-value of less than 0.05 was considered statistically significant for this analysis.

3.5 Qualitative Data Analysis

Constructivist grounded theory was used as the methodological approach for the qualitative aspects of this research (Charmaz, 2006). Grounded theory, founded by Glaser and

Strauss, is a methodology that sets out to produce or discover explanatory theory of a process from systematically obtained data (Chun Tie et al., 2019). The constructivist version of grounded theory attributed to Kathy Charmaz identifies the researcher to be embedded in the research process, taking an active role in the construction of experience and meaning alongside study participants, as well as locates the research within both historical and current social and situational contexts (Birks et al., 2019; Charmaz, 2017). Ideally, a researcher guided by grounded theory would conduct interviews in tandem with analyzing qualitative data, allowing the researcher to implement theoretical sampling of concepts and developing theory further in ongoing interviews as concurrent data collection and analysis is a hallmark of the process (Timonen et al., 2018). However, as noted by Timonen and colleagues (2018), this ideal is not always practical in projects with time constraints, and a grounded theory can be developed despite these limitations. In this study, the student principal investigator initiated the majority of data analysis, with the exception of memo writing, following the completion of data collection.

Once all eight interviews were transcribed, the student principal investigator coded the data using Dedoose software (*Dedoose Version 9.0.17, Web Application for Managing, Analyzing and Presenting Qualitative and Mixed Method Research Data*, 2021). This initial pass-through of coding generated 87 unique codes that were closely grounded in excerpts from study participants. To improve rigor, two of the eight transcripts were also independently coded by the student principal investigator's advisor using this initial codebook to assess interrater reliability followed by discussion to compare the application of codes. The resulting codes were then categorized according to content, such as "positive NBS disclosure and subsequent events" and these categories were examined for relationships between individual codes, such as "lacking support in managing care" and "wanting formal psychological/emotional support". Five major

categories were identified and were reviewed through dialogic engagement with two metabolic geneticists, two metabolic genetic counsellors, and a metabolic dietitian who are regularly immersed in metabolic-related care and interact with family caregivers of children with IEMs, to increase the validity of these categories. These categories then informed the development of a constructivist grounded theory and the construction of a theoretical model to explain the process of family caregiving for children with IEMs. This theory is presented with thick description to increase credibility (Tracy, 2010). The gender of family caregivers and their children with IEMs was removed from the presented interview results and quotations to protect the identity of study participants.

3.6 Patient Engagement

We incorporated patient engagement where possible in this study to ensure that the research being conducted was relevant and valuable to the patients that it affects. In preparation of this study protocol and study instruments, we met with the Children's Hospital Research Institute of Manitoba's RAC to review our study invitation letters, the survey, and the interview guide. Revisions were made following this meeting to accommodate the feedback provided by RAC members. In addition, we had hoped to recruit one or two study partners who have personally experienced accessing complex care for a child as a family caregiver with low income. Recruitment of a study partner was attempted by a member of the RAC who posted an invitation letter on a Facebook group for family caregivers of children requiring complex care. Unfortunately, we were not successful in identifying a study partner to be recruited to this study and time constraints limited our recruitment efforts.

Chapter 4: Survey Results

4.1 Demographic Characteristics of Family Caregivers

The demographic characteristics of survey respondents are described in Table 1. A total of 25 surveys were completed for a response rate of 25.5%, with 20 respondents returning the supplied paper survey by mail and 5 respondents completing the online survey on REDCap (Harris et al., 2009). All but one of the 25 (96%) family caregivers who responded to the survey were parents, including 23 mothers and one father, although one grandmother (4%) was also included. Seven respondents (28%) were between the ages of 20 and 29 years, six (24%) were between 30 and 39 years, ten (40%) were between 40 and 49 years, and one respondent (4%) was 50 years or older. One respondent (4%) did not report their age. A majority of respondents were women (22/25, 88%), although two respondents were men (8%) and one respondent was Two Spirit (4%). The most common ethnicity reported was European origins (13/25, 52%). Other reported ethnic backgrounds included Metis (5/25, 20%), Asian origins (3/25, 12%), African origins (1/25, 4%), Latin, Central, and South American origins (1/25, 4%), and First Nations (1/25, 4%). Two respondents reported having an ethnic background not listed, including Middle Eastern (1/25, 4%) and Canadian (1/25, 4%). The level of Westernized education varied among survey respondents, with five (20%) reporting some high school education, seven (28%) reporting having a high school diploma or equivalent, two (8%) reporting a trades certificate or diploma, four (16%) reporting some college or university with no degree received, five (20%) reporting a college/university certificate or diploma below the bachelor's level, seven (28%) reporting a bachelor's degree, and three (12%) reporting a university certificate, diploma, or degree above the bachelor's level. One respondent (4%) reported having Indigenous knowledge. Most (17/25, 68%) respondents were currently employed. Survey respondents were asked to provide either annual or monthly household income before deductions. If only monthly

household income was reported, it was converted to annual household income by multiplying it by twelve. Annual household income was relatively evenly distributed among survey respondents, with three respondents (12%) reporting less than \$30,000, five respondents (20%) reporting \$30,000 to \$49,999, four respondents (16%) reporting \$50,000 to \$99,999, four respondents (16%) reporting \$100,000 to \$149,999, and three (12%) respondents reporting over \$150,000. Six respondents (24%) did not provide their household income. The median annual household income reported by respondents was \$54,000. Finally, the distance from where the family caregiver lived to Health Sciences Centre, the tertiary care hospital, was estimated from the first three digits of respondent's postal codes. Ten respondents (40%) were within 30 minutes of the hospital and likely resided within the city of Winnipeg. Most other respondents lived rurally, with two (8%) respondents living 30 minutes to 1 hour away from tertiary care, seven (28%) living 1 hour to 1.5 hours away, three (12%) living 1.5 to 2 hours away, and three (12%) living over 2 hours away from tertiary care, one of whom lived over 5 hours away in a remote location which would require MedEvac in the case of an emergency such as a metabolic crisis.

Table 1. Demographic characteristics of 25 survey respondents.

Type of Caregiver	<i>n</i> (%)
Parent	24 (96%)
Grandparent	1 (4%)
Age (in years)	
20 – 29	7 (28%)
30 – 39	6 (24%)
40 – 49	10 (40%)
50 +	1 (4%)
No response	1 (4%)
Gender Identity	
Two Spirit	1 (4%)
Man	2 (8%)
Woman	22 (88%)

Ethnicity*	
First Nations	1 (4%)
Metis	5 (20%)
European origins	13 (52%)
Latin, Central, and South American origins	1 (4%)
African origins	1 (4%)
Asian origins	3 (12%)
Other	2 (8%)
Education*	
Some high school	5 (20%)
Graduate high school or equivalent	7 (28%)
Trades certificate or diploma	2 (8%)
Some college or university, no degree	4 (16%)
College/university certificate or diploma below the bachelor's level	5 (20%)
Bachelor's degree	7 (28%)
University certificate, diploma, or degree above the bachelor's level	3 (12%)
Indigenous knowledge	1 (4%)
Employment Status	
Employed	17 (68%)
Not employed	8 (32%)
Annual Household Income (\$)***	
< 30,000	3 (12%)
30,000 – 49,999	5 (20%)
50,000 – 99,999	4 (16%)
100,000 – 149,999	4 (16%)
150,000 +	3 (12%)
No response	6 (24%)
Distance to Tertiary Care	
< 30 minutes	10 (40%)
30 minutes – 1 hour	2 (8%)
1 – 1.5 hours	7 (28%)
1.5 – 2 hours	3 (12%)
> 2 hours	3 (12%)

*More than one response accepted per respondent

**Median annual household income of \$54,000

4.2 Characteristics of Children with IEMs Receiving Care

The characteristics of the children who survey respondents were caring for as well as some additional care-related variables are described in Table 2. There was a relatively even distribution of ages of children who were being cared for, with six (24%) respondents reporting that they cared for a child between the ages of 0 and 2 years, four (16%) reporting a child

between 3 and 5 years, four (16%) reporting a child between 6 and 8 years, six (24%) reporting a child between 9 and 12 years, and five (20%) reporting a child between 13 to 16 years of age. Fifteen (60%) of the children were reported to be assigned female at birth and ten (40%) were reported to be assigned male at birth. A majority (17/25, 68%) of respondents reported that their child was diagnosed on NBS, meaning that they were typically diagnosed prior to symptom onset rather than upon clinician suspicion following the onset of symptoms. The most common form of IEM management reported was dietary restrictions (17/25, 68%). About half of respondents (12/25, 48%) reported using medical formula or foods to manage their child's IEM. Other management strategies included vitamins or other supplements (10/25, 40%), medications (7/25, 28%), and surgery such as insertion of a gastrostomy tube (2/25, 8%). Five respondents (20%) reported other management strategies such as frequent feeding, screening and monitoring via blood spot cards, or using a continuous glucose monitoring system.

Eight of the survey respondents (32%) reported that the COVID-19 pandemic itself or related restrictions had impacted access to care for their child, with two of those eight (25%) reporting a positive impact and all eight (100%) reporting a negative impact. Finally, in terms of financial impacts, there was a variety of responses on what care-related out-of-pocket expenses were required in the past 12 months. Eight respondents (32%) reported that they did not have any out-of-pocket expenses in the past 12 months. Three respondents (12%) reported spending between \$1 and \$99 on this care, five respondents (20%) reported spending between \$100 and \$499, six respondents (24%) reported spending between \$1000 and \$4999, and three (12%) respondents reported spending over \$5000. The maximum out-of-pocket expenses in the past 12 months listed was \$51,000, which was attributed to being spent on home modifications and specialized aids/devices related to their child's diagnosis. Otherwise, the most common cause of

out-of-pocket expenses was medications, averaging over \$400 per year for those who disclosed these expenses. When asked if they had experienced financial hardship because of their caregiving responsibilities, most respondents (20/25, 80%) reported ‘No’. However, four respondents (16%) reported that they had indeed experienced financial hardship in the past 12 months, with one commenting “I do not take all work shifts that are available to me.” A second respondent commented that they were unable to get a full-time position due to COVID-19 restrictions and that the cost of their child’s restricted diet “made it hard financially.” A third respondent commented that “all of the uncovered medications and device cost[s] are not insignificant. It would be nice to see more of the requirements covered by health care.” One survey respondent (4%) did not provide a response to this survey question.

Table 2. Characteristics of 25 children of survey respondents.

Child’s Current Age (in years)	<i>n</i> (%)
0 – 2	6 (24%)
3 – 5	4 (16%)
6 – 8	4 (16%)
9 – 12	6 (24%)
13 – 16	5 (20%)
Child’s Sex Assigned at Birth	
Male	10 (40%)
Female	15 (60%)
Was your child diagnosed via NBS?	
Yes	17 (68%)
No	8 (32%)
How is your child’s diagnosis managed?*	
Dietary restrictions	17 (68%)
Medical formula or foods	12 (48%)
Medications	7 (28%)
Vitamins or other supplements	10 (40%)
Surgery	2 (8%)
Other	5 (20%)
Has the COVID-19 pandemic itself or related restrictions affected access to care for your child?	
Yes	8 (32%)
<i>If yes, positive impact</i>	2 (25%)
<i>If yes, negative impact</i>	8 (100%)
No	17 (68%)

Estimated care-related out-of-pocket expenses in the past 12 months (\$)	
0	8 (32%)
1 – 99	3 (12%)
100-499	5 (20%)
500-999	0 (0%)
1000-4999	6 (24%)
5000 +	3 (12%)
During the past 12 months, have you experienced financial hardship because of your caregiving responsibilities?	
Yes	4 (16%)
No	20 (80%)
No response	1 (4%)

*More than one response accepted per respondent

As a proxy for condition severity, family caregivers were asked how many clinic visits on average their child required per year by different modes including in-person appointments, phone calls, Telehealth, and virtual appointments for any clinic visit related to their IEM diagnosis (Figure 1A). For respondents who reported at least one clinic visit, the minimum was 1 visit and the maximum was 24 visits across all modes. Of these, in-person visits ranged from 1 visit on average per year to 20 visits on average per year, phone call appointments ranged from 1 to 12, Telehealth visits ranged from 1 to 2, and virtual appointments ranged from 1 to 4. One respondent (4%) reported that their child required zero visits on average per year across all modes. Additionally, family caregivers were asked how many trips to emergency or urgent care and how many hospital admissions their child required on average per year (Figure 1B). Eight of the 25 associated children (32%) were reported to require trips to emergency, with a minimum of 1 trip to emergency or urgent care on average per year and a maximum of 4 trips per year. Three of these children were reported to require hospital admission, with a minimum of 1 admission on average per year and a maximum of 2 admissions on average per year. Seventeen of the associated children (68%) were reported to require zero trips to emergency or urgent care and

zero hospital admissions on average per year. In general, children who did not require trips to emergency or hospital admission had fewer clinic visits per year, whereas those who required trips to emergency and hospital admissions received more frequent follow-up. When family caregivers were asked to rate the severity of their child’s condition on a scale from 0 to 10, with 0 being not serious and 10 being extremely serious, a majority of the respondents (14/25, 56%) reported their perceived severity of their child’s condition to be an 8 or more (Figure 2). Five (20%) respondents reported their perceived severity of their child’s condition to be a 4 or 5, and four respondents (16%) reported their perceived severity to be a 2 or less.

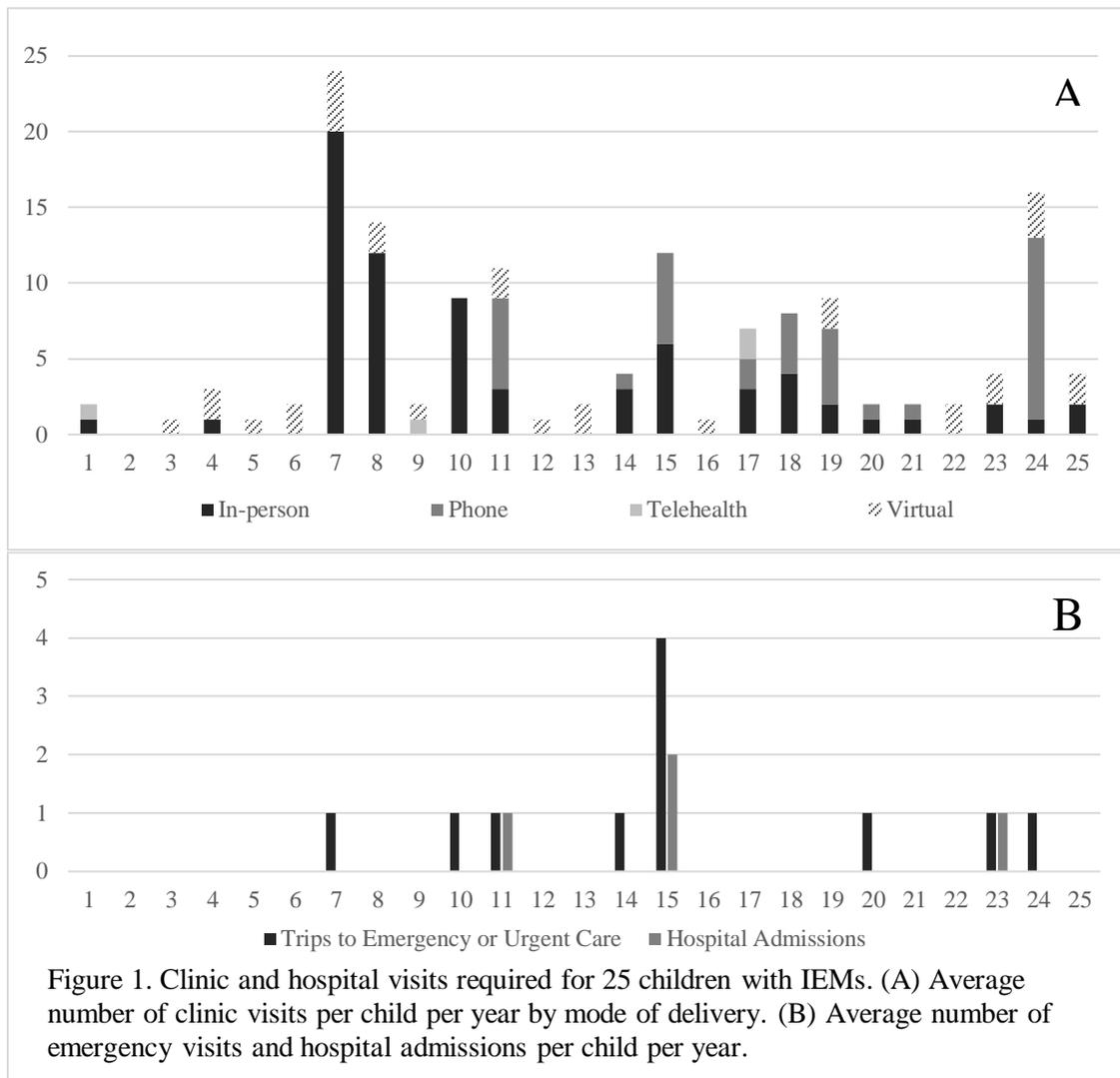
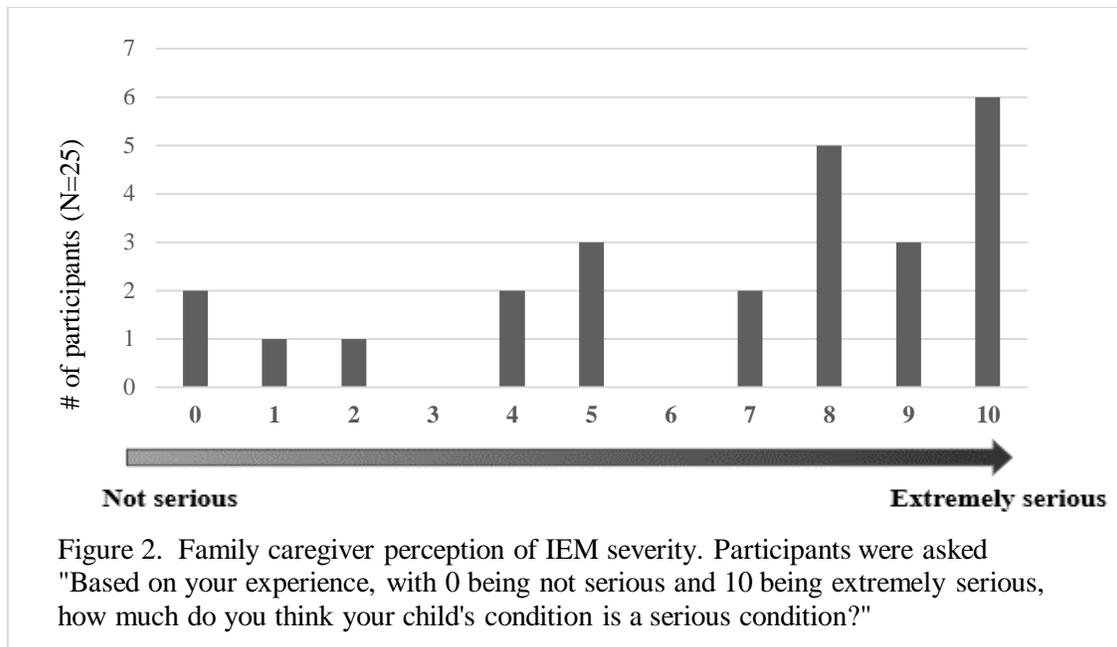


Figure 1. Clinic and hospital visits required for 25 children with IEMs. (A) Average number of clinic visits per child per year by mode of delivery. (B) Average number of emergency visits and hospital admissions per child per year.



4.3 Open-Ended Survey Responses

The survey instrument contained open-ended questions to explore factors that facilitated access to care and those that caused difficulties or acted as barriers to accessing care as well as to explore the impact of the COVID-19 pandemic and related restrictions on access to care. When asked what factors had helped in accessing care for their child, the most common response (6/25, 24%) was that the health care team or metabolic team was helpful. Three respondents (12%) reported that either technology or telephone/virtual appointments were helpful and three (12%) reported that it was their family doctor who was helpful. Two respondents (8%) reported that the metabolic dietitian was helpful, two (8%) reported that having tools to do blood spot tests at home was helpful, and two (8%) reported that therapy was helpful. Many other facilitating factors were reported once across all surveys (4%) including government travel grants, daycare, postal services, the hospital social worker, having friends in similar situations, the ability to stay home, having a metabolic protocol letter, and medication or specialized food coverage.

On the other hand, when asked what factors caused difficulties or prevented access to care, ten respondents (40%) reported “nothing.” However, for the survey respondents who reported a factor that caused difficulties, the most common response was the lack of knowledge that healthcare providers had of the condition (2/25, 8%). Other factors reported once across all surveys (4%) included difficulties with transportation, language barriers, sudden increases in medication costs, remote living location, distance between Health Sciences Centre and rural communities, and expensive diet. Additionally, one respondent commented that that a difficulty was “the fact that [my child’s] condition doesn’t fall under normal ‘umbrellas’ and [I] had to fight for any assistance,” and another commented that “COVID-19 has made virtually every aspect of accessing care for our child more difficult.”

For the eight respondents who reported that the COVID-19 pandemic and its related restrictions had an impact on their access to care, all specified a negative impact. Negative impacts of COVID-19 reported included having more phone appointments and fewer in-person appointments which resulted in fewer physical examinations and longer wait times for results and tests. One respondent commented that this negative impact meant that the “doctor can’t physically see my [child].” Additionally, it was reported that speech therapy and physiotherapy were no longer available due to COVID-19. Finally, two respondents noted the increase in cost for specialty foods that are needed to meet the unique dietary requirements of their children, with one commenting “I worry more about shipping and supply chains [...] Food costs have also increased exponentially and we have fewer ways to cut costs with a restricted diet.” However, two caregivers also specified at least one positive impact from the COVID-19 pandemic, such as its restrictions leading to more telephone appointments which resulted in less travelling for appointments as well as less time-consuming appointments.

4.4 Bivariate Analysis of Survey Data

Bivariate analysis of survey data using Fisher's exact test revealed that survey respondents were significantly more likely to perceive their child's condition as a 5 or more on a scale of 0 to 10, with 0 being not serious and 10 being extremely serious, if their child required one or more trips to emergency or urgent care on average per year ($p = 0.009$) or if they did not have a bachelor's degree or higher Westernized education ($p = 0.043$).

Additionally, survey respondents were more likely to report that access to care had been impacted by COVID-19 if their child required one or more trips to emergency or urgent care on average per year ($p = 0.04$) or if they were 35 years of age or younger ($p = 0.011$). These younger respondents were also more likely to report that their child required at least one trip to emergency or urgent care on average per year, although this association was not statistically significant ($p = 0.067$). Additionally, survey respondents who reported having a bachelor's degree or higher Westernized education were significantly less likely to live over an hour from tertiary care ($p = 0.01$). On the other hand, those who were 35 years of age or younger were more likely to live over an hour from tertiary care, although this association was not statistically significant ($p = 0.05$).

This analysis also revealed that survey respondents who reported having some high school and no other Westernized education were significantly more likely to have reported spending \$500 or more on care-related out-of-pocket expenses in the past 12 months ($p = 0.017$). Additionally, survey respondents who were caring for a child under the age of 6 years or who were 30 years of age or younger themselves were both significantly less likely to have an annual household income over the provincial median annual household income of \$68,147 (Statistics Canada, 2016) ($p = 0.001$ and $p = 0.004$, respectively). Survey respondents who reported an

annual household income over the provincial median were less likely to report financial hardship in the last 12 months, although this association was not statistically significant ($p = 0.069$).

Finally, survey respondents who were over the age of 30 years were less likely to be interested in participating in an interview as were respondents who reported that access to care was not impacted by the COVID-19 pandemic, although these associations were not statistically significant ($p = 0.069$ and $p = 0.70$, respectively).

Chapter 5: Interview Results

5.1 Survey Responses of Interview Participants

A total of 8 interviews were completed, ranging from approximately 17 minutes to 28 minutes, with an average of 23 minutes. The demographic characteristics of these interview participants which were collected from their survey responses are described in Table 3. All (8/8, 100%) of the family caregivers who participated in an interview were parents. Four participants (50%) were between the ages of 20 and 29 years, one participant (13%) was between 30 and 39 years, and three participants (40%) were between 40 and 49 years. A majority of participants were women (6/8, 75%), although one participant was a man (13%) and one was Two Spirit (13%). The most common ethnicity reported was European origins (3/8, 38%). Other reported ethnic backgrounds included Metis (1/8, 13%), Asian origins (1/8, 13%), and First Nations (1/8, 13%). Two participants reported having an ethnic background not listed, including Middle Eastern (1/8, 13%) and Canadian (1/8, 13%). The level of Westernized education varied among interview participants, with one (13%) reporting some high school education, three (38%) reporting having a high school diploma or equivalent, one (13%) reporting a trades certificate or diploma, one (13%) reporting some college or university with no degree received, one (13%) reporting a college/university certificate or diploma below the bachelor's level, two (25%) reporting a bachelor's degree, and one (13%) reporting a university certificate, diploma, or degree above the bachelor's level. None (0%) of the interview participants reported having Indigenous knowledge. Most (6/8, 75%) participants were currently employed. Annual household income of the interview group varied, with two participants (25%) reporting less than \$30,000, three participants (38%) reporting \$50,000 to \$99,999, one participant (13%) reporting \$100,000 to \$149,999, and one (13%) participant reporting over \$150,000. One participant (13%) did not provide their household income. The median annual household income reported

by interview participants was \$54,000. With regard to the estimated distance from where the family caregiver lived to the tertiary care hospital, four participants (50%) were within 30 minutes of the hospital and likely resided within the city of Winnipeg. All other participants lived rurally, with two (25%) living 1 hour to 1.5 hours away, one (13%) living 1.5 to 2 hours away, and one (13%) living over 2 hours away from tertiary care.

Table 3. Demographic characteristics of 8 interview participants.

Type of Caregiver	<i>n</i> (%)
Parent	8 (100%)
Grandparent	0 (0%)
Age (in years)	
20 – 29	4 (50%)
30 – 39	1 (13%)
40 – 49	3 (38%)
50 +	0 (0%)
No response	0 (0%)
Gender Identity	
Two Spirit	1 (13%)
Man	1 (13%)
Woman	6 (75%)
Ethnicity*	
First Nations	1 (13%)
Metis	1 (13%)
European origins	3 (38%)
Latin, Central, and South American origins	0 (0%)
African origins	0 (0%)
Asian origins	1 (13%)
Other	2 (25%)
Education*	
Some high school	1 (13%)
Graduate high school or equivalent	3 (38%)
Trades certificate or diploma	1 (13%)
Some college or university, no degree	1 (13%)
College/university certificate or diploma below the bachelor's level	1 (13%)
Bachelor's degree	2 (25%)
University certificate, diploma, or degree above the bachelor's level	1 (13%)
Indigenous knowledge	0 (0%)
Employment Status	
Employed	6 (75%)
Not employed	2 (25%)

Annual Household Income (\$)***	
< 30,000	2 (25%)
30,000 – 49,999	0 (0%)
50,000 – 99,999	3 (38%)
100,000 – 149,999	1 (13%)
150,000 +	1 (13%)
No response	1 (13%)
Distance to Tertiary Care	
< 30 minutes	4 (50%)
30 minutes – 1 hour	0 (0%)
1 – 1.5 hours	2 (25%)
1.5 – 2 hours	1 (13%)
> 2 hours	1 (13%)

*More than one response accepted per participant

**Median annual household income of \$54,000

The characteristics of the children whom interview participants were caring for as well as some additional care-related variables are described in Table 4. The ages of children who were being cared for varied, with three (38%) participants reporting that they cared for a child between the ages of 0 and 2 years, two (25%) reporting a child between 3 and 5 years, two (25%) reporting a child between 9 and 12 years, and one (13%) reporting a child between 13 and 16 years of age. Five (63%) of the children were reported to be assigned female at birth and three (38%) were reported to be assigned male at birth. Most (7/8, 88%) interview participants reported that their child was diagnosed on NBS. The most common forms of IEM management reported were dietary restrictions (7/8, 88%) and using medical formula or foods (6/8, 75%). Other management strategies included medications (2/8, 25%) and vitamins or other supplements (1/8, 13%). One participant (13%) also reported frequent feeding as another management strategy.

Five of the interview participants (63%) reported that the COVID-19 pandemic itself or related restrictions had impacted access to care for their child, with one of those five (20%) reporting a positive impact and all five (100%) reporting a negative impact. Finally, in terms of

financial impacts, two participants (25%) reported that they did not have any out-of-pocket expenses in the past 12 months. Two participants (25%) reported spending between \$1 and \$99 on this care, one participant (20%) reported spending between \$100 and \$499, and three participants (38%) reported spending between \$1000 and \$4999. When asked if they had experienced financial hardship because of their caregiving responsibilities, most participants (6/8, 75%) reported ‘No’, although two participants (25%) reported that they had experienced financial hardship during the past 12 months.

Table 4. Characteristics of 8 children of interview participants.

Child’s Current Age (in years)	<i>n</i> (%)
0 – 2	3 (38%)
3 – 5	2 (25%)
6 – 8	0 (0%)
9 – 12	2 (25%)
13 – 16	1 (13%)
Child’s Sex Assigned at Birth	
Male	3 (38%)
Female	5 (63%)
Was your child diagnosed via NBS?	
Yes	7 (88%)
No	1 (13%)
How is your child’s diagnosis managed?*	
Dietary restrictions	7 (88%)
Medical formula or foods	6 (75%)
Medications	2 (25%)
Vitamins or other supplements	1 (13%)
Surgery	0 (0%)
Other	1 (13%)
Has the COVID-19 pandemic itself or related restrictions affected access to care for your child?	
Yes	5 (63%)
<i>If yes, positive impact</i>	1 (20%)
<i>If yes, negative impact</i>	5 (100%)
No	3 (38%)
Estimated care-related out-of-pocket expenses in the past 12 months (\$)	
0	2 (25%)
1 – 99	2 (25%)
100-499	1 (13%)
500-999	0 (0%)

1000-4999	3 (38%)
5000 +	0 (0%)
During the past 12 months, have you experienced financial hardship because of your caregiving responsibilities?	
Yes	2 (25%)
No	6 (75%)
No response	0 (0%)

*More than one response accepted per participant

5.2 The Experience of Family Caregiving for Children with IEMs

The following constructivist grounded theory was assembled based on the experiences shared during individual interviews by eight family caregivers accessing care for children with IEMs in Manitoba (Figure 3).

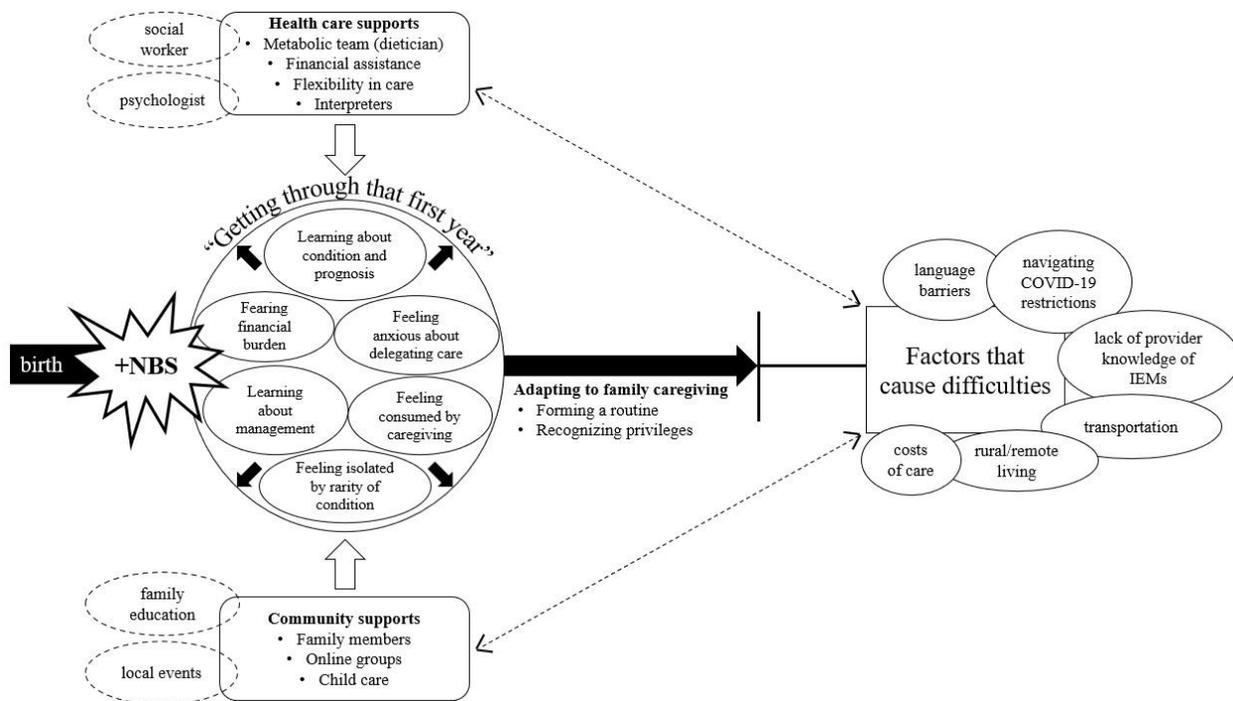


Figure 3. Theoretical model of the experience of family caregiving for children with IEMs.

The experience begins following the birth of the child and is upheaved by the impact of an unexpected positive newborn screen and confirmatory diagnosis of an IEM. In some cases,

the experience may instead be upheaved by the onset of symptoms followed by clinical diagnosis of an IEM. Family caregivers are met with expanding stressors that push their limits in “getting through that first year.” As they try to cope with their child’s diagnosis of an IEM and the great level of care that is required, there are a myriad of factors that cause difficulties for family caregivers which push back on their ability to adjust despite their resilience. There are also numerous factors that can help family caregivers to mitigate the stressors and difficulties that overwhelm them especially in their first year. Over time, family caregivers adapt to the demands of caregiving, although some of the factors that cause difficulties remain throughout their experience of accessing care for their child.

5.2.1 Receiving a Positive Newborn Screen and Clinical Diagnosis

There is already a great deal of stress and excitement associated with the experience of giving birth to a child. This stress can be compounded by delivery complications and postpartum recovery. Many parents leave the hospital following the birth of their newborn expecting to have an opportunity to rest and recover. In the case of positive NBS, disclosure is often accompanied by instruction for family caregivers to take their newborn for further testing. In the most urgent cases, family caregivers are instructed to immediately bring their child directly to the emergency department to meet with the metabolic team. This experience can have an explosive impact on family caregivers’ lives (Figure 3a).

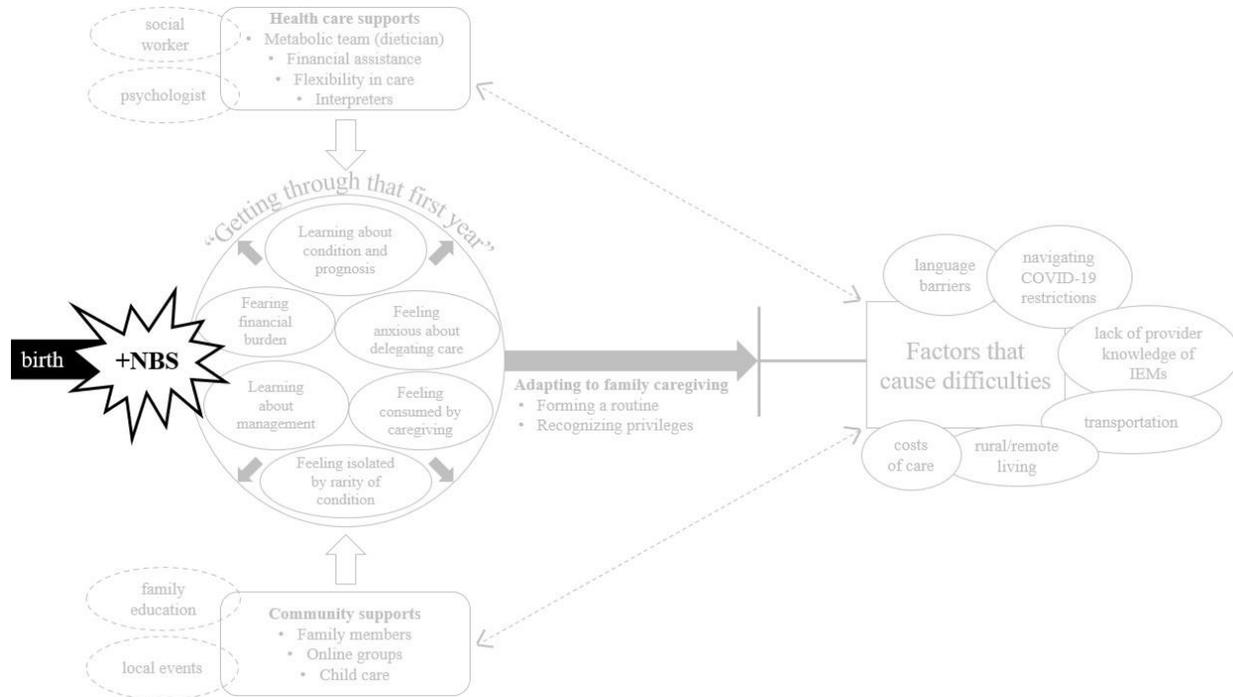


Figure 3a. Theoretical model of the experience of family caregiving for children with IEMs, highlighting the impact of receiving a positive newborn screen and clinical diagnosis.

Urgently acting on positive NBS

The impact of receiving a positive NBS result and following diagnosis of an IEM so early after birth is highlighted by Family Caregiver 13.

“Uh, well in the beginning it was, I guess overwhelming. [My child] was diagnosed with [an IEM] at 6 days old so it was a lot to take in.”

– Family Caregiver 13

Receiving urgent instruction upon this positive NBS can be a traumatic experience for family caregivers. Family Caregiver 8 described the exhaustion they felt following delivery and the loss of autonomy that they experienced upon being instructed to bring their newborn back to the hospital. They also describe the anxious waiting period that began upon arrival.

“We had already been at home trying to settle in from being away from home for a whole week [to have a Caesarean section]. So we were overwhelmed. And we got a call from the metabolic doctors, and they had – I guess they were trying to

scare us or something because the way they reacted towards us, tried to tell us that we needed to go back to the hospital right away. It was urgent and that we didn't really have, like, a choice. We just [wanted] some rest. But we ended up going back to the hospital just after that phone call. And we waited the rest of the night in the hospital while they did some more tests on [my child]. So it was – honestly, [my child's] diagnosis was probably one of the worst experiences I've had since [they were] born.” – Family Caregiver 8

Family Caregiver 19 echoes the urgent action that was required upon receiving their child's positive result and subsequent time that is spent waiting in hospital for confirmatory testing and first contact with members of the metabolic team.

“We got a call [...] telling us to come to the hospital because [our child's] newborn screening had been flagged. They kind of talked us through what [the specific IEM] could be on the phone, and then said we had to go for further testing. So, we went like right then and there. And then, I sat in the hospital with my [child] for the rest of the day and then we were admitted overnight and then we were in contact with [the metabolic geneticist]” – Family Caregiver 19

The extent of urgent action is amplified by the distance that the family caregiver must travel to bring their newborn to the hospital following positive NBS disclosure. Family Caregiver 23 describes the requirement of a MedEvac due to their remote living location.

“[Winnipeg was] where we got MedEvac to when [our child] was born. So we developed a relationship with the doctors and the team that was on the ground there.” – Family Caregiver 23

Experiencing a diagnostic odyssey

As not all children with IEMs are diagnosed by NBS, this theory could be adapted to show the difficulties prior to receiving a diagnosis upon clinical suspicion of an IEM. In this scenario, the experience of family caregiving prior to the diagnosis of an IEM would be more complex and involve unique challenges, and a diagnosis may come with more relief than shock. For example, Family Caregiver 6 was caring for a child who had not been diagnosed by NBS. Instead, their child experienced a diagnostic odyssey that lasted over ten years across multiple

countries, in which they received multiple misdiagnoses before ultimately receiving their current IEM diagnosis.

“There were so many ups and downs.” – Family Caregiver 6

To move to Canada with their child, Family Caregiver 6 waited alone for six years while their spouse immigrated to Canada as a student. During this period before moving to Canada, their child was struggling with repeated dietary changes due to their misdiagnoses. This caregiver then describes the relief that came with having proper dietary management once a final diagnosis was received.

“We had to take [my child] off of food- like, because the [misdiagnosis] right... so [they] love[d] to eat like fries, but [they] had to stop. Very, emotionally, very disturbing right? [...] I got [them] eating fruit, vegetable, but every time [they start] vomiting. [...] But we were unaware [that the diet was improper], right? We started getting [them] eating fruits, everything. And it was the same. [...] So, everyone was like, ‘why [are they] different?’ [and thought it was] weird that [they] don’t like fruit, chocolate, [but] that’s how [they were]... what can we do? [...] So, those times were very tough for us, but later on when [they were] diagnosed [with the IEM] then it was like ‘finally’ for us. [...] [They] can eat what [they] love now.” – Family Caregiver 6

Family Caregiver 6 also recounted the financial challenges that they incurred in Canada prior to being seen by the metabolic service, due to one of the misdiagnoses which resulted in their child taking ten tablets of a medication three times a day.

“After coming here, the one difficulty we face[d] was we find that the medication [for the misdiagnosis] was expensive. [...] And we were looking for some help but [we didn’t get] good help. [...] We didn’t have good jobs between both of [us being] students for medical anything. [...] We were in too much financial burden, from like back home [when] we spen[t] so lots of money on [our child] going up the road every year and then spending so much money on [their]... on top of that, medicine, and even after coming here. So, we’d had zero...we have had no financial support.” – Family Caregiver 6

Now that Family Caregiver 6 has connected with the metabolic service and their child has received a proper diagnosis, they are grateful for the support they now receive which has improved their experience of caregiving.

“So, we feel so far lucky after coming here. Like, for... it is far better, we are so lucky.” – Family Caregiver 6

5.2.2 “Getting through that first year” Following Diagnosis

A child’s diagnosis of an IEM leads family caregivers into an overwhelming period of intense education and growing anxiety, especially throughout the first year (Figure 3b).

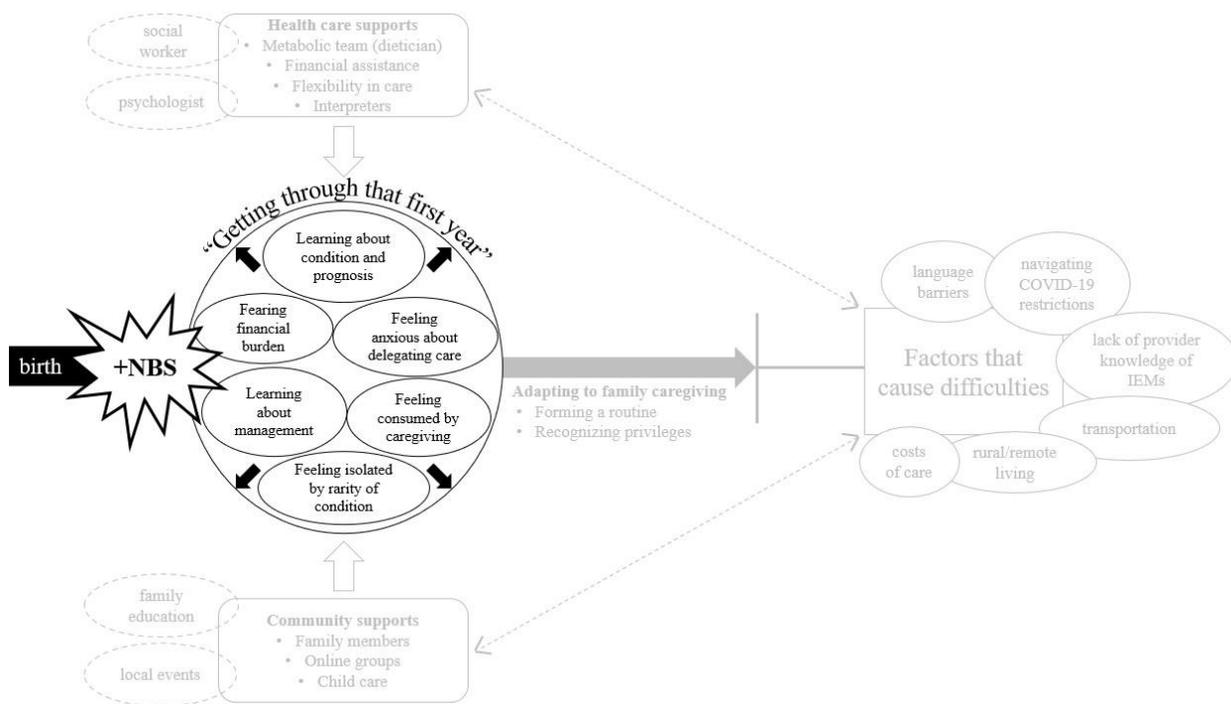


Figure 3b. Theoretical model of the experience of family caregiving for children with IEMs, highlighting the experience of “getting through” the first year following diagnosis.

This period includes learning about the specific condition and its prognosis as well as learning about specific IEM-related management. Family caregivers feel anxious about delegating this care to others who have not received this education from the metabolic team and tend to personally ensure appropriate management on their own, which leads to feeling

consumed by caregiving. Additionally, family caregivers feel isolated by the rarity of the condition as friends, family members, and non-genetics healthcare providers tend to have limited or no prior knowledge of the condition. As family caregivers learn more about the costs of the required management for their child's IEM, they begin to fear financial burden. All these processes immediately following diagnosis contribute to expanding the stress that family caregivers experience as they navigate "getting through that first year."

Learning about condition and prognosis

Family Caregiver 23 described the anxiety-inducing experience of reading about their child's specific IEM diagnosis online following initial counselling about the condition from the metabolic team.

"You Google it and then you find these people. And it was a really hard – like, it was hard at the beginning because it ranged from extreme to mild, right? And you just did – like, people's experiences with it were so varied. Right? Like, some people's children were, you know, they died or there were mental issues and stuff like that. And, you know, [...] they warned us. [...] One of the doctors [or] the genetic counsellor there at the hospital, that they told us, like, everybody's experience with this is very different. Where you want to just, you know, be careful and know that not all the cases are going to be the same and so that was good. But with time it, you know? Yeah." – Family Caregiver 23

Family Caregiver 23 continued to detail the experience of learning about the condition, including the uncertain prognosis that can be associated with an IEM.

"I think it just takes experience, right? Like, until you start to live it. Like, you really don't know. Like, and you don't know that – you don't even know what when they looked at [my child] and the genes and stuff and even then they couldn't even tell you that it was going to be mild or how [they were] going to react. I mean, they kind of tell you too like, you know, [they] might be mild for now but, you know, when [they hit] puberty and things get out of whack or something, you're like you just don't know. You kind of live with a little bit of, 'I don't know, what's going on', kind of thing." – Family Caregiver 23

Family Caregiver 8 shared how receiving information on the treatments available for their child's IEM quelled some of their anxiety.

“After we looked into it, it wasn't so terrifying because we know that it's now treatable.” – Family Caregiver 8

Family Caregiver 24 spoke about how the most significant difficulty especially in the early stages of receiving their child's diagnosis was the lack of psychological support during this intense period of processing their child's diagnosis, and how they struggled to seek out external support as it was not provided by the metabolic team.

*“Yeah, I mean it was very much the onus was on me to seek out those extra supports and that can feel very, very overwhelming when it's separate from your kid's diagnosis and team because you already have so many things to do... to then have to source out your own support that way is really challenging and I think it's a pretty universal experience of many parents getting that diagnosis that it's very, very hard to process. And so, I think it would be beneficial to have those emotional or psychological supports built-in to that system a little bit better.”
– Family Caregiver 24*

Learning about management

Family Caregiver 13 reported that the most difficult part of learning about management was learning how to manage their child's diet, including measuring protein intake.

“The most difficult is... I guess at the beginning, is learning the different foods and what the amount of protein is for certain foods, and just getting in the habit of having to measure everything.” – Family Caregiver 13

They continued to describe this experience, beginning at the transition from breastfeeding to using metabolic formula to care for their newborn following diagnosis and how management changed with their child's increasing age.

“With [my child's IEM], [management is] all based on diet for the most part, so when [they were] a baby I did have to stop nursing [...] for a few days, to get the levels down where they should be. And [there is also] the metabolic formula that

[they] need to drink daily. [...] But, as [they] grew older it's all managing diet, so calculating the amount of protein [...] and weighing foods and doing all that to make sure [they are] getting the proper amount of protein that was allotted, work with our dietitian and metabolic doctors to make sure [my child] stays healthy.

– Family Caregiver 13

Family Caregiver 8 highlighted the anxiety that can be associated with being in charge of dietary management, including fear of witnessing signs of metabolic distress if the diet is not followed carefully.

“Caregiving has a lot to do with how much [my child] eats and [...] drinks because the formula [they are] supposed to have to balance out how much [they are] eating and if [they] eat too much. Then [my child] starts to act different too, like [their] levels get too high or they get too low. So really you notice when someone's not OK. And when [my child] is content and taken care of. [...] Sometimes it's scary because I do look for the signs of, like, seizures and whatnot every once in a while, it's one of my biggest fears to see [my child] go through that. And so far, I've been doing a good job but there have been a couple of scares.” – Family Caregiver 8

Family Caregiver 19 describes their satisfaction with the experience of learning how to manage their child's condition at home.

“[The metabolic geneticist] was really, really wonderful. We liked him. And he kind of talked us through it and then we were able to go home the next day. We had somebody show us kind of how to use a blood glucose detector and kind of walk us through any of [their] protocols. So then, yeah, we went home after that and kind of ever since then we've just monitored blood sugars for [my child].”

– Family Caregiver 19

However, they also shared Family Caregiver 8's anxiety of being responsible for their child's metabolic control, wanting to mitigate the possibility of harm resulting from their own knowledge deficits by seeking out as much education as possible.

“I would say I probably try to get more appointments or for me I want to be as knowledgeable in the situation as I can be because I'm [my child's] number one care provider so what I don't know is detrimental to [my child] essentially, so I'm trying to learn as much as I can.” – Family Caregiver 19

Family Caregiver 19 also described their experience of trying to figure out aspects of management on their own with their partner. They detailed not being able to identify clear patterns or triggers to their child's drop in blood sugar, instead citing a parent's instincts, and they yearned for more education on factors that can influence a child's metabolic state.

“Me and my [partner] really at the beginning tried charting [our child's] feeds and [their] correlation to blood sugar but we realized that was no correlation, so I think that is something that talking to new parents of kids with [the same IEM] would be important. And [...] the other ways that energy leaves your body, so things to look for, like I find my [child] can't really go outside when it's cold, because [they] drop way too low. I didn't think of that until after the first time we brought [them] outside this winter, and it wasn't that cold. I brought [them] inside and just tested out of mom gut and [they were] quite, quite low on [their] blood sugar. So, just kind of... I don't know if there was any more like classes or any other ways to gain knowledge of it... I found that everything I had to learn was on my own and that was tricky because I didn't 100% know that what I was reading was correct, if it aligned with what my [child's] needs were, and things like that, so I think just more education would be the best there.”

– Family Caregiver 19

Finally, Family Caregiver 19 acknowledged the limits of searching for information about managing IEMs online as well as the privilege of having access to online resources.

“I think like you can Google and look up as much as you could, but not every site is completely accurate and I don't think it's really smart to just go off the internet, you know, so we did talk to our doctor. We looked on Mayo Clinic a lot and we did as much reading as we could, but also, I don't know, we had a lot of access to internet, we had a lot of access to sources of knowledge, but I think if we didn't have that access things could have been a lot harder.” – Family Caregiver 19

Feeling isolated by rarity of condition

Family Caregiver 24 described the burden of educating others about their child's IEM and how isolating that felt throughout the first year.

“[The first year] felt extremely isolating, and that's because my family was adjusting to that diagnosis [...] It was very, very isolating and I had to sort of teach everybody [about my child's diet].” – Family Caregiver 24

Family Caregiver 19 pointed out that even their friends who are in healthcare professions had never heard of their child's IEM and how this contributed to their anxiety. They contrasted this experience to family caregiving for a child with a well-known condition such as diabetes.

“The other thing that I would say too is because uhm... like [my child's IEM] is... it's rare. Like, when I first learned about it, I'd never heard of it. And talking to my friends who, even some of them who are in the healthcare field, have never heard of it. I think exposure to what [the IEM] is or what other metabolic disorders are so that people understand, because for me it's the lack of understanding and knowledge in towards [my child's IEM] that causes me anxiety [...] When you think about diabetes lots of people go oh, I know it's diabetes, or the general concept of diabetes, but nobody knows what [my child's IEM] is or even other genetic disorders similar to this.” – Family Caregiver 19

Family Caregiver 23 echoed the desire to have their child's IEM understood by others, yearning for connection with other families with similar experiences.

“It would be nice if we had other families around that have had the condition or – you know what I mean? [...]. Lots of people have kids that have issues and, you know, they relate but to have somebody actually understand, you know, what the [specific metabolic pathway] is, would be kind of nice.” – Family Caregiver 23

They added that, while others can relate to certain parts of the experience of family caregiving for a child with an IEM, to “be a parent and live it” is a very distinct experience.

“It's just, you know, I come from a science background and it's really intriguing but to be a parent and live it is totally different. But then, you know, once you tell somebody “oh we just do this four times a day”. Like, often there's so many people that have [...] something that they relate to, right? You actually realize that there are so many different health issues out there and a lot of people have gone through a lot of different things that you don't realize. Until you open that door.” – Family Caregiver 23

Fearing financial burden

Family Caregiver 24 highlighted that, despite their socioeconomic privileges, they experienced fear and anxiety when initially learning about management and the financial burden that can arise when caring for a child with an IEM.

“Not every place offers a budget for medical foods or for formula and so that was initially a very big fear for me, even though I have a good job, was how I was going to afford this because that is a real fear for people in other places”

– Family Caregiver 24

Feeling anxious about delegating care

Family Caregiver 19 outlined that although they preferred performing aspects of care on their own in the beginning, their experience of being the sole caregiver able to feed their infant has taken a toll on their mental health. They describe the anxiety they felt about the possible scenario of having to delegate care to someone else if something were to happen to make them unable to care for their child.

“I think when I first started nursing my [child], it was very important to me to almost like do it all on my own, because I didn’t want to miss a feeding. So if there was maybe more guidance to saying like it’s more important to get two people maybe to be able to feed, introduce the bottle, because I think that is where I’ve struggled a lot. Because I really up until this point have been the only one able to care for my [child]. And I think in a lot of hospital situations, I don’t think they ever tried to put a decision on me to feed, they said do what works for you, but I think in this situation if they say like look I know this is hard, but it’s going to be a very hard thing, it would be very important for you to at least introduce the bottle and have the baby be able to take the bottle. Because I also had a lot of anxiety like what happens if I died or something spontaneously and like nobody could feed my kid. It was a huge stress for me. So, I think just like that whatever choice you want to feed your child is fine, but I would... if I had more guidance telling me you should do both ways, I think I would have listened more to health care professionals than just doing whatever I thought was best. Because I think what I thought was best has been led down the road to not being what was best for my mental health and things like that.” – Family Caregiver 19

They also shared the anxiety of being hypervigilant and being skeptical of others' abilities to appropriately care for their child given how much of care relies on a parent's intuition and cannot be instructed.

“Also, to even just leave my kid with my in-laws you know, or my parents, or anything like that. They don't understand what it is and if I don't trust 110% a person to be almost hyperaware of my kid, I'm nervous to leave [them]. And I'm nervous when I have to leave [them] at daycare, [they are] not yet in daycare, but its one of those things that is causing us stress because even myself, the one or two times I've noticed [they have] been low, has been only on a [parent's] intuition. So, it's really tricky to say okay, watch for my kid's change in moods or anything like that or sleepiness or anything like that, but when I tested [them], [they] didn't have any of those symptoms. And [they were] quite low.” – Family Caregiver 19

This anxiety often leads to family caregivers to prefer to personally ensure appropriate management at all times, as highlighted by Family Caregivers 6, 8, and 19.

“Because [my child] sometimes [they get] super low all of sudden, [...] after an hour [they] sort of get down, and we need to feed [them] immediately. That uhm... so those were some difficult concerns, we had to worry when [they] go... when [they] went somewhere, like anything for camping or when [they are] at school. And yeah, so like... [my partner] or me had to be with [them]. [...] We can't leave [them] with friends or family, so that is a difficulty.” – Family Caregiver 6

“Well, I've been pretty obsessive, I guess you could say, since finding out about [my child's] condition. So I'm kind of just attached to [them]. I don't really like to leave [them] alone with other people. But [they do] have day care for when I go to work and that's about it.” – Family Caregiver 8

*“I personally have made sure [my child has] eaten every two hours, pretty well.”
– Family Caregiver 19*

Feeling consumed by caregiving

In the early stages following diagnosis, the intense and overwhelming requirements that accompany family caregiving for a child with an IEM, including learning how to manage all aspects of diet and accessing frequent medical care, are emphasized by Family Caregiver 24.

“What does caregiving look like? Well, early on, it was consuming. Absolutely consuming. So, in the first year, caregiving involved mixing breastfeeding with pumping with mixing formula and all of the measurements and weighing that we had to do for [my child’s] diet. We had many, many doctors’ appointments in the first year including going to the hospital twice a week initially for blood draws at the lab.” – Family Caregiver 24

They continued sharing their experience of how the immense time burden of frequent medical visits and long wait times pushed their limits and led to them advocating to be able to do more aspects of caregiving from home to relieve some of this pressure.

“I was at my complete breaking point having to run to the hospital every week still at that point, it took a whole year of my maternity leave, and that was a real pain too because you can never make an appointment at the lab and so you’d have to go and haul your baby and wait in whatever lineup happens to be there that day, even though you had to go every week. So that was enormously stressful and a humongous time burden for me. So, I would say like things that have helped are being able to do more of it at home which has just come with [my child’s] age. And then, as [they have] gotten older, [they don’t] require as many follow up appointments, just because that first year is so critical. So, some of that has just changed because of [their] age.” – Family Caregiver 24

Additionally, Family Caregiver 24 expressed that there was a lack of both flexibility in care and psychological support during these especially stressful stages of caregiving, during which a family caregiver is still processing their child’s diagnosis.

“It was just like the stress of how much contact there was and how many appointments, and we always had to be out and doing things. I didn’t have flexibility or freedom in how I fed [my child] but also in how I used my time because there was just so many demands on phone calls and meetings and appointments. That was... that was very, very stressful. I really don’t know that all of the appointments really needed to be that frequent or that long. I guess just like, having the support of our clinic [helped] but also not feeling like there was enough support to really process the diagnosis... like from a psychological perspective, there was zero support.” – Family Caregiver 24

Family Caregiver 7 echoed the necessity for extremely frequent medical visits during the first year following positive NBS and diagnosis of their child’s IEM.

“When [they were] six months... one month to like one year, every day we went for [their] blood tests” – Family Caregiver 7

Family Caregiver 19, who had previously shared their struggles of being the sole caregiver able to feed their child, indicated that all of these exhausting requirements of family caregiving can occur whilst waking up every couple of hours to feed an infant during the night.

“Yeah, so [my child] was exclusively breastfed up until six months, kind of when [they] started solids. And then, [they are] still breastfed and now on solids as well. [...]. But, for me I think it was more just like kind of the stress of taking on all of the feedings because [they] wouldn't take a bottle [or] formula [...] so that was a little bit difficult. But we made it through and so that was good. And at night, [the metabolic team] told us we could go a little bit longer [between feeds]. That was about six months ago I believe it was, but [my child] was really kind of in a pattern so [they are] still up every two to three hours, even now.”
– Family Caregiving 19

They reaffirmed the anxiety that occurred during this overwhelming first stage following diagnosis of their child's IEM and foreshadowed the relief of “getting through that first year.”

“Yeah, I think it's impacted me more than my [partner] in a way. Because [they] now [are] just... ‘okay, I'll help you when I can but I can't help you when it comes to feedings.’ Because [our child] would not take a bottle from [them], [they] would try, and we just left it too long. So, I don't know [laughs] it's just I think it just causes me a lot of anxiety, and I think it's getting through that first year will be a lot better than the year, because I'm getting more and more confident every day, but I think for the sole provider it's very... it's an overwhelming disorder.” – Family Caregiver 19

The feelings and impact of being consumed by caregiving did not necessarily end completely after the first year, as outlined by Family Caregivers 6 and 23.

“Yeah, because I had to go with lack of sleep during the first few years, because I have to stay with [my child] for the day. And then I have to go for night duty and during the daytime my [partner] did [their] day work, and that was really tough for me because it impacts my health also. [...] And then in day we can't also really free time to go out somewhere [...] I'll be sleeping, so actually they think ‘oh you can't go from the start of the day.’ [...] that's how we had to manage. Those were difficult years.” – Family Caregiver 6

“It’s constant as a caregiver too. Like, it’s funny because we don’t – we don’t realize it sometimes and then, like, we’ll have friends over and we’ll be like, we’ll have to give [our child] a medicine and we don’t even think about it anymore and they’re like, oh my god, you’re going to do that four times a day? It’s like, yeah.

Doing this four times a day for [several] years. [Laughter]”

– Family Caregiver 23

5.2.3 Factors that Cause Difficulties for Family Caregivers

As family caregivers navigate accessing care for their child, there are numerous factors that cause difficulties for them along the way (Figure 3c).

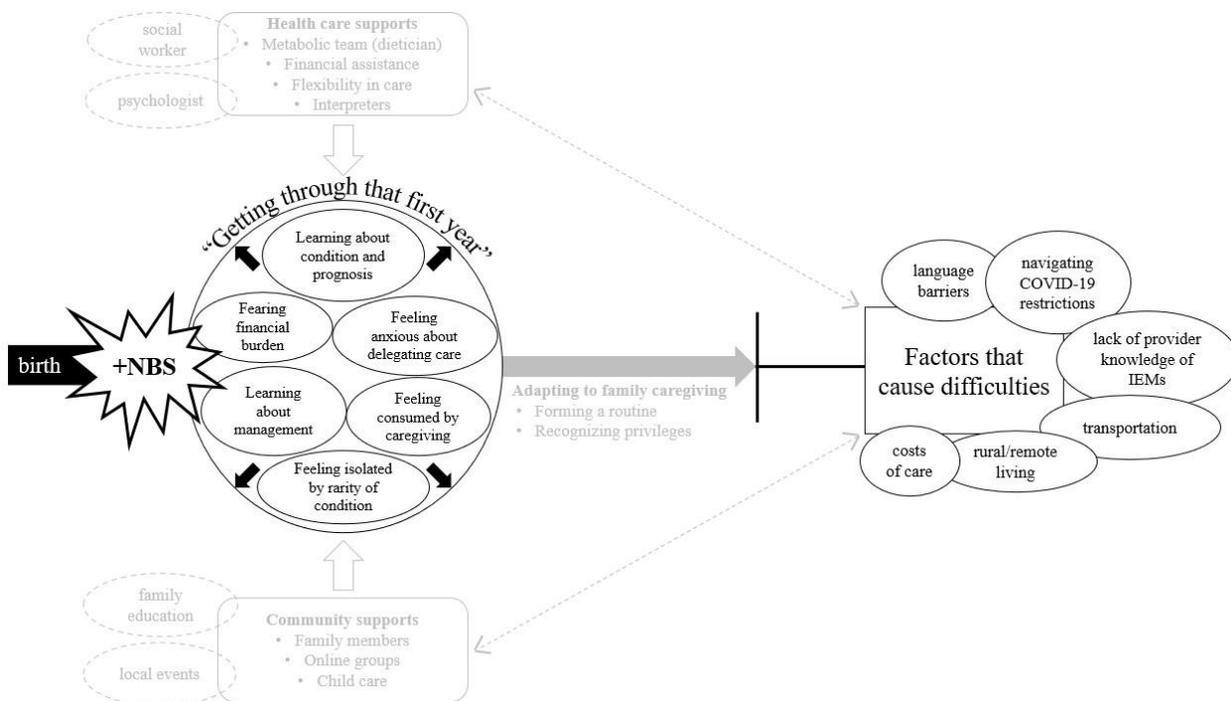


Figure 3c. Theoretical model of the experience of family caregiving for children with IEMs, highlighting the factors that cause difficulties for family caregivers.

Family caregivers are quickly introduced to the costs of IEM-related care, including costs stemming from obtaining medical foods and products as well as the costs involved with frequent access to care such as parking fees and travel expenses. Family caregivers’ living in rural or remote communities face these challenges with the added difficulties of reduced access to specialized care, longer wait times for the shipping of care-related supplies, and further distances

to travel to receive care even in an emergency. Even those who live within the city may experience transportation difficulties, such as navigating public transit in inclement weather. Additionally, family caregivers may experience language barriers attempting to access care, including in obtaining transportation and education about the condition. Once at health care facilities, such as laboratories or urgent care centres, caregivers often struggle to navigate differences in the level of care resulting from lack of knowledge about IEMs from non-genetics providers.

The COVID-19 pandemic and related restrictions present a new and unique challenge to accessing care. Family caregivers worry about their children with IEMs being exposed to COVID-19 and must navigate appointment cancellations due to COVID-19 infection. COVID-19 restrictions that limit the number of individuals that may accompany a child during care, especially for emergency department visits such as following positive NBS, lead to caregivers lacking support and feeling alone.

Costs of Care

Family Caregiver 17 thoroughly outlines the various costs of caregiving that can cause financial difficulties such as medication and other management supplies as well as transportation to appointments and childcare for other children in the home. They also highlight that due to the nature of rare disease, many of these costs are not covered by benefit plans.

“I mean, [providing care has] always kind of been a priority, so I wouldn’t say that it’s hindered us from doing it, but just the economic feasibility and stuff, having to get the stuff that’s necessary to [our child’s] health at our own costs out of our own pocket right because... hasn’t always been great but like I said well it’s gotta be done. [...] The [medical product] is roughly \$40 a month, so just the combination of [those] and [especially the test strips]. [And] the financial impact of [going into the city for medical visits], right like... Prices of gas are going up,

and then taking the day off work, and then you have to eat a couple of meals while you're in Winnipeg, and it just all adds up financially. [...] Transportation [has a financial impact] for sure. [...] We have the family usually that can help out with the kids, but that would be another added expense if that weren't the case. And then the medication that isn't covered because a lot of these metabolic disorders from my understanding are fairly rare and a lot of the medications aren't covered under even the benefit plans." – Family Caregiver 17

Family Caregiver 8 echoed many of these costs and further outlined the additional expenses that can be incurred living rurally, where diet-appropriate grocery selection is limited and prices are elevated.

"When I go shopping, it now costs me a lot more because trying to give [my child] food that isn't going to throw [their] balance off too much, but that [they] could eat as much as [they] want because [they are] a kid and [they] like to eat. It's just difficult to buy the food that [they] can't eat as much of just like we could without it harming [them]. [...] It's like, you, know organic food is always more expensive than normal store kind of food. [...] And I guess that's where it's impacted my income because my grocery bill is a lot higher now. Well, mostly because we have to go to the city to do the shopping, they don't really offer a lot of that kind of food where we are. [...] And the food that they do have here is a lot more expensive." – Family Caregiver 8

Family Caregiver 24 also resonated with the expenses of diet management for children with IEMs, adding the financial difficulties that can be experienced as a single parent.

"Yeah. I mean like, early on, a lot of the financial impact was with, you know, travel to and from appointments, and I did not live far so I can't say that was as even significant as parking. But, as [my child] started eating real foods, like [their] diet is very expensive because a lot of what [they] can eat freely is like fresh produce, which is extremely expensive, and moreso after COVID. And many like gluten-free types of products that fit within [their] diet are significantly more than a comparable non-gluten-free product. So, that piece of it is... yeah, like the grocery bills are enormous. And I do have a good job, but I am now a single parent and yeah, I notice that grocery bill for sure." – Family Caregiver 24

Family Caregiver 23 highlighted the vital impact that programs providing coverage for medications and medical foods have on their costs of caregiving, acknowledging that these expenses would be unmanageable without such programs.

“The other thing financially that has probably [an] even bigger [impact is that] [...] [my child is] in a program that basically covers [their] medications at 100% [...] And I see the price tag on what it would cost and that would be quite substantial for somebody to have to cover if – I think it’s probably about \$32,000 a year, I think, for the [medication]. [...] So that would be quite a lot. The [other medication] is a little bit cheaper. Maybe a couple of thousand dollars a year or something, maybe. So pretty minimal compared to the other ones. Yeah. But that’s covered. We just pay for shipping. [...] Part of that program too [...] they basically pay for certain specialty foods like low protein – low to no protein, like, noodles and flours and stuff like that. So they have a [...] specialty food shop and [...] they have a webpage and you can go on and you can just kind of order all of these specialty foods. And yeah. And that’s covered.” – Family Caregiver 23

Family Caregiver 13 compared the types of programs covering the costs of medical foods, suggesting that programs which direct bill to the government provide greater financial security than reimbursement programs.

“Uh, at the beginning it was I guess difficult because the way we... uhm... we do get funding from the government to help with the cost of specialty foods... they are quite pricey. But, at the beginning, when we first started to have to purchase the specialty foods, the program ran where we had to dish out the money first and then submit receipts and then get reimbursed. So that could be a little challenging at times because you’re putting it on your credit card and then yeah, you get that receipt, but then you have to submit it, so then you’re waiting for them to reimburse you, so it’s kind of financial restraint that way, but that changed [and we are now] able to order foods from a couple places and they direct bill the government, so we don’t have to worry about getting reimbursed or dishing out money first, so that is nice. [...] Other than that, no [income is not a factor] because the formula is covered and the medication that [my child] is on now is covered.” – Family Caregiver 13

Family Caregiver 24 added that having additional flexibility with food budgets provided by these programs would be valuable.

“I mean I think like in some ways, what items we always kind of like to use, like we get a food budget allowance, but it has to be for medical foods, so you... like I don’t see an amount, I have an amount. But it’s just through the medical food companies. But where it would serve me better because some items that [...] you know, produce costs and bread and pastas that [my child] can eat off the shelf, but there’s no flexibility really with that. So, in that there is a bit of a financial

burden because there's access to funding but they're not flexible"
– Family Caregiver 24

Additionally, Family Caregiver 24 shared their experience of advocating to make changes to what metabolic formula was covered by the government, once again emphasizing the importance of flexibility in these types of programs.

"It has been very hard to influence what the province will agree to pay for. So there, you know, I had come up against this moreso in my early stages, like wanting to try other formulas and them not being on the formulary and you know, I've had good advocates who argued to get things on the formulary, but that [...] was not easy at all. [...] I think if you are in that position if you're looking for something that isn't covered by the province, then yeah there are barriers to care [...]. It's not a particular barrier for us right now because my [child] is happy with what [they] have, but you know, if you're in that position where your kid doesn't want what you provided, and what you provided is the only thing that is paid for, that is a bit of a challenge." – Family Caregiver 24

In an account that highlights the obstacles that can be encountered when attempting to obtain financial assistance to cover costs of care, Family Caregiver 13 presented a frustrating scenario in which coverage for their child's medication was jeopardized following their partner's loss of insurance.

"One difficulty... I guess would be with the [medication], dealing with the company that distributes [it], because at one point we didn't have insurance and when we agreed to the trial, we were told that you know it wouldn't get taken away from [my child], and then when my [partner] switched jobs and we had lost our health insurance, then it was like well, our system is, or like the program is changing so now if you don't have insurance then you don't qualify for this, uhm you know then [my child] wouldn't be able to go on the trial. But we managed to get that all figured out, because we do have health insurance again, but it was more of a hassle at that point, and frustrating, and feeling like we weren't, I guess, getting the help we were told we would get. You know, like I wouldn't have agreed to the trial if I had known it would've been taken away or it could get taken away, the medication, because the medication is expensive, like nobody can afford it without benefits, so I guess that was a little concerning and kind of took us back a bit. Uhm... yeah, that would... that's probably the only kind of time that I felt like we were getting worked against. And it was like, in order to, I guess, qualify for their bridging program, they were wanting us to stop [my child's]

treatment and make [their] levels go higher than they have ever, like I've never had [their] levels, in order to qualify, so... that seemed like it was kind of working backwards, but fortunately we got it figured out and [they are] still on the medication and we didn't have to go to that option to qualify for the bridging program.” – Family Caregiver 13

Rural/Remote Living

Family Caregivers 8 and 17 both described the challenges of living over an hour away from tertiary care at Health Sciences Centre, including lacking laboratory services to do IEM-related blood tests closer to home and needing to take time off of work travel to the city to access specialized care.

“When [my child] was born we lived in Winnipeg but now we moved out of the city because of COVID. I just wanted to get away from all the people. But yeah. We tried getting [their] blood tested regularly here where we are and [the lab wasn't] able to do the kind of tests that we need. And no other place that we know of in the city does it either if it's not the Health Sciences Centre. So it has made our life a lot [more difficult] not being in the city and having the access to the Health Sciences Centre [...] We moved pretty far, so when we need something it's about an hour-and-a-half away. That's probably been the biggest impact, is the accessibility to everything.” – Family Caregiver 8

“Before COVID, we were going in every 6 months to a year to talk to our doctor and now we have Telehealth usually every 6 months to a year and [my child] gets bloodwork done to check [their] levels and that kind of thing. [...] We are from a smaller municipal community which is about an hour and a half outside of Winnipeg, so I mean, that hasn't always been easy. Usually, you've gotta take days off from work to go in and that kind of thing. Uhm, we used to get our bloodwork done at the local hospital but our lab no longer does that so we have to go into [a different city] or Winnipeg to get the bloodwork done now as well, so that's been a little bit of a hurdle but I mean everything is there to get done so as far as I'm concerned it's been quite good.[...] So, the Telehealths have been good because we don't have to travel quite as far.” – Family Caregiver 17

Furthermore, Family Caregiver 23 introduced the unique experience of living remotely and having to travel to Winnipeg to access tertiary care, and how this initially led their family to consider whether relocating would be necessary.

“One of our issues – our biggest issue with dealing with the condition is our remoteness. We live [remotely] in a small town [...], so we have a hospital but they don’t have the capacity to deal with [IEM-specific] testing. [...] So we’re 500 kilometers from basically the nearest hospital that deal with [condition]. About an hour flight. Yeah. So, you know, when [our child] was little, we considered moving to Winnipeg it would be closer to the, you know, to support. And then [they] got older and so, you know, it was just – [they were] more mild and mild and it was just, you know, we could – we stay where we love to live, right? So, the remoteness definitely was an issue. It’s something that we can overcome because we drive, whatever, get out there, and [they don’t] have crises all the time or anything. Yeah.” – Family Caregiver 23

Additionally, Family Caregiver 23 acknowledged that one significant drawback of living remotely is the requirement of MedEvac in the event of a metabolic crisis.

“Anytime there is a threat of a crisis we get MedEvac to Winnipeg. It’s quite an ordeal.” – Family Caregiver 23

Relevant to families living rurally or remotely, Family Caregiver 8 voiced the impact that living further away from Winnipeg had on the time required to receive management supplies such as requisitions for blood tests and medical food orders.

“Well, the whole mail thing, everything’s in the mail and you have to wait super long for whatever you order. And all [my child’s] medical food is shipped from Toronto or something, somewhere in Ontario or The States. I can’t remember where they said it was from. But it does take a while to get here. Not that I’m impatient or anything but it’s caused the – it’s caused some panics a couple of times [when] [they don’t] get [their] formula on time, or food.”
– Family Caregiver 8

Transportation

Family Caregivers 6 and 7 both shared their similar experiences of having to navigate public transit with their children in lieu of driving, especially during frigid Manitoban winters.

“We had to cancel our... like off from work between the two of us because we have to take [our child] for appointment and I wasn’t driving, so for a couple of years, so if I had to go [without my partner] then we had to take buses. Then we were from here to downtown, we had to take two buses, especially in the winter...”

oh, we walked in snow... cold weather... it's difficult so year, here [my child] wished to have [my partner who would drive] whenever we go to appointment, not me, but we have to compromise, I have to go without work routine, so that's a difficulty. [...] The most [difficult factor] was taking [them] to appointments like by bus. It was really difficult, because we were.... we're in this weather environment, everything right. And yeah, it was difficult for us."

– Family Caregiver 6

"Like sometimes I went [to appointments] by bus because my father in law, he went to work sometimes by car. We sometimes like if car at home, we go by car. [...] Yeah, like [when] I went by car- it's good. But by bus it's difficult. [...] Like the bus, it sometimes... like I late for my [child's] appointment. Sometimes I'm waiting for the bus, sometimes the bus late and the weather is very cold, it's like... for [my child]... it's very cold, very difficult."

– Family Caregiver 7

Family Caregiver 7 further described their experience of taking their child to appointments without access to a car, mentioning that they must take a taxi or ensure that they purchase bus tickets ahead of time.

"Yeah, like, sometimes we buy a [bus] ticket. [...] If we go by car, by taxi, [then] we didn't pay for that ticket for the bus."

– Family Caregiver 7

Language Barriers

Family Caregiver 7 shared their personal difficulties in navigating language barriers, citing the challenges involved with requesting a taxi by phone to take their child to medical appointments.

"Helping with the taxi, or... nobody help me with that. [...] Sometimes if my [partner] is not at home, [or] someone [else to] help me with that. Like, [calling a] taxi or things... it would be very difficult."

– Family Caregiver 7

Lack of Provider Knowledge of IEMs

Family Caregiver 6, who previously shared their experience of navigating their child's diagnostic odyssey, described the lack of provider knowledge about IEMs that hindered the

process of accessing appropriate care once they immigrated to Canada. It took multiple referrals to different providers and a long wait for their child to finally receive an appointment with the metabolic team for assessment and diagnosis.

“I think for the first, like, for the very first appointment, what we do with family doctor, at the beginning we went for physical to the family doctor, and like he was- he said that ‘oh I’m totally unaware of this kind of disease, I really have, honestly, I have no knowledge about this kind of disease’. Oh, okay, then what should you do, then you have to go to pediatric doctor, even though she has no idea where [my child] should be seen, like what kind of specialist. So, we were first referred first to the pediatric doctor. And two pediatric doctors, then one here sent us to the genetic doctor, and I think for the first appointment it took some while time. We didn’t get in right away.” – Family Caregiver 6

Family Caregiver 19 was also impacted by lack of provider knowledge of IEMs. Even when they presented their child’s metabolic protocol letter to staff at their local hospital, the staff did not acknowledge the urgency of care required. Family Caregiver 19 continued to highlight the differences in the level of care that was received when they took their child to their local hospital where pediatric supplies required for IEM-related care were limited, compared to when they received care at the Children’s Hospital at Health Sciences Centre.

“Yeah, [the hospital closest to me] was like... the staff there... had never heard of it, had never heard of [this IEM]. They didn’t really know, there wasn’t a significant urgency, even with the metabolic [...] protocol letter. They did get me into a room, but it was very slow... slow paced and moving. When I went to Children’s [Hospital] after, they got me into a room right away, got [my child] hooked up to [an] IV, made sure everything was good. And also, at [the other hospital], half the rush was they didn’t have any supplies for children that young, that small. So I think maybe in that situation, they should, even though it’s not a children’s hospital, have more access to like small IVs and things like that because half the time they were running around looking for anything that could fit [my child].” – Family Caregiver 19

Similarly, Family Caregiver 8 described the difficulties in obtaining IEM-related blood tests at their local laboratory, where lack of provider knowledge led to Family Caregiver 8

feeling that they had a burden to educate staff about how to collect IEM-specific samples. They indicated that these difficulties meant that their only option was to travel farther to Health Sciences Centre to have these tests performed.

“The knowledge that other medical facilities have of the condition [is another difficulty]. Because, well, when we try and go get [my child] – when we used to go and try and get [their] tests done somewhere else, like, at the local hospital or the closest clinic, they draw blood normally. Well, for [child’s name] they kind of just shut down. They didn’t know what to do; they didn’t know how to do it. It was supposed to be colder and spun or something like that; frozen. Didn’t know how to read the requisition. It was really confusing to try and tell them what they needed to do because I’m not exactly sure. I just know from looking at the requisition that this is what needs to happen. [...] And it was frustrating because that’s when I realized that I could really only go to the Health Sciences Centre to get [their] blood tests. So I tried a different couple other places and they didn’t know how to take from an infant. They didn’t know how to read the requisition. They didn’t know exactly what kind of test that needed to be done [...]. It just made it really confusing.” – Family Caregiver 8

Navigating COVID-19 Restrictions

Family Caregiver 19 recounted the experience of travelling to and arriving at the hospital following urgent instruction after disclosure of positive NBS. Hospital restrictions limiting the number of individuals that can enter to accompany a child led to Family Caregiver 19 feeling alone and lacking emotional support in this overwhelming experience.

“I think at the very first doctor’s visit when we were all rushed in that was where I noticed it the most because it built a panic in us, like we were trying to get in the hospital and we had security guards stopping us saying ‘you can’t come in, you can’t have more than one parent’, like we’d already called ahead and said there’s two of us. It was a snowstorm, so my [partner] had to drive but I had to feed [my child], and then we had my [other child] with us because we didn’t have any like babysitters or anything because it was so quick. So, that kind of was unfortunate. And then, me just being so postpartum in the hospital by myself was really hard. I think being able to have the support of my [partner] there would have drastically reduced my anxiety and my sense of overwhelmed-ness. And I think too just trying to get to appointments on time when you’re stopped, going through all the COVID protocols.” – Family Caregiver 19

Family Caregiver 8 voiced similar concerns regarding these hospital restrictions, especially when accessing follow up care for their child alongside their own recovery from Caesarean section and being unable to physically carry their child.

“Well, at first they made it really hard for me because I wasn’t allowed to go in with my partner. [My partner] would be carrying [our child because] I was recovering from a Caesarean, so I wasn’t really lifting anything. And when we first had [our child] that’s when we had the most tests and we had to go to the doctors more often. And they gave us a hard time every, single time we went to one of those doctor’s appointments [for their] blood testing. [My partner] would hold [our child] and I would just walk in and [security] would give us a hard time because it’s only one parent allowed. And I understand that but at the same time [my partner] doesn’t know what to do; [they’re] just carrying a baby. And I couldn’t really carry the baby at the time because they told me not to lift too much. So I had a hard time really explaining myself and the needs that I had at the time because the restrictions were so strict. [...] Honestly, it’s made me feel like I have to do everything alone. When you have a child, you’re supposed to have your support systems in place and everything. But with the restrictions, they kind of like cancelled all that out.” – Family Caregiver 8

Family Caregiver 13 raised the point that the pandemic has caused many people to feel more stressed in general, including family caregivers for children with IEMs. They also highlighted that virtual care due to the pandemic resulted in fewer physical examinations for their child."

“Uhm, I wouldn’t have to say that the pandemic changed anything for caring for [my child]. Like, in regard to [their] diet and everything. Uhm, I think it kind of affects everybody... these things, in not the same way. It’s been stressful for everybody, right, like it’s... freaky... different... Uhm, but for [my child’s] overall care, minus not going to the hospital for [their] appointments and having to do them virtually... like, I’ve talked to the dietitian and the metabolic doctor, but they actually haven’t actually seen [my child] for probably a year since the pandemic started.” – Family Caregiver 13

Family Caregiver 6 added that the logistical implications of COVID-19 can include infection in the family interfering with accessing care for their child and resulting in appointment cancellations.

“One time we had to cancel because I had COVID. So, yeah, and that time even my [partner] was not available, because if one person is COVID [positive] then everyone has to stay home. That time, we had to cancel [our child’s] appointment that way.” – Family Caregiver 6

An additional concern brought forth by Family Caregiver 8 included the fear that COVID-19 restrictions could negatively impact care, citing COVID-19 stigma as a factor that had delayed access to care.

“Well, there has been a couple times where [my child] got pretty sick and I didn’t want to take [them] in because I didn’t want [the doctors] to think it was COVID. I had tested [my child] and it was COVID-negative but they still get so worked up as soon as you have a symptom. It’s like you’re not allowed to be sick to go see a doctor. I’ve brought negative test results and everything to prove that [my child] doesn’t have COVID so that they would see [them].” – Family Caregiver 8

5.2.4 Factors that Facilitate Family Caregiving for Children with IEMs

There are a number of factors, both within the health care system and in the community, that can support family caregivers and mitigate the factors that cause difficulties (Figure 3d). Health care supports are directly related to the care provided by the metabolic team, including medical geneticists, metabolic genetic counsellors, and dietitians. Additional professionals such as psychologists and social workers could also be members of the metabolic team. Family caregivers can also receive support from their communities, including aid from family members, advice and emotional support from online groups, and assistance from childcare services. Community supports can also include local events that aim to provide family members with education about the condition as well as to connect different family caregivers of children with IEMs.

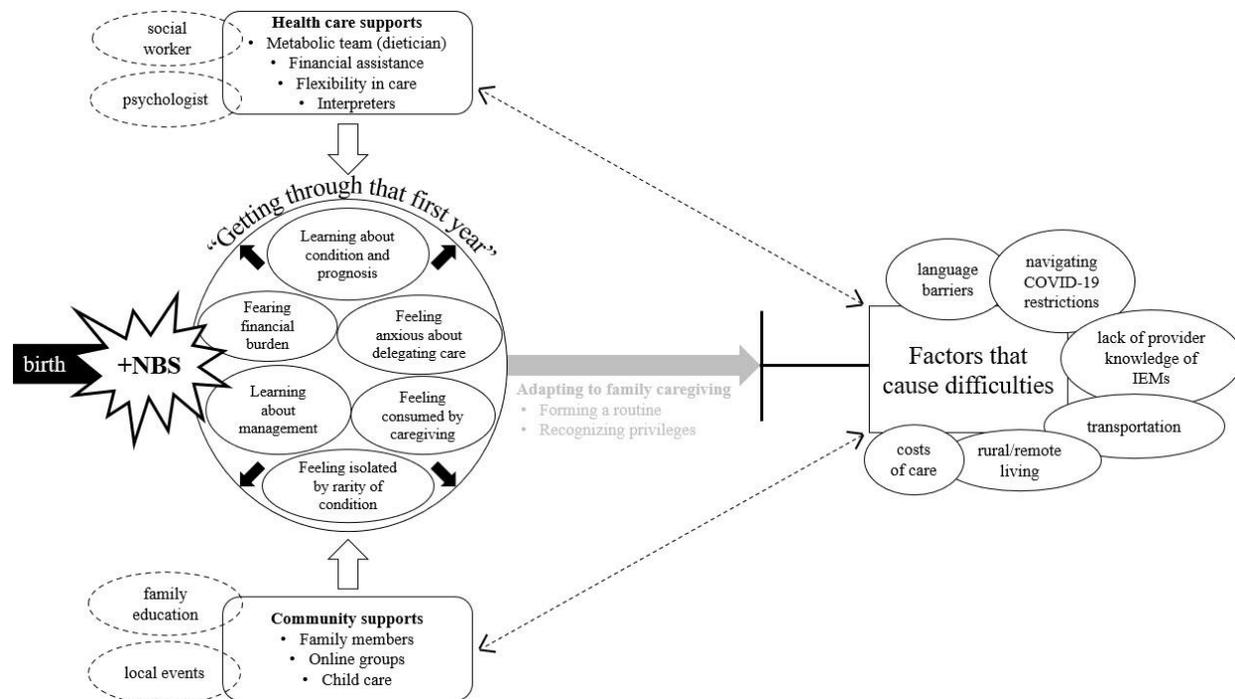


Figure 3d. Theoretical model of the experience of family caregiving for children with IEMs, highlighting factors that facilitate family caregiving for children with IEMs.

Health Care Supports: Metabolic Team

Overall, the family caregivers interviewed reported good experiences with meeting and building relationships with the metabolic team. Family Caregivers 19 and 13 voiced their satisfaction with initial and ongoing care provided by the dietitian, geneticists, and genetic counsellors on the team.

“We [...] got in contact with the metabolic specialist the day after we were in the hospital. [...] [He has] given us his number and he says ‘contact us at any time’ as well with the dietitian, I’ve reached out to her multiple times and she gets back to me always within a day or two.” – Family Caregiver 19

“I think our metabolic team has been amazing. Like, I couldn’t ask for a better metabolic doctor or a better dietitian. So... we work well together. [...] Our dietitian has been a huge help. I know when I have questions, I contact her. The doctors are, have been great as well, but it’s you know we have our regular appointments and I talk to them then, I don’t... but I’ll call the metabolic doctors in regard to [my child’s] diet and that, but who we talk to, that’s I guess the dietitian that we deal with more. [...] In the beginning, we had a genetics

counsellor and that, which was you know a huge help at the beginning especially with being overwhelmed with all the additional information that we now had to, you know... learn, in order to care for our child. [...] The support has been good.” – Family Caregiver 13

Family Caregiver 13 also highlighted their satisfaction particularly with the initial education and support they received from the dietitian.

“I’d probably have to say the dietitian [was the most significant help] just because they were able to work with me and help me, I guess become more familiar and at ease with the measuring and the counting, the calculating, of all the protein intake [...] and working closely with me to make sure that [my child’s] levels stay on par and where we like to see them.” – Family Caregiver 13

Family Caregiver 23 expressed that, even in emergency situations, they were satisfied with the care provided by the metabolic service.

“Well, the doctors and stuff are always on the schedule and booking things at the time. Like, you know, we have an appointment every six months or whatever and they’ve been, like, the metabolic on call doctor is great. I mean, we recently had to call the metabolic doctor on call and it’s – actually a couple times. And it’s pretty smooth. Even when you feel like you’re in an emergency situation, it feels pretty good.” – Family Caregiver 23

Health Care Supports: Psychosocial Support

Family Caregiver 24 reported receiving emotional support from the dietitian and questioned whether this support extended beyond the scope of practice of a dietitian.

“Well, I mean I will say that our metabolics dietitian has been incredibly kind and supportive and very, very patient and she has gotten us through all of my worst moments, so I feel like having that as a support has been a huge contributor to my resilience. That being said, I feel like at many times she did many people’s jobs, so I don’t know if that was ever strictly what her job was, it extended far beyond being a dietitian. And because she was the only one working in that position, so. Yeah, I mean like that, the level of support that I was offered through that particular person in her position was very helpful in getting me over the hump of it.” – Family Caregiver 24

Family Caregiver 8 expressed a different side of this experience, emphasizing that they did not feel that they had anyone that they could talk to for this type of support when the dietitian was unavailable.

“[One difficulty is] how often I talk to the dietitian. I know that she’s, like, one of the only dietitians in the area and she goes on holidays pretty often. So when I have a question I just – I feel like there should be somebody else I can talk to other than just her. Because, you know, she’s a person too; she has her own life and everything. Like, sometimes I get that feeling like I do need to talk to somebody and there’s nobody to talk to.” – Family Caregiver 8

Family Caregiver 24 highlighted the desire to have additional formal psychological support such as the availability of a social worker or psychologist, especially during the period of processing their child’s diagnosis.

“I think maybe what a barrier could [be] is that there just are not as many supports available to parents in that stage of supporting a kid with a lot of medical needs. You know, I think I was surprised that there were no social workers or psychologists or anybody who was there to sort of facilitate processing the information, like the diagnosis and the things that have to change immediately when you get that diagnosis.” – Family Caregiver 24

They continued to express how critical this lack of support was for them during the first year following diagnosis, adding that even other people in their life had been shocked to hear there was no professional support available to help cope in this adjustment period.

“So, as you know, and in speaking to a lot of people that I know who hear this story and how you get diagnosed, like everybody is shocked that there isn’t. Like, ‘what do you mean you just leave the hospital, and you get this information and you give [your child] formula?’, like? It’s, when I think about it, like I think it could be a lot more supportive. I think that we got very lucky in that our dietitian is a... like... a very good person [laughs] who took on more than this role, or who takes on more than her role because there’s no one else. But, I think that that would have been very, very helpful in... like I didn’t manage the first year very well at all.” – Family Caregiver 24

Family Caregiver 6 expressed that ongoing emotional support would also benefit families as they navigate issues that can arise when caring for a child with an IEM, such as addressing misconceptions and social stigma of having a rare condition.

“Families are also impacted from emotional- I’m thinking of emotional load for that kind of behaviour [when kids are treated differently by others due to their IEM], it affects their emotions, right? [...] Because that happened back at home, we sent [our child] to the school when [they] started grade 1, we explained everything to the school and then they... the teacher told all the parents also and they were like ‘oh is that a contagious disease? We keep our children, their food away from [your child]’ ... like what?! [...] That could happen over here [in Canada] also. I have no idea about over here, but we should be aware of those things also. That absolutely matters for families, what to do with this kind of thing for their kids.” – Family Caregiver 6

Family Caregiver 8 highlighted that noticing how their child’s IEM affects daily life in similar social situations was a major source of emotional distress.

“I spend every day with [my child]. And noticing the way that [their IEM] affects [them] every day is honestly the biggest challenge because [they don’t] get to have the same snacks as everyone, and they question [my child]. But nobody really treats [them] differently. It’s just, like, because no one really knows that [they have] the condition; they just wonder why [my child] can’t eat the same things.” – Family Caregiver 8

Family Caregiver 7, who also reported their child has developmental delay, spoke of the added difficulties they experience related to their child’s speech and behaviour as a family caregiver lacking support.

“Like- I don’t know, it is like... very difficult because [my child] doesn’t understand, and [they are] like always running and touching everything... [they] can’t wait for a minute like sitting for a minute. [...] when [they] need something, [they are] like... [they don’t] speak like... I didn’t understand [them]... [they] grabbed my hand and take me to the sink for the water and when [they] grab my hand to in front of the stove, [they] want to some food. Yeah, if [they]... like, we doesn’t understand [them]... [they get] very angry and [they cry]... [...] Like when we go outside, [they are] always running and [they]... when we are at the... like beside the road, I said ‘come, you can’t go to the road, street’, [they don’t]

understand, [they cry] and [they] like lying in the ground, [they are] like... Yeah, it's difficult... [...] Yeah, like always like I am running with [them], yeah, [they] can't like stopping and sitting for ten minutes." – Family Caregiver 7

Health Care Supports: Financial Support

Family Caregiver 17 emphasized the impact of receiving financial support from the metabolic team by being given management supplies and how this mitigated stress in times “when money was tighter.”

“The willingness of the health care aides to go above and beyond to help us out [is the most significant]. It definitely relieves a lot of the stress. [...] When money was tighter, they were providing the glucose meter and extra test strips for us.”
– Family Caregiver 17

They detailed the ways that the metabolic team had helped, including working with the dietitian to advocate for coverage of a new form of the medical product their child needs.

“The staff at [Health Sciences Centre] have always been very good to us. Whether it was giving us blood sugar monitors and these test strips and that kind of thing, trying to minimize the cost, so like I've said they've always been very helpful in that regard. I know our dietitian has been trying to get a [new form of medical product] rather than [our current medical product], we've been trying to get that funded through our benefits, so she's been working on that I guess it has this whole process.” – Family Caregiver 17

Family Caregiver 24 echoed this support that the metabolic team has contributed to through advocacy in having costs of care such as medical foods covered.

“Our metabolic clinic has been very good at advocating [...] because we have a medical food budget and we don't pay for formula which is also enormously helpful.” – Family Caregiver 24

Family Caregiver 19 also described receiving financial support from a benefits plan to help cover the costs of obtaining management supplies.

*“We were able to get a glucose monitor, all [my child’s] testing strips and pokers and all that stuff, like we have a benefits plan so it isn’t really a costly thing”
– Family Caregiver 19*

In regard to travel costs, Family Caregiver 23 spoke of the financial support they received through a Provincial Travel Grant that helped them to travel from their remote community to the city for medical visits.

“There’s times when [...] we just make a trip to the city. That is financially supported through the [Provincial] Travel Grant. [...] I haven’t gone in a while. I feel like it might be, like, up to around \$300, \$3- or 400 per trip. So the idea of that is that your gas is covered and your hotel is covered and maybe a little bit of food or something like that.” – Family Caregiver 23

While most family caregivers interviewed highlighted the significant financial support that they received, Family Caregiver 6 emphasized that, especially for newcomers to Canada, there is a need for formal support to help family caregivers identify and navigate obtaining these financial resources.

*“Especially if you are new [to Canada] when you don’t have much knowledge about the policy, care rules and of community health, then it’s like you’re lost. [...] Nowadays we have come to know that there are other... many government [programs] and non-government [programs] which help support families with low income and if they are on some difficulties, disabilities, or anything there--- but at that time [after immigrating to Canada], we were totally unaware,”
– Family Caregiver 6*

Health Care Supports: Flexibility in Care

Family Caregiver 24 reported that, once they had advocated to be able to collect blood spot samples at home for IEM-related testing, they experienced a significant shift in wellbeing as a family caregiver as a result of the increase in flexibility with their time.

“The thing that shifted the most for me, in terms of my wellness and time, was being able to take on more of those care pieces without having to go in for appointments all the time. That was really, really helpful. [...] Over time we’ve

been able to take some things off of, you know, having to go into the hospital for appointments and being able to do things at home. So, once [my child] was almost a year old, then I was allowed to start doing [their] blood draws at home instead of having to go into the lab. I will say, I had to have like a full on meltdown in order to get permission to do that. [...] [My child] doesn't have nearly as many appointments now so there is somewhat more flexibility in how the appointments can work around the rest of my life.” – Family Caregiver 24

Family Caregiver 8 echoed this experience, voicing that being able to do the blood spot collection at home was better, despite the time required to mail the sample for testing, suggesting that this could enable the family to travel more as well.

“The access to the medical facilities [has caused difficulties], really. Because not every, like, hospital or every blood clinic knows how to do this kind of test. And it's been one of the biggest difficulties, I would say, because you have to get [your child's] blood tests every so often. And not being able to just go to a normal doctor and get [their] blood tested and be able to travel. But now that [they are] over a year, you could do it at home so that's been nice. It's all been by mail. [...] [Mailing the blood spot cards] usually takes about a week. It's not as fast as it was if you would just go there and do it. But that's honestly better than having to go there once or twice a week.” – Family Caregiver 8

Family Caregiver 8 also voiced concerns with the flexibility of scheduling follow up appointments for their child with the metabolic team in general, specifying that the appointments provided often conflicted with their set schedule at work.

*“I usually take Mondays off because of [my child's] blood tests and everything. I just have this whole set up with my company that I take Mondays off so that I could care for [my child], take [them] for [their] doctor's appointments. But when they make the appointment for a day that isn't a Monday, then I have to try and rearrange things and possibly reschedule. And they know that I have Mondays off too, so [...] the communication between us is a little hard. [...] I would say their scheduling [is the most significant difficulty]. Because I know that they're busy people already and they have other cases and whatnot. But at the same time I think I'm just trying to provide for my [child] and my schedule is really, really routine. And then it kind of puts me off balance when they know I have certain days off and they go and make it, like, in the middle of the week or something.”
– Family Caregiver 8*

In regard to the changes of service delivery due to the COVID-19 pandemic, Family Caregiver 24 reported that the transition from in-person to virtual appointments was smooth and that they preferred to stick with this mode of care as often as possible due to the convenience and time-saving benefits of accessing care from home.

“I will say that even though COVID has been a huge pain for lots of people, I have appreciated the shift for our appointments because when we were having to go in-person they were exceptionally long and there was always a lot of waiting involved and it just felt like an excessive amount of time, and now that things are allowed to be virtual, I feel like that has streamlined those appointments also, like they used to be many hours long, or of my time, and now they’re not, so I would say like the option to have virtual appointments has actually been helpful. [...] I appreciate that, [...] [When] I don’t have to like take that much time off of work, and I don’t have to drive, and I don’t have to pick kids up and pay for parking then that’s an improvement for me. If it doesn’t have to be in person, I would prefer it not to be.” – Family Caregiver 24

They reemphasized that COVID-19 had more of a positive impact on their experience accessing care for their child, highlighting that they are still receiving all the support they need from the dietitian.

“I don’t think that [COVID-19] negatively impacted our access to care. Like, I think it has actually positively impacted. Like, I’ve never ever had a hard time getting in touch with anybody at the clinic if I needed to, and that is 99% of the time our dietitian. So, I wouldn’t say that it’s impacted our care at all because I am in contact with her as much as I ever was. And like I’ve said before, I think having some of those virtual options has improved my experience of care because I can always... I don’t particularly enjoy those metabolic check ins because I don’t always get the point of them, they’re not overly useful to me or my [child] I don’t find. So, I feel like because I talk to the dietitian regularly, like that, that is the care. So, I sort of appreciated that there is a virtual option that can sort of account for personal inconvenience factors to that appointment, because there’s no real need for them to be in person.” – Family Caregiver 24

Family Caregiver 6, who previously spoke about the difficulties using public transit to get to appointments during Manitoban winters, also appreciated the time-saving benefits of the transition from in-person to virtual appointments and being able to access care from home.

“Oh, yeah, it’s like, in some ways good, because we don’t have to go to the hospital for appointments. We started doing video conferences, and that helps so like... if we have to deal with, because [my child] has to deal with dietitian, genetic doctor and the liver doctor, right. And they all three of them come in video conference at the same time. [Whereas] if we go to hospital then we have to make different appointment days for liver doctor and different day for genetic doctor and dietitian. But during those times, all three in the same videoconference and like, at the same time. It’s time saving. [...] They send lab test requisitions right to our home. So, we just have to go for blood test and if [my child] has to do ultrasound then in that case we have to go to the hospital, in that case only. I think it’s far better, especially in winter. It’s very useful. [...] We get online very well every time, actually we had two times those two videoconference calls and both were good, clear, no problem with connection.” – Family Caregiver 6

Family Caregiver 23 preferred a mix of both in-person and virtual care in an effort to balance the benefits of accessing care from home, such as not having to travel long distances, with the benefits of attending in-person appointments, where metabolic team members can physically see their child.

“Well it’s [been] at least a year that we didn’t actually see the specialist in person. But it wasn’t a bad thing as much, right? I mean, now you realize now you don’t need to take the trip every three or six months, so that’s OK. [...] It’s made things a bit easier. You don’t have to go out of town for three days. So, I guess it saves time. [...] It’s nice to be in-person, though. It really is. Like, you know, you do the virtual appointment. And I think especially for the doctors, like, to see [my child] and how [they are] moving and up and running is very different in-person versus remotely. So I wouldn’t want to do virtual all the time. [...] So if was, you know, every second appointment was virtual or whatever that would be convenient.” – Family Caregiver 23

Although Family Caregiver 19 perceived value in accessing care from home in certain situations, they ultimately preferred to have appointments in-person when possible, citing the benefits of in-person communication and relationship building.

“I guess I would have had a couple more phone appointments and things like that [due to COVID-19], which in the past I wouldn’t have had. Although, I don’t know if that’s necessarily a negative because with my kids... well, ultimately with my kids at home that’s due to COVID, so I guess you could see it from both sides. But if I were sick or something I could access my appointment and I think my next

appointment is virtual as well. I do think though I prefer to go in to talk to the doctors if that's possible, because I just find you're more focused, you ask better questions, they get more personable with your child, they care more when they feel like they have a relationship maybe.” – Family Caregiver 19

Family Caregiver 17 reported that they would rather have in-person appointments, despite the reduced travel costs of accessing care closer to home, due to the benefit of “getting it all done in one day.”

“I would say probably not going to the doctor face-to-face [was the most significant difficulty]. Even though it might be a little more cost effective in some scenarios, that just not having that same conversation, and [...] the ease of getting it all done in one day, right?” – Family Caregiver 17

They elaborated that transitioning to Telehealth appointments caused significant difficulty to accessing care due to the increase in schedule coordination required by them to arrange care.

“Basically once COVID hit, we no longer had any appointments in Winnipeg that we would go to, they all got switched over to Telehealth and then we were doing lab work at other hospitals and stuff. So, it all became a little bit more confusing and harder to juggle. [...] So, I mean it feels like maybe things haven't been as on schedule with the Telehealth. [...] Now we have to get the forms and what [my child] can get tested for mailed to us and so by the time we get that in and then take a day to go to the city to get the lab work done, that kind of thing, it's been a little longer than it should be in some situations. [...] Care isn't quite as immediately accessible it seems like; we're not talking to the doctor face-to-face and there's just more variables. It's not just one single appointment now right, now we've got to go to the mail, then we have to make an appointment, run to [the city], then wait for them to get back ahold of us to schedule a Telehealth rather than just one appointment where everything gets done kind of thing. [...] It just gets harder to kind of keep track of everything.” – Family Caregiver 17

On the other hand, Family Caregiver 17 indicated that they felt more positively towards having some appointments by phone, suggesting that these one-on-one conversations were helpful gaining information from the metabolic team.

“Yeah, there has been some phone appointments there that we were doing as well as the Telehealth, and those actually weren’t too bad. They weren’t too bad either, you know. It was a little bit more one-on-one, and we got some good info out of the phone conversations.” – Family Caregiver 17

Family Caregiver 8 reported that they would prefer in-person appointments particularly for the initial stages of learning how to manage their child’s diet, especially given the internet connectivity issues they experienced during this process due to living rurally.

“Well, the communication out here is not very strong, so when we do Zoom calls it’s really staggered. And I know that when I was learning to try and count [...] all the protein that [my child] eats, I had a hard time understanding because of how the video was lagging and everything. Trying to hear, like, every other word. Trying to write down notes and whatnot. So it’s just, I don’t know, that kind of stuff would just be better in-person. [...] The education piece is a big part, I think. I personally am a hands-on learner; I feel more comfortable face-to-face when somebody’s trying to teach me something.” – Family Caregiver 8

Family Caregiver 7, who previously discussed language barriers, did not like virtual or phone appointments due to health care providers having fewer options for explaining information and their experience with internet connectivity issues.

“Yeah, that, like... [my child’s] appointment like... every time by phone. Yeah this is like a little bit difficult for me, because when I went there face-to-face, I better understand. [...] Because like, when I didn’t understand some question, I can’t like... they can’t explain more for me. Yeah, when by phone... like, I didn’t like appointment by phone. Yeah, sometimes like the internet like the frozen and yeah...” – Family Caregiver 7

Health Care Supports: Interpreter Services

Family Caregiver 7 emphasized the importance of having an interpreter present for each of their child’s medical appointments.

“When [my child] has appointment, always time [they have] an interpreter. And yeah, that good, like always I call the doctor, he [has an] interpreter for [my child]. This is a good thing.” – Family Caregiver 7

Community Supports: Family

Family Caregiver 24 described their knowledge of past local events in which family members could receive education opportunities, for example on the child's IEM-related diet. They reported that there were no similar events available during the time they had cared for their child, which led to lack of support and burden to educate family on the condition and its management.

“I guess thinking about it there would be some things that would be more helpful. [...] I know that there used to be events where kids with similar conditions could meet and there might be like some family education opportunities and that has not happened ever since my [child] was born. So, I don't know if that's 100% due to COVID, because there definitely was a chunk of time where there wasn't COVID and it didn't happen, so I don't know if that's the only reason, but it would have been helpful [...] for our family to learn a little bit more about the diet. Yeah, so, that was hard [to teach family] at the beginning, but now everything is great, like people know how to cook for [my child] and will bring food [that I can feed them and that's really helpful, and so now I'd say I have support.”

– Family Caregiver 24

On the other hand, Family Caregiver 17 acknowledged that family support had been a great help, especially in regard to obtaining childcare for their other children when accessing care for their child with an IEM.

“Family for sure [has helped], like, being able to... like, we home school. So, yeah just having family around who also homeschool so we can send the other kids there... we don't have to worry about hiring a babysitter or that kind of thing [...] Family has definitely been a big help!” – Family Caregiver 17

Family Caregivers 7 and 6 both spoke about having limited to no family support, with Family Caregiver 6 specifying that this was in part due to their immigration to Canada.

“Just like my family helps me when... no, nobody helps me watch after [my child], nobody, just like my mother in law. [...] Sometimes when I have appointments, she watches after [my child].” – Family Caregiver 7

“We don’t have [any] family members here [in Canada], we were all by ourselves [when we immigrated]. So, those were really difficult days.”
– Family Caregiver 6

Community Supports: Online Groups

Family Caregiver 23 shared their experience of receiving support from online communities following their child’s diagnosis, which helped them to learn more about and access resources for their child’s IEM.

“When [my child] was first diagnosed we, you know, you do a lot of Googling and stuff like that and we found a pretty big support group from The States. [...] There was one lady in particular that was running this group. She was really good in kind of helping us, you know, for the first few, just, you know, the whole world that you’re now learning about and you’re scared and you don’t know what’s going on and then she was really good with, I think, giving some resources and webpages stuff like that” – Family Caregiver 23

However, they cautioned that the internet also has the potential to invoke anxiety when searching for medical information.

“I mean, the internet is a scary place, right? For anything, right? You know, you bruise on your fingers and you Google it and that’s not good.”
– Family Caregiver 23

Family Caregiver 13 reported occasionally receiving support from online communities.

“There is a [specific IEM] Facebook page that sometimes has information on it or support that way.” – Family Caregiver 13

Similarly, Family Caregiver 19 spoke highly of an online support group that they joined immediately following their child’s diagnosis. They recounted the experience of receiving reassuring messages from other family caregivers who had older children with the same IEM diagnosis and could speak from experience that children with the IEM could lead a “relatively normal life.”

“Right after I got out of the hospital, I was extremely overwhelmed, that was also probably due to my postpartum. And so I found the [specific IEM] support group and I kind of just put out there like oh my god I don’t know what I’m going through, like anyone else have any advice? And I got tons and tons of messages that I could do it, that it was going to get better, that it was very difficult to think about in the moment but that so many kids with [the same IEM] could lead that relatively normal life. And I think all those thoughts overwhelmed me, but hearing that from other members of the support group like honestly has probably been the number one thing that has eased my anxiety towards it.”

– Family Caregiver 19

They continued to describe the support and advice they received from other family caregivers in the online support group, highlighting that people in the group could better empathize with their experience than those in their local community.

“It just gave me a lot of reassurance talking to other families who were in that exact situation, because it didn’t matter who I talked to that I knew, nobody understood because nobody had even heard of [IEM diagnosis] before. But then I found the support group on Facebook and it was everything that I was feeling, so many other [family caregivers], and people who had it themselves could relate on a level that my family and friends couldn’t relate to. So I also learned like a lot of good tricks, I learned helpful tips for travelling in the summer, helpful tips for travelling in the winter, things like that that somebody who is not knowledgeable in [my child’s IEM] wouldn’t know. And even just to calm my nerves and just letting them know how I felt like I was in the hospital, I don’t know, there’s so many things that I often check on there or I do my best to help other families on there with my input when they are asking for it. So, I don’t know, I just found that very helpful.” – Family Caregiver 19

However, Family Caregiver 19 also expressed a desire to have more local support.

“I’ve really learned a lot from other people [on Facebook] who also have [the same IEM]. So, I don’t know if there would be a way that you could find support groups in your area because the ones on Facebook are all over the world, so you could be talking to people in the United States, China, like anywhere really. So that was really, really helpful to me though, but I would think that if you could find support groups or little things in your community, I think that would be really helpful as well.” – Family Caregiver 19

Community Supports: Local Events

As other family caregivers who were interviewed reported in different contexts, Family Caregiver 13 voiced how beneficial previously attending a local education event for their child's IEM was, highlighting the positive ongoing impact it had on managing their child's diet.

"I know there were a couple times when [my child] was younger, like we did do a [local event], so there was I guess a couple other [family caregivers] that already had a little bit older kids that were there and taught some recipes and stuff to make the low-protein foods, but still make it like it's "ours". So, I learned how to make low-protein perogies and stuff like that, that we still make today. So, that was helpful." – Family Caregiver 13

Community Supports: Childcare

Family Caregiver 8 described the positive impact that daycare had, highlighting that they had not experienced any challenges with childcare relating to their child's IEM.

"[Daycare has] been really helpful. They try to understand and they don't treat [my child] any differently, which is really important to me." – Family Caregiver 8

Similarly, Family Caregiver 19 acknowledged the benefits of having access to daycare and other programming, although they described the struggles they experienced in balancing care for other family members while attending appointments for their child with an IEM when these childcare services were not available due to COVID-19.

"With the COVID and not having a lot of people around and being extra nervous about [my child] getting a cold or a flu, it's been pretty well me and my [partner] doing [all their care]. I haven't really had a lot of care in that end of things. I also have two other kids at home, so one has high- special needs, so taking care of both of them is very tricky actually. [...] Right now, I'm lucky that I can be home with my children. But my other foster [child] has high level needs. So, to get [my child with an IEM] to go to [their] doctors' appointments, plus bringing my other one, I can't go on my own. So, I have to make sure that [my foster child] is in [their] programming and that [my other child] is in daycare, to be able to take them. I don't know if also too COVID has affected that, you know like you're

really not supposed to bring more than one person to their appointment. So, but, my foster [child] right now [they are] not in program, and so I actually have missed an appointment, I couldn't bring them all in.” – Family Caregiver 19

5.2.5 Adapting to Family Caregiving for Children with IEMs

The final aspect of this constructivist grounded theory is the process of family caregivers’ adaptation to accessing care for their children with IEMs (Figure 3e). The intensity of care required by family caregivers typically decreases over time, and they begin to adapt once the tasks of managing care for their child with an IEM become routine and a part of daily life. One coping strategy that family caregivers use in this process is recognizing their own privileges as a family caregiver, whether these be socioeconomic advantages or the metabolic stability of their child’s condition.

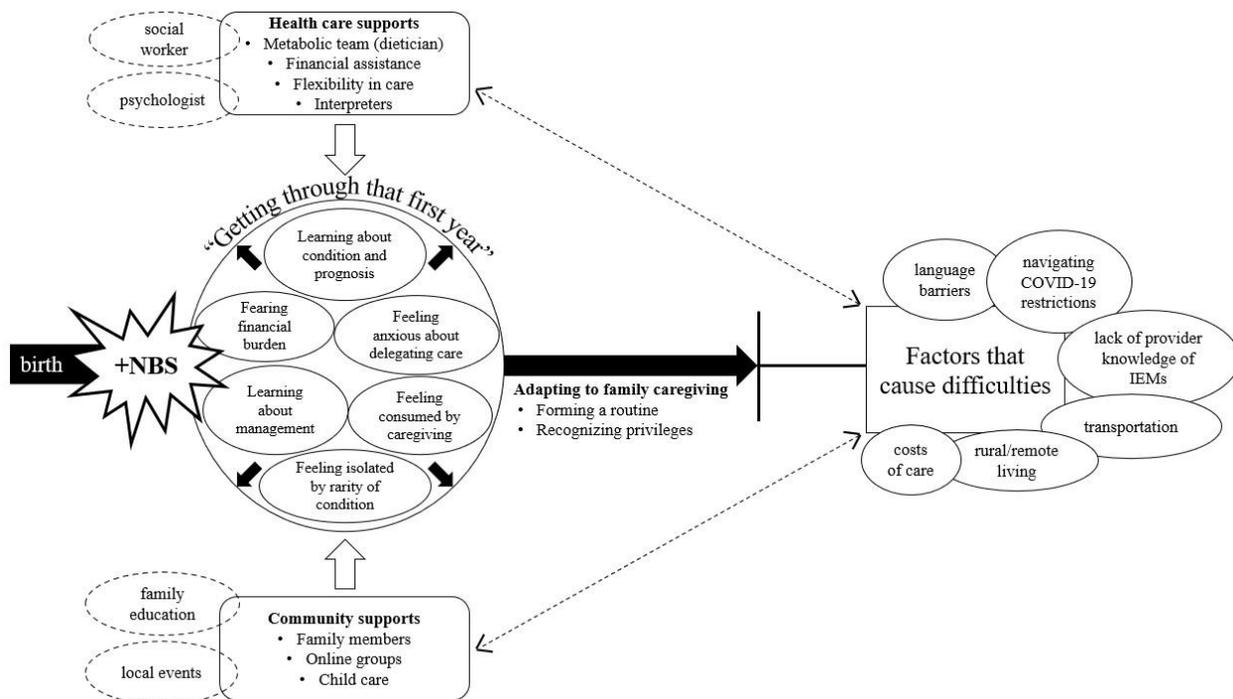


Figure 3e. Theoretical model of the experience of family caregiving for children with IEMs, highlighting the process of adapting to family caregiving for children with IEMs.

Forming a routine

Family Caregiver 8 expressed that caregiving for their child got easier over time as they developed a routine of care requirements at home. They also drew attention to the future when their child will be able to continue this routine on their own.

“As we adapted to the new way of our life it got easier. And now it’s – you don’t even notice that it’s going to be different; [my child] just gets blood tests to make sure that [they eat] more or drink more. [...] [They] have gotten really used to the poke [of at-home blood spot collection] so it’s like [they don’t] even notice; it goes really easy. It’s not like going to the doctors [who] have to, like, strap [their] arm and [they are] all overwhelmed by new people. [...] And when [my child is] older [they’ll] be able to do it [themselves] and then won’t have to worry as much. Because [...] it’ll just be part of [their] routine.” – Family Caregiver 8

Family Caregiver 13 described that while they had gotten into a routine, there were still many ongoing requirements of managing their child’s diet

“Ideally [now that I’m in the habit of measuring everything], there’s no more kind of restraints or difficulties in managing [my child’s] diet. [...] Two years ago, [my child] was able to get onto [medication] which [...] has helped increase [the] amount of protein [they are] allowed to take. So, that is good, and [they are] allowed to eat a bit more regular food. Still doesn’t eat meat, no high protein foods, [they] can have regular pasta now, but... Yeah, for the most part, I still calculate everything [they] eat and measure it out and make sure [they are] taking [the] medicine and formula daily as required.” – Family Caregiver 13

Family Caregiver 24 echoed Family Caregiver 13’s report. They summarized the extensive ongoing tasks of daily care for their child that still consume their daily life, despite their becoming more resilient over time.

“I’ve gotten more resilient because I’ve gotten more confident, and I’ve had the support of the dietitian. But, aside from that, I don’t know... I mean, you just do it because you have to. [...] And [my child] still ... [they do] not have as many appointments as [they] used to have, nor [do they] have as many as other kids with metabolic disorders, but [they do] have more. We’ll have to do yearly metabolic whole blood panel, we have [...] [their] dietitian appointment... we’re having that every couple of months. And then we have one or two metabolic clinic

appointments per year and we've been having them every 6 months. But then on the daily it's ordering [their] food, weighing [their] food, buying [their] food, communicating with school, communicating with daycare, trying to figure out budgeting for [their] food because we get a medical food budget but a lot of the food that [they eat] still comes from the grocery store and it's extremely expensive. Yeah, so there's no spontaneity in my life [laughs] raising a child, everything is planned and budgeted for in terms of [their] protein allowances, where we can eat and what [they] can eat if we go there, so [they have] enough protein to have it, so there's no part of any day with [them] that isn't... where we don't think about food.” – Family Caregiver 24

Recognizing privileges

Family Caregiver 24 acknowledged numerous socioeconomic privileges that they benefit from, listing access to daycare, stable employment, and a personal vehicle as advantages in accessing care for their child, and noting that caring for a child with an IEM would be more difficult without these privileges.

“I've been lucky in that regard, like I didn't have childcare issues for my other [child] because [they were] in daycare. I mean, parking... parking would be a huge barrier in that... like I never did the math of what I paid for that many doctors appointments, but it was a lot of money. It was a lot of money. So, I mean no I didn't financially feel the impact of that, but I think many families would. And I mean it's not that easy to access, like there were many times where I thought I'm lucky to have a job and I'm lucky I have a car, because I don't know how you would do all of this if you didn't have easy access to a car. So, it didn't personally impact me because I had those things, but, I mean, you realize how privileged you are because it's hard and you have all those things. So yeah, like it was expensive. I wouldn't consider it to be like a huge burden for me but I guess I haven't really thought about it, that most people don't spend their money that way.”
– Family Caregiver 24

Similarly, Family Caregiver 23 also noted socioeconomic privileges, sharing that their household income was sufficient to absorb potential unexpected financial impacts of caring for a child with an IEM.

“Our personal income is good, so we could absorb the financial problems if it wasn't supported by those groups. But it helps a lot. Like, yeah. It just alleviates that worry that, you know, what if you don't have enough money next year to pay

*for drugs or whatever? I think, like, just, we don't need to worry about that.” –
Family Caregiver 23*

Additionally, they continued to acknowledge the benefits that they receive from owning their own company and having control over their own work schedule.

“Our works are – we're both – we both own our own companies. Kind of modify our schedule as required to – for time off and stuff. Like, that's never been an issue.” – Family Caregiver 23

Family Caregivers 19 and 13 echoed the advantages of having flexibility in their work schedules, citing the freedom to take time off work and to attend virtual appointments on lunch breaks.

“The time off work is not necessarily [a] problem, and the transportation, we're lucky enough that we have the ability to buy a vehicle and we have our licences and everything like that.” – Family Caregiver 19

“There was never an issue with travel plans. A couple of the virtual appointments, I was in the office at work, but I kind of just had to take my break earlier or later just to coincide with our appointment time. [...] I have also another, [my child with an IEM] has a younger sibling as well. It was never an issue, we would just... I would just take [my other child] with us as well and kind of do what we needed to do. My work was never an issue, like they are very flexible with my hours so luckily, I am able to kind of juggle that around” – Family Caregiver 13

Family Caregiver 17, while sharing that they are lower income, also acknowledged their own socioeconomic advantages in having enough to live comfortably and contemplated how things could be worse if their child's condition were more severe.

*I mean, yeah, like I said it's always been kind of a priority for us and I mean, we're lower income for sure, but at the same time we have enough... we have enough to pay the bills and have food on the table, so it hasn't made a difference in [our child] getting adequate care or not. But, I mean at the same time it is a financial burden and I could see, especially single parent families, really struggling with it. Especially if conditions were worse for the child.
– Family Caregiver 17*

Family Caregiver 17 continued, adding that if their child's condition were less stable and required more frequent follow up appointments, this would likely add further difficulties to the coordination of care.

“I mean, it’s been fairly mild, there hasn’t really been any significant setbacks in [my child’s] diagnosis or in [them]... everything’s been going fairly smooth” [...] Even with the fact that [...] [they have] always been in the healthy range. But you know if things were to get a little rocky, it might make it a little bit harder to track [care].” – Family Caregiver 17

Similarly, Family Caregiver 19 acknowledged privileges related to condition severity, suggesting that the costs of family caregiving would be even higher caring for a child requiring more frequent hospital visits.

*“I’m pretty sure of ourselves right now, like our income has not really been an impact on it. We have like I say, we have insurance, so [my child’s] glucose monitor, [their] test strips, all that, is not really a financial burden for us. And I don’t think that, maybe, that [their] genetic disorder is causing any more financial problem to us than anything else would. We have been to the hospital and there’s your typical expenses there, eating food there, parking, things like that. But I don’t think that’s narrowing down just because [they’ve] got a specific disorder. But I think if we were in the hospital more often, those costs could add up. We’ve just been fortunate enough not to have to go that often.”
– Family Caregiver 19*

Family Caregiver 23 highlighted that caregiver responsibilities and stress would also be higher caring for a child requiring a stricter diet than their own.

“Yeah. I mean, overall my experience has been pretty good. [My child has] been mild on the spectrum, so we haven’t had any crises. We’ve had moments where we’re we weren’t sure. [...] So care giving for us is – [they’re] a pretty normal child. [...] We just basically need to give [them] medicine three times a day – four times a day, sorry. And diet, so [they are] on a protein-restricted diet. So we need to carefully watch and record what [they eat]. So every day, you know, we have a book and we write down what it is [they’ve] been eating. [...] Again, because [they are] mild on the spectrum, there is that we’ve always had a little bit of wiggle room. You know? So, if we had a day where we kind of forgot to write in the book or if one day, you know, [they] went over [their] allocated 10 grams or

whatever, there was never – no fear in that. Right? So, like, [if they were] extreme and always having these crises and stuff, like it could be a lot more of a stressful situation.[...] We were pretty lucky that [they were] so mild.”

– Family Caregiver 23

They continued, acknowledging that a child requiring a stricter diet and more reliant on the use of medical foods would add additional expenses to the cost of caregiving.

“Financially, myself and my [partner], we’re doing well [financially]. Like I just can’t – you know, it would be – it would be a lot if – I mean, [my child] doesn’t eat a lot of the specialty foods – again because [they are] mild. [...] So, you know, if you were dependent on these things. It is very expensive to buy. It’s like they have the price tag on the webpage. And if you had to buy it without it being funded, sponsored, whatever, that – yeah. It would be very expensive.”

– Family Caregiver 23

Family Caregiver 6 added that family caregivers caring for children with less stable conditions or for children with comorbidities such as developmental delay may encounter more difficulties and require more support.

“I have questions, like there are lots of different kinds of genetic disease, right? And [my child] is like... almost like physically, mentally, [they are] well developed. [They have] no problem with [their] mental and physical growth, right. But there are some other genetic diseases that really affect them with their physical and mental growth. So, I don’t know about what kind of help government gives to those kinds of families, so, I think we should really go digging- we should be concerned whether those families are getting enough help or not.”

– Family Caregiver 6

Finally, Family Caregiver 19 shared the lingering thoughts of what caregiving could have been like had their child’s IEM been more severe, even picturing worst-case scenarios.

“We were lucky that nothing happened, like I’ve heard other horror stories like kids have gone into shock and things like that. So, I’m just lucky that I had a big baby, I had a good eater from the very get go. [...] Because I think about the first night that we were home by ourself, before we were aware, and my [child] slept a good four hours. Which I was actually impressed with. And [they were] okay, but other kids have not been okay.” – Family Caregiver 19

Chapter 6: Discussion & Recommendations

6.1 Discussion

6.1.1 Summary of Findings

The primary aim of this project was to explore family caregivers' experiences in caring and accessing care for their child with an IEM in Manitoba, with a goal of highlighting the experiences of family caregivers with low income. The study population, both for quantitative surveys and qualitative interviews, reported a median annual household income (\$54,000) below the provincial median annual household income of \$68,147 (Statistics Canada, 2016). Therefore, this study was somewhat successful in achieving this goal of sampling for family caregivers with lower income, although most participants did not report experiencing financial hardship due to their caregiving responsibilities. As such, it is possible that study participants still had socioeconomic advantages compared to other family caregivers who did not participate in this study, especially as there are many factors impacting the ability of families with low income to participate in research such as stress due to lack of resources, poverty-related family difficulties, time demands, and scheduling conflicts (Schnirer & Stack-Cutler, 2012). However, the focus on family caregivers with low income as outlined in recruitment materials distributed for this study likely had an impact on the experiences shared by family caregivers with higher income as demonstrated by their numerous comments on socioeconomic privilege and reflections on how their experiences may have been different if they had a lower income. Therefore, the wording included in the study recruitment letter has shown to be a strength of this study and has elucidated novel perspectives on the financial aspects of caring for children with IEMs.

While the interview participant group appeared to be fairly representative of the larger survey respondent group, there were some differences between these two groups. The interview participant group had a larger proportion of family caregivers under the age of 30 years as well

as those who had no college or university education and who identified as having ethnic origins other than European. The interview participant group also had a larger proportion of family caregivers who were caring for a younger child, a child who had been diagnosed following positive NBS, and a child who required management in the form of dietary restrictions and medical foods. Finally, the interview participant group also had a larger proportion of family caregivers who reported that access to care was impacted by the COVID-19 pandemic. Some of the family caregivers who were interviewed in this study received their child's positive NBS and diagnosis during the height of COVID-19 and restrictions, whereas others received their child's diagnosis prior to onset of the pandemic. Additionally, as previous literature has demonstrated that younger family caregivers and those caring for younger children with IEMs who require dietary management are at greater risk for decreased wellbeing (Abdelaziz et al., 2019; Hatzmann et al., 2009; Shaji Thomas et al., 2021), it is possible that these family caregivers were more inclined to share their experiences and thus were more likely to be interested in an interview. These factors likely played a role in the development of the grounded theory which was constructed primarily based on interviews conducted with these families.

The constructivist grounded theory presented describes the process of family caregiving for children with IEMs with potential for acute decompensation in Manitoba. This theory evolved from the initial research question which intended to explore family caregivers' experiences specifically in accessing metabolic-related care into an explanation of the broader process of family caregiving for children with these IEMs. The theoretical model presented visualizes the overwhelming expansion of stressors that accumulate especially in the first year of family caregiving for a child with an IEM following positive NBS. The model also highlights the role that health care and community supports can play in alleviating the pressure that family

caregivers face in “getting through that first year”. Finally, the model demonstrates how family caregivers adapt to the demands of caregiving despite the difficulties that “push back” on them throughout this process. While this theory was constructed to describe the process of family caregiving specific to the participants in this study and does not attempt to generalize findings to other populations, many findings are consistent with previous literature. For example, Carpenter and colleagues (2018) described family caregivers’ anxiety regarding delegating care to others in early stages of caregiving. Additionally, Rose and colleagues (2019) reported on the costs of care relating to medical food supplies, and both Carpenter and colleagues (2018) and Packman and colleagues (2007), among others, reported on the difficulties that family caregivers experience due to the lack of knowledge about IEMs from non-genetics providers and the general public. Furthermore, Rajasekar and colleagues (2020) described that while family caregivers gained more experience in managing IEM-related care over time, lack of support was often a forefront issue. Lamoureux and colleagues (2015) and Packman and colleagues (2007) report that family caregivers of children with IEMs would benefit from formal psychosocial supports and financial assistance, respectively. Therefore, these findings support the validity of the theoretical model constructed in this study.

This study also sought to further elucidate factors that facilitate and factors that cause difficulties to accessing care for children with IEMs. Overall, study participants reported being satisfied with care provided by the metabolic team and generally spoke highly of the support received by the dietitian, although they expressed that other roles were missing on the team such as a social worker or a psychologist. This led to many family caregivers relying on the dietitian for psychosocial support and feeling unsupported when the dietitian was not available unless they were able to seek out external support on their own. Additionally, many study participants

found that the benefits of accessing care from home during the transition from in-person to virtual care due to the COVID-19 pandemic was highly valuable due to increased flexibility in care. Similarly, family caregivers found being able to perform other aspects of care, such as blood spot tests, at home relieved some of the strains of accessing frequent care. While many family caregivers reported no difficulties in accessing care, others reported challenges related to the costs of care, lack of knowledge of non-genetics providers, language barriers, transportation, and navigating COVID-19 restrictions. Family caregivers also reported substantial expenses related to their children requiring dietary management. Further, another secondary objective of this study was to explore the perceived impact of the COVID-19 pandemic and related restrictions on access to care. While many family caregivers appreciated the increased flexibility that the transition to virtual care provided, many also reported negative impacts such as fewer physical exams, longer wait times and more coordination for testing and receiving results. Although the final secondary objective of this study was to compare the experiences of urban and rural family caregivers, bivariate analysis did not reveal any statistically significant associations, except that younger family caregivers and those with less Westernized education were more likely to live over an hour away from tertiary care. However, through the experiences that family caregivers shared during interviews, it was clear that living rurally or remotely posed additional challenges to accessing care, such as requiring family caregivers to request additional time off work, increasing travel costs, and prolonging the time to receive important management supplies.

6.1.2 Study Limitations

While valuable and actionable information was collected as part of this study, there are some limitations to consider. First, as discussed in previous studies exploring the experiences of

family caregiving for children with IEMs, the heterogeneity of IEMs included in this study could be seen as a limitation due to the variability in the types of management strategies and level of care required across different IEMs (Arpaci et al., 2020; Chow et al., 2022). Additionally, while specific IEM diagnoses were not reported in this study to protect the identities of study participants and their children, most family caregivers were caring for a child with an IEM at mild risk of decompensation according to expert ratings of IEMs (Gramer et al., 2014). It is likely that family caregivers who care for children with IEMs at higher risk of acute metabolic decompensation would have unique experiences in the process of family caregiving. These family caregivers may be less likely to participate in research due to the potential increase in time and energy required to care for their child. Furthermore, due to the identifiable nature of rare conditions such as IEMs and since the thesis advisor for the student principal investigator is a care provider on the metabolic team, it is possible that participants may have felt pressure to respond to survey questions and interviews differently despite the fact that measures were taken to protect their identities.

This study was additionally limited by the wording of Question 21 in Part B of the survey instrument which explored whether family caregivers had experienced “financial hardship because of their caregiving responsibilities”. As discussed by Arpaci and colleagues (2020), family caregivers may be hesitant to label expenses related to their child’s care as financial burden or hardship due to their emotional attachment to their child. It is possible that a more sensitive way to pose this question would have been, “Did you ever have difficulty making ends meet at the end of the month?” as this would address financial hardship without the perception of placing blame on the child. Furthermore, while this study intended to highlight the experiences of family caregivers with low income, the requirement of participants to complete the survey and

interviews in English due to the lack of an interpreter available could have prevented interested family caregivers from participating in this study. Another factor that could have impacted recruitment was that the honorarium was not advertised until the consent form for individual interviews, meaning that it did not act as an incentive to increase response rate but rather served to thank those who were already interested in participating in an interview and compensate them for their time.

Finally, a limitation in the constructivist grounded theory was the time constraints on data collection which led to all interviews being conducted at once prior to the initiation of theory construction. While some qualitative researchers believe this is acceptable for studies with limited timelines (Timonen et al., 2018), this meant that this study lacked the iterative process of theoretical sampling in which the initial theory was investigated further in subsequent interviews to achieve a theory that is highly grounded in data (Birks et al., 2019).

6.1.3 Future Directions

This study was the first to explore the experiences of family caregivers with low income caring for children with IEMs in Manitoba. While attempts were made to involve a patient partner in this study, future research should strive to engage patients in participatory research. Additionally, there were important areas of family caregiving which were not explored in this study due to time constraints, including the exploration of the social impact of family caregiving on relationships such as friends, immediate family members, and extended family. Future research should explore these dynamics as well as collect data on family structure to investigate the impact of family caregiving for children with IEMs on siblings or other children in the home with or without IEMs and other care needs. While some family caregivers in this study expressed that caregiving for their child with an IEM had an impact on their own health, this was not

explored in detail and could be investigated further in future studies. Finally, as this study focused on the experiences of family caregivers for children with IEMs, a suggested topic of future research would be to explore the financial aspects of adults with low income accessing their own IEM-related health care.

As this topic of research involves family caregivers with low income, it is important to address the shared histories of colonization and disempowerment that have impacted child health outcomes for Indigenous populations (Coombes et al., 2018). Previous research has identified that Indigenous peoples encounter numerous obstacles to accessing health care, including lack of culturally appropriate care, language and cultural barriers, racism, remoteness and distance from health care providers, transportation and financial difficulties, whereas there has been limited research on factors that facilitate access to care (Coombes et al., 2018). As such, future studies should attempt to obtain Indigenous perspectives on family caregiving for children with IEMs by collaborating with interested Indigenous communities. One possible area of future research could be exploring the experiences of First Nations family caregivers caring for children with glutaric acidemia type 1, as there is a known founder population in Manitoba. Preferably, these future studies should be led by Indigenous researchers as there are many limitations and biases of Western views of research methodology and concepts of health (Coombes et al., 2018).

Additionally, family caregivers who are immigrants or refugees to Canada face systemic barriers in accessing health care for their children, including substantial financial barriers (Salami et al., 2020). For example, many immigrants and refugees lack universal healthcare coverage upon their initial arrival to Canada, and although many health services are free, those that are not covered, such as medications and medical foods, can be prohibitively expensive (Salami et al., 2020). Furthermore, immigrants and refugees often have lower income due to

being employed in lower wage jobs (Salami et al., 2020). As well, refugees are often placed by government resettlement programs in suburban areas without access to public transportation or a personal vehicle, transportation can be an added obstacle to accessing health care for their children (Salami et al., 2020). For these reasons, future research should focus on exploring the unique perspectives of immigrant and refugee family caregivers who are accessing care for their children with IEMs.

6.2 Recommendations for Supporting Family Caregivers of Children with IEMs

Many of the difficulties experienced by family caregivers caring for children with IEMs in Manitoba can be alleviated by improving the support they receive at the health care and community levels. The following recommendations are made to improve the experience and wellbeing of family caregiving for children with IEMs based on the findings of the present study as well as previous literature: (1) provide formal psychosocial support, (2) offer support in navigating financial resources, (3) deliver education to family members, (4) organize local community events, and (5) increase flexibility in care.

6.2.1 Recommendation #1: Provide Formal Psychosocial Support

Providing formal psychosocial support to family caregivers of children with IEMs in Manitoba would greatly improve caregiver wellness, especially during the intensifying stress of the first year following positive NBS and diagnosis. Notably, the stresses of this first year can be compounded for family caregivers who have traumatic birth experiences and for those who experience postpartum depression. As decreased caregiver wellness is associated poorer outcomes related to child health (Hassall et al., 2022; Shaji Thomas et al., 2017), improving caregiver wellness should be a priority for the metabolic service. The family caregivers interviewed in this study expressed heavy reliance on the single dietitian involved on the

metabolic care team at Health Sciences Centre to receive their psychosocial support and acknowledged that this was not necessarily the formal role of a dietitian. Furthermore, family caregivers voiced that they did not have elsewhere to obtain support, especially given the all-consuming nature of providing IEM-related care which hindered them from being able to seek out external supports on their own. Previous literature has demonstrated the success of formal psychosocial support programs, with Rajasekar and colleagues (2020) finding that incorporating additional psychosocial supports such as clinic-associated workshops addressing the mental and emotional needs of family caregivers for children with IEMs had led to clear improvements in caregiver wellbeing. In some Canadian metabolic clinics, this psychosocial support is provided by a psychologist integrated with the metabolic team (Lamoureux et al., 2015). We recommend that a psychologist position be integrated with the metabolic team in Manitoba to ease family caregivers' anxiety and stress throughout caring for their child with an IEM, including assisting caregivers as they process their child's diagnosis and adapt to the extreme ongoing demands of managing IEM-related care.

6.2.2 Recommendation #2: Offer Support in Navigating Financial Resources

Offering support in navigating financial resources would benefit family caregivers of children with IEMs in Manitoba by decreasing difficulties related to the many costs of IEM-related care, which would improve caregiver wellness (Arpaci et al., 2020; Packman et al., 2007). Many family caregivers highlighted the significant benefits of receiving financial support from the metabolic team and their provincial government, including provision of management supplies and plans to cover the costs of these supplies and travel. However, these family caregivers, despite their acknowledged socioeconomic privileges, also expressed that obtaining coverage was often difficult and that they required assistance from the dietitian to advocate for

coverage of new medical products and foods. Importantly, one family caregiver who had immigrated to Canada voiced that there is need for formal support to help family caregivers identify and navigate obtaining these financial resources, as many newcomers to Canada are unaware of the programs and benefits that are available to them. Previous literature confirms that family caregivers who are immigrants or refugees often have limited knowledge of the Canadian health care system and resources available, and have substantial financial barriers (Salami et al., 2020). The addition of a social worker on the metabolic team would help family caregivers caring for children with IEMs in Manitoba to overcome these hurdles. While the metabolic service in Manitoba previously had a specific social worker dedicated to the team to provide additional support and to assist families in accessing these resources as described by Hartley and colleagues in 2011, this is unfortunately no longer the case. Social workers are commonly included in other metabolic services, with Lamoureux and colleagues (2015) reporting over 50% of participating clinics having a dedicated social worker position. We recommend that a social worker position be added to the Manitoban metabolic service and that their scope of practice include but not be limited to helping patients overcome these modifiable barriers, for example by assisting with insurance coverage, navigating resources, and addressing other financial difficulties faced by family caregivers of children with IEMs (McBrien et al., 2018).

6.2.3 Recommendation #3: Deliver Education to Family Members

Delivering education directly to family caregivers, their extended family members, and the community at large would improve the wellbeing of family caregivers of children with IEMs. Many of the family caregivers interviewed in this study reported feeling incredibly isolated following their child's positive NBS and diagnosis due to the rarity of the condition and resulting lack of awareness of family members and the general public of IEMs. This had multiple

downstream effects, causing family caregivers to feel anxious and often unwilling to delegate care to others for fear of others' lack of competence to maintain metabolic control, which led caregivers to assume all responsibility of caregiving thus feeling further isolated and consumed by the demands of care. This finding has been reported in previous literature, for example by Carpenter and colleagues (2018) who highlighted that a lack of knowledge on appropriate management had the potential for life-threatening consequences for a child with an IEM. Furthermore, the family caregivers interviewed in this study described the challenges of being required to teach their family members about their child's diet on their own, which is also supported by previous literature describing caregiver anxiety due to the food-oriented nature of many social gatherings which would require cooperation from others to adhere to their child's dietary restrictions (Carpenter et al., 2018; Fidika et al., 2013). If family members were educated on management and engaged early on following positive NBS and diagnosis, they would be better equipped to be more involved in care during the first year when caregivers feel most overwhelmed. This assistance from the metabolic team in educating others who participated in childcare has previously been reported to be highly valued by family caregivers of children with IEMs (Piercy et al., 2017). We recommend that the metabolic service form an education team to deliver education to extended family members by offering support via in-person and virtual group education appointments. This would serve to enhance caregiver wellness by improving ease of communication among family members, creating a wider support system for caregivers, and normalizing life outside the immediate family (Carpenter et al., 2018; Rajasekar et al., 2020).

6.2.4 Recommendation #4: Organize Local Community Events

Organizing local community events would improve family caregivers' experiences of caring for a child with an IEM by raising awareness of IEMs in the general public and helping to connect them to other families with similar experiences. The family caregivers in this study longed for connection with other caregivers who shared experiences in caring for a child with an IEM, which often led them to finding online groups of these caregivers worldwide to share advice on aspects of care such as dietary management and travelling with a child with an IEM. However, these family caregivers longed for groups and events that could connect them within their local communities. One family caregiver who was interviewed highlighted that they previously had the opportunity to attend a local event for their child's IEM and reported it had been a highly beneficial experience that had lasting positive impacts on their experience as a caregiver. We recommend that the metabolic team organize and support the organization of local community events to raise awareness of IEMs and to connect families with similar IEMs, as these events would improve the experience of caregiving for children with IEMs by decreasing the isolation felt by caregivers and improving the knowledge of IEMs in the general public, including among non-genetics health care providers.

6.2.5 Recommendation #5: Increase Flexibility in Care

Increasing the level of flexibility in care provided to children with IEMs would benefit the wellbeing of family caregivers by providing options that decrease the time, energy, and expenses related to travelling to access in-person care. While family caregivers of children with IEMs in this study had mixed preferences on accessing virtual and in-person care, there were clear benefits to continuing to offer virtual appointments to family caregivers who preferred accessing care from home when possible. These findings are supported by the literature, in which

virtual care has been shown to increase caregiver satisfaction by significantly reducing health care-related costs, travel, time off work, and waiting time for appointments (Baumbusch et al., 2022; Ferro et al., 2021; LeBlanc et al., 2020; Rovelli, Zuvadelli, Paci, et al., 2021).

Additionally, virtual care protects children with chronic conditions such as IEMs, with previous literature reporting that virtual monitoring of these children reduces unplanned hospitalizations and medical visits (Ferro et al., 2021). Furthermore, virtual care reduces the risk of exposure to viral infections such as COVID-19 which is especially important for children with IEMs who have risk of acute metabolic decompensation as a result of illness (Ferro et al., 2021; Rovelli, Zuvadelli, Paci, et al., 2021). Where virtual care is not feasible, such as for certain biochemical testing required for children with IEMs, there is a role for home care services to perform sample collection and other required care, such as through the addition of a metabolic nurse to the metabolic service (Brunetti-Pierri et al., 2020). Finally, the experiences shared by family caregivers through interviews in this study revealed a need for greater availability of psychosocial supports and other resources outside of typical office hours or when members of the metabolic team are unavailable, such as having staff available in the evenings or on weekends. For example, a recent study conducted in Italy identified that having an additional dietitian available on Saturdays was beneficial to patients with IEMs as they could better access this care between their work and social lives (Rovelli, Zuvadelli, Paci, et al., 2021). Overall, we recommend that the metabolic team continue to provide virtual care as an option to family caregivers who prefer it and to expand the amount of caregiving that can be managed at home such as home monitoring of blood levels, as well as to strive to increase the availability of psychosocial supports such as the dietitian, psychologist, and social worker.

6.3 Conclusions

This research exploring the experiences of family caregivers with low income caring for children with IEMs informed the construction of a grounded theory to describe their experiences caregiving for their child in Manitoba. The theoretical model developed to visualize this theory highlights five main aspects of the process of family caregiving for a child with an IEM: (1) Receiving a positive newborn screen and clinical diagnosis, (2) “Getting through that first year” following diagnosis, (3) Factors that cause difficulties for family caregivers, (4) Factors that facilitate family caregiving for children with IEMs, and (5) Adapting to family caregiving for children with IEMs. Overall, family caregivers found that caregiving was overwhelming and consuming, especially during the first year following positive NBS, and they outlined substantial costs of IEM-related care such as managing their child’s diet and obtaining other care-related supplies. Family caregivers felt isolated and were lacking psychosocial support as well as assistance with navigating financial resources. However, the transition from in-person to virtual care as a result of the COVID-19 pandemic and related restrictions appeared to ameliorate some of the stress of caregiving by increasing the flexibility of care and providing family caregivers with greater control over their limited time. This research reveals the unmet needs of family caregivers of children with IEMs in Manitoba, especially in the psychosocial and financial domains. We recommend that psychologist and social worker positions be integrated into the metabolic team at Health Sciences Centre in Winnipeg, Manitoba to address these unmet needs and improve outcomes for family caregivers and their children with IEMs. We also recommend that the metabolic service form an education and outreach team to provide education directly to the extended family and community of children with IEMs as well as to organize local events to connect family caregivers and to increase awareness of IEMs among the general public and other non-genetics health care providers. Implementation of these recommendations would improve

the wellbeing of family caregivers and increase equitable access to metabolic care in Manitoba and potentially beyond.

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APPENDICES

Appendix A: Consent to Contact Form

Study Title: Exploring the experiences of family caregivers with low income accessing health care services for children with inborn errors of metabolism

Student Investigator: Katherine Chimney, BSc, Genetic Counselling Student, Department of Biochemistry and Medical Genetics, Rady Faculty of Health Sciences, University of Manitoba

Supervisor: Dr. Aizeddin Mhanni, MD, PhD, Department of Biochemistry and Medical Genetics, Rady Faculty of Health Sciences, University of Manitoba; Clinical Geneticist, Genetics & Metabolism Program, Health Sciences Centre, Winnipeg, Manitoba

The purpose of this study is to explore the experiences of family caregivers who are accessing care for children with metabolic disorders. We are especially interested in hearing the experiences of family caregivers with low income, because previous research has been biased towards families with higher incomes. However, family caregivers of children with metabolic disorders are being invited to participate in this study regardless of income. As a result of the study, we hope to enable genetic counsellors to better support families and advocate for improved access to care.

Participants will be asked to complete a 15-20 minute survey and some participants may be asked to complete a 30-40 minute interview to further explore their experiences of accessing care for a child with a metabolic disorder.

Individuals are eligible to participate if they are the primary family caregiver for a child who has been diagnosed with a metabolic disorder and they would be able to complete the survey and interview in English.

I, _____ confirm that I discussed this study with
(name of clinician)

_____ who meets the above eligibility criteria and agreed to be contacted
(name of potential participant)

by mailing address _____

or by telephone at _____ or email at _____.

Best time to contact the potential participant:

Morning Afternoon Evening Weekend

Signature: _____ **Date:** _____

Appendix B: Study Invitation Letter

Dear Potential Participant,

*My name is Katherine, and I am a student in the Genetic Counselling Program at the University of Manitoba. I am conducting a study to better understand the experiences of family caregivers who are accessing health care services for children with metabolic disorders. **The goal of this study is to enable genetic counsellors to better support families and advocate for improved access to care.***

The information collected in this survey will be part of a research project to help us understand the experiences of family caregivers with low income who are caring for children with metabolic disorders. We are interested in hearing the experiences of family caregivers, especially those with low income, because previous research has been biased towards families with higher incomes. However, any family caregiver receiving this study letter, *regardless of income*, is invited to complete this survey.

We would be very grateful if you would please complete our 15-20 minute survey either by filling out the attached paper survey OR by using the web link provided:

[survey link]

If you choose to complete the paper version of the survey, please use the return envelope and postage provided to return your completed survey by mail.

Your completion of the survey will serve as your consent to participate in this research. You have the option not to complete this survey. Please do not provide personal information regarding your last name or birthdate as this will help to ensure that your answers will not be identified back to you.

This study has been approved by the University of Manitoba Research Ethics Board. If you have any questions about this survey, please contact Katherine or Dr. Aizeddin Mhanni with the contact information below and we would be happy to speak with you.

To access a list of resources, please visit www.GetYourBenefits.ca or contact us if you wish to receive a physical copy of the resource booklet.

Sincerely,

Katherine Chimney
Genetic Counselling Student
Biochemistry & Medical Genetics
[redacted]

Dr. Aizeddin Mhanni
Clinical Geneticist, Associate Professor
Biochemistry & Medical Genetics
[redacted]

Appendix C: Study Reminder Letter

Dear Potential Participant,

You are receiving this letter because you have previously been invited to complete a survey. This letter is to remind you that we would be very grateful if you would please complete our survey about your experience with accessing healthcare for a child with a metabolic disorder. If you have already completed the survey, thank you very much for your valuable contribution to this project. If you are still considering completing this survey, please respond by **January 15, 2022** to ensure that your responses are included in this study.

The goal of this study is to enable genetic counsellors to better support families and advocate for improved access to care.

The information collected in this survey will be part of a research project to help us understand the experiences of family caregivers with low income who are caring for children with metabolic disorders. We are interested in hearing the experiences of family caregivers, especially those with low income, because previous research has been biased towards families with higher incomes. However, any family caregiver receiving this study letter, *regardless of income*, is invited to complete this survey.

We would be very grateful if you would please complete our 15-20 minute survey either by filling out the attached paper survey OR by using the web link provided:

[survey link]

If you choose to complete the paper version of the survey, please use the return envelope and postage provided to return your completed survey by mail.

Your completion of the survey will serve as your consent to participate in this research. You have the option not to complete this survey. Please do not provide personal information regarding your last name or birthdate as this will help to ensure that your answers will not be identified back to you.

This study has been approved by the University of Manitoba Research Ethics Board. If you have any questions about this survey, please contact Katherine or Dr. Aizeddin Mhanni with the contact information below and we would be happy to speak with you.

To access a list of resources, please visit www.GetYourBenefits.ca or contact us if you wish to receive a physical copy of the resource booklet.

Sincerely,

Katherine Chimney
Genetic Counselling Student
Biochemistry & Medical Genetics
[redacted]

Dr. Aizeddin Mhanni
Clinical Geneticist, Associate Professor
Biochemistry & Medical Genetics
[redacted]

Appendix D: Survey Consent Disclosure

Study Title: Exploring the experiences of family caregivers with low income accessing health care services for children with inborn errors of metabolism

Study Description: Thank you for accessing this study. The information collected in this survey will be part of a research study to help us understand the experiences of family caregivers who are caring for children with metabolic disorders. We are interested in hearing the experiences of family caregivers, especially those with low income, because previous research has been biased towards families with higher incomes. However, any family caregiver who received the invitation letter, *regardless of income*, is invited to complete the survey. **The goal of this study is to enable genetic counsellors to better support families and advocate for improved access to care.**

Student Investigator: Katherine Chimney, BSc, Genetic Counselling Student, Department of Biochemistry and Medical Genetics, Rady Faculty of Health Sciences, University of Manitoba

Supervisor: Dr. Aizeddin Mhanni, MD, PhD, Department of Biochemistry and Medical Genetics, Rady Faculty of Health Sciences, University of Manitoba; Clinical Geneticist, Genetics & Metabolism Program, Health Sciences Centre, Winnipeg, Manitoba

Your Role: This survey will ask you a series of questions regarding your experience as a family caregiver accessing care for a child with a metabolic disorder. It will take approximately 15-20 minutes to complete this survey.

At the end of the survey, you will be asked if you are interested in the possibility of participating in a 30-40 minute individual interview to help us better understand your experience of accessing care for your child. You can let us know if you are interested by providing your first name and contact information at the end of the survey for us to contact you OR you can contact us. Your contact information will not be linked to your survey responses and will only be accessible to the study team.

Potential Risks and Benefits: There are very few risks to participating in this study. You do not have to answer any question that makes you feel uncomfortable or that is upsetting. There may not be any direct benefit to you or your child for your participation in this study, but information gained in this study may help other children with metabolic disorders and their family caregivers have improved access to care in the future.

Your completion of the survey will serve as your consent to participate in this research. Your participation is voluntary, and you have the option not to complete this survey.

You are not required to provide any personal information such as your name, email address or telephone number, although this information may be provided if you are interested in being contacted for an interview. Please do not provide personal information regarding your last name or birthdate as this will help to ensure that your answers will not be identified back to you.

All survey responses, including contact information if provided, will be kept confidential. Online surveys will not be able to track your IP address or email address if not provided. Electronic information will be stored as secure files on a password-protected computer. The University of Manitoba Health Research Ethics Board may review study data for quality assurance purposes.

If you have any questions or concerns about this survey, need help filling it out, or would like to participate in an individual interview, please contact Katherine Chimney at [redacted]. You may also contact the study supervisor, Dr. Aizeddin Mhanni, at [redacted].

This study has been approved by the University of Manitoba Health Research Ethics Board. If you have questions specifically about your rights as a study participant, please contact the University of Manitoba Research Ethics Board at (204) 789-3389.

Appendix E: Survey

A survey to explore the experiences of family caregivers accessing health care services for children with metabolic disorders

PART A: The following questions ask about the child you are caring for:

1. **What is your child's current age?**
 ___ Year(s) and ___ Month(s)
 2. **What sex was assigned to your child at birth?**
 Male Female Prefer not to answer
 3. **What is your child's diagnosis?**
 Medium chain acyl-CoA dehydrogenase (MCAD) deficiency
 Glycogen storage disease (GSD)
 Very long chain acyl-CoA dehydrogenase (VLCAD) deficiency
 Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency
 Glutaric aciduria type 1 (GA1)
 Pyruvate carboxylase deficiency (PCD)
 Pyruvate dehydrogenase (PDH) deficiency
 Isovaleric acidemia (IVA)
 Urea cycle disorder (UCD)
 Other, please specify: _____
- Please note that your child's specific diagnosis will be kept confidential in this study and will not be directly reported.*
4. **Was your child diagnosed based on a positive newborn screen?**
 Yes No
 5. **What age was your child when their symptoms first started?**
 ___ Year(s) and ___ Month(s)
 6. **What age was your child when they were diagnosed with this disorder?**
 ___ Year(s) and ___ Month(s)
 7. **Does your child have any additional diagnoses that complicate their care?**
 Yes, please specify: _____
 No
 8. **On average, how many clinic visits (to any relevant specialist) does your child attend per year?**
 In-person: ___ Phone Call: ___ MBTelehealth: ___ Virtual (Microsoft Teams): ___
 9. **On average, how many trips to emergency or urgent care does your child require per year?**

 10. **On average, how many hospital admissions does your child require per year?**

 11. **On average, how many days does your child have to stay in hospital during each admission?**

 12. **How is your child's diagnosis managed? (Please check all that apply.)**
 Dietary restriction, please specify: _____
 Medical formula or foods, please specify: _____
 Medication, please specify: _____
 Vitamins or other supplements, please specify: _____
 Surgery (Example: tube insertion), please specify: _____
 Other, please specify: _____

PART B: *The following questions ask about your experience as a family caregiver for the above child:*

1. **What is your relationship to the child that you are caring for?**
 - Parent
 - Foster Parent
 - Grandparent
 - Sibling
 - Aunt/Uncle
 - Other, please specify: _____
2. **Based on your experience, with 0 being not serious and 10 being extremely serious, how much do you think your child's condition is a serious condition? (Please circle your answer.)**

0	1	2	3	4	5	6	7	8	9	10
Not serious					Extremely serious					
3. **What is your current age?** ____ Years
4. **What is your gender?**
 - Two Spirit
 - Non-binary
 - Woman
 - Man
 - Other, please specify: _____
5. **What is your ethnicity? (Please check all that apply.)**
 - First Nations
 - Metis
 - Inuit
 - European origins
 - Caribbean origins
 - Latin, Central, and South American origins
 - African origins
 - Asian origins
 - Oceania (Pacific Islands) origins
 - Other, please specify: _____
6. **Do you identify as a member of a racialized group? (For example, a community that is subject to discrimination, language barriers, historical trauma, and/or colonization)**
 - Yes No Prefer not to answer
7. **What are the first three digits of your postal code? (For example, if your postal code is "R3E 0W2", please only provide "R3E")**
8. **What education do you have? (Please check all that apply.)**
 - Indigenous knowledge
 - Some high school
 - Graduated high school or equivalent
 - Trades certificate or diploma
 - Some college or university, no degree
 - College/university certificate or diploma below the bachelor's level
 - Bachelor's degree
 - University certificate, diploma, or degree above the bachelor's level

9. Are you currently employed?

- Yes
- No

If yes to the previous question, please list all positions you currently hold:

10. Please choose to answer one of the two following questions:

On average, what is your total **annual** household income before deductions?

\$

OR

On average, what is your total **monthly** household income before deductions?

\$

11. What has helped you access care for your child?

12. What has caused difficulties or has prevented you from accessing care for your child?

13. Has the COVID-19 pandemic itself or related restrictions affected access to care for your child? Please consider either positive and/or negative impacts.

- Yes
- No

If yes to the previous question, has it been a positive or negative impact? (Please check all that apply.)

Positive, please specify: _____

Negative, please specify: _____

The following questions (#14-21) ask about expenses you may have encountered in the past 12 months because of your caregiving responsibilities. These are out-of-pocket expenses that are not reimbursed.

14. In the past 12 months, have you had any out-of-pocket expenses for **home modifications** to accommodate your child's needs?

- Yes
- No

If yes to the previous question, what is your best estimate of these expenses?

\$

15. In the past 12 months, have you had any out-of-pocket expenses for **professional services** for your child's healthcare? (Example: physical/speech therapy)

- Yes
- No

If yes to the previous question, what is your best estimate of these expenses?

\$

16. In the past 12 months, have you had any out-of-pocket expenses for **hiring people to help** with your child's daily activities?

- Yes
- No

If yes to the previous question, what is your best estimate of these expenses?

\$

17. In the past 12 months, have you had any out-of-pocket expenses for transportation, travel, or accommodation because of your caregiving responsibilities?

- Yes
- No

If yes to the previous question, what is your best estimate of these expenses?

\$

18. In the past 12 months, have you had any out-of-pocket expenses for specialized aids or devices for your child's use?

- Yes
- No

If yes to the previous question, what is your best estimate of these expenses?

\$

19. In the past 12 months, have you had any out-of-pocket expenses for prescription or non-prescription medications for your child's use?

- Yes
- No

If yes to the previous question, what is your best estimate of these expenses?

\$

20. During the past 12 months, have you had any other care-related out-of-pocket expenses? (Example: medical diet)

- Yes, please specify: _____
- No

If yes to the previous question, what is your best estimate of these other expenses?

\$

21. During the past 12 months, have you experienced financial hardship because of your caregiving responsibilities?

- Yes, please specify: _____

- No

Are you (and/or another primary caregiver for your child) interested in the possibility of participating in an individual interview to share your experience of accessing care for your child?

- Yes
- No

If yes, please provide your preferred primary phone number as well as an alternate phone number or email address in case we cannot reach you at your primary number.

First Name: _____

Primary Phone Number: _____

Alternate Contact Information: _____

Best time to contact: *(Please check all that apply).*

Morning Afternoon Evening Weekend

Thank you very much for your valuable participation in this survey!

Appendix F: Consent Form for Individual Interview

Study Title: Exploring the experiences of family caregivers with low income accessing health care services for children with inborn errors of metabolism

Student Investigator: Katherine Chimney, BSc, Genetic Counselling Student, Department of Biochemistry and Medical Genetics, Rady Faculty of Health Sciences, University of Manitoba

Supervisor: Dr. Aizeddin Mhanni, MD, PhD, Department of Biochemistry and Medical Genetics, Rady Faculty of Health Sciences, University of Manitoba; Clinical Geneticist, Genetics & Metabolism Program, Health Sciences Centre, Winnipeg, Manitoba

Advisory Committee: Dr. Cheryl Rockman-Greenberg, MD; Dr. Kristin Reynolds, PhD; Melanie Napier, MSc, CCGC

You are being asked to participate in a research study involving an individual interview. This interview will explore your experience as a family caregiver accessing care for a child with a metabolic disorder. Please take your time to review this consent form and discuss any questions you may have before you make your decision. Please also ask the study team to explain if there is any words or information that you do not understand. You may keep a copy of this form for your records.

The student investigator who will be conducting your interview is Katherine Chimney, a graduate student in the Genetic Counselling Program at the University of Manitoba. This research study is being completed as part of this student's Master's program.

Purpose of Study: The goal of this study is to raise the voices of families with low income caring for children with metabolic disorders. We hope that our gained understanding of the experiences of these family caregivers will enable genetic counsellors to better support families and advocate for improved access to care.

Participant Selection: You are being invited to participate in this study because your child is currently receiving care from the Genetics & Metabolism Program at Health Sciences Centre in Winnipeg. As previous research has been biased towards families with higher incomes, we are interested in hearing the experiences of approximately 15 family caregivers with low income. You can participate in this interview if you speak English, and you are the primary caregiver of a child who has a metabolic disorder.

Study Procedures:

- You will be asked to participate in a phone or video conference interview.
- This interview will be 30-40 minutes in length.
- During this interview, you will be asked questions regarding your experience accessing care for your child with a metabolic disorder. These questions will help us understand what factors have helped you access care for your child and what factors have caused difficulties or prevented you from accessing this care.
- The interview will be audio recorded and the student investigator may take notes during the interview. The audio recording will be transcribed by the student investigator and research team for analysis.

Potential Risks: There are very few risks to participating in this study. However, it is possible that talking about your experiences with accessing care for your child might be upsetting, emotional, or distressing for you. You do not have to answer any question that makes you feel uncomfortable or that is upsetting. If you need any additional help or support, you may contact the Klinik Crisis Line (1-888-322-3019), or we will help you find other resources specific to your needs.

Benefits: There may not be any direct benefit to you or your child for your participation in this study, but information gained in this study may help other children with metabolic disorders and their family caregivers have improved access to care in the future.

Costs: There is no cost for you to participate in this study, aside from the time it takes to conduct the interview.

Payment for Participation: Following your interview, you will receive a \$25 pre-paid credit card as a gesture of appreciation for your time. It can be mailed or emailed to you.

Privacy and Confidentiality: All records containing personal information, such as names, email addresses, and telephone numbers will be kept strictly private and confidential during the study. As metabolic disorders are rare conditions, your child's specific diagnosis will also be kept private. Study records will be labelled with a coded ID number, which will be assigned to you so that neither your name nor your child's name will be used in the study records. If the results of this study are presented in a meeting or published, your words may be used to highlight a point. To keep you anonymous, all personal information such as names, places, pronouns, and specific diagnoses will be removed from all study reports so that it will not be possible to identify that you were in the study.

This study will use the Zoom platform to collect data, which is an externally hosted cloud-based service. Their privacy policy is available here: <https://explore.zoom.us/en/privacy/> While the University of Manitoba Health Research Ethics Board has approved using the platform to collect data for this study, there is a small risk of a privacy breach for data collected. We will disable 'Cloud recording' and 'Automatic recording' and enable 'Local recording', meaning the session will be recorded to the local desktop rather than through Zoom. The file will be deleted once the session is uploaded onto our secure server.

All study records (including interview transcripts and audio recordings) will be kept in a secure location accessible only to the student investigator and supervisor. Any databases containing personal information will be password protected using a password known only to the student investigator and supervisor and stored on a secure computer at the University of Manitoba. All study records, including audio recordings, will be kept for 7 years following the completion of the study in September 2022. Only the research team and the Genetic Counselling Program will have access to these files. The collection and access to personal information will comply with provincial and federal privacy legislations.

Safety:

Your confidentiality may be broken if you describe one of the following:

- Harm to yourself or others.
- Abuse or neglect of a child.
- Inappropriate or incompetent practice of a healthcare professional.

Alternatives: An alternative is to not participate in this research study.

Voluntary Participation and Withdrawal: It is your choice to take part in this study. You should not feel any pressure to participate. You can refuse to participate or decide to stop participating in the study at any time and for any reason. If you withdraw from the study, all data you provided will be destroyed. Your decision not to participate or to withdraw from the study will not affect your child's medical care.

Questions: If you have any questions or concerns about this study, please contact Katherine Chimney at [redacted] You may also contact the student supervisor, Dr. Aizeddin Mhanni, at [redacted]. If you have questions specifically about your rights as a study participant, please contact the University of Manitoba Research Ethics Board at (204) 789-3389.

Consent Signatures:

1. I have reviewed and understand all 3 pages of this consent form.
2. I have had the opportunity to ask questions, all of which have been answered to my satisfaction.
3. I understand that by signing this consent form, I have not waived any of my legal rights as a participant in this study.
4. I understand that my records, which may include personal information, may be reviewed by the research team working with the student investigator as described in the Privacy and Confidentiality section of this document.
5. I understand that my participation is voluntary, that I may stop participating in this study at any time, and that my data may be withdrawn prior to publication.
6. I understand that I will be provided with a copy of the consent form for my records.
7. I am agreeing to participate in this study and am providing verbal consent to the researcher to sign on my behalf.

Participant name: _____ Date: _____

Participant phone number: _____

I, the undersigned, have fully explained the relevant details of this research study to the participant named above and believe that the participant has understood and has knowingly given their verbal consent.

Name: _____ Date: _____

Signature: _____ Role in the study: _____

Appendix G: Interview Guide

Thank you very much for your interest in participating in an interview. As you know, I am interested in your experience with accessing metabolic-related health care services for the child you are caring for.

- 1. Tell me about your experience of caring for your child** (*i.e. what is your relationship to the child you are caring for, when and how were they diagnosed, how long have you been caring for them, how is your child's diagnosis managed, and what has caregiving looked like?*)
- 2. What factors have facilitated accessing care for the child that you are caring for?** (*i.e. what has helped you access care when your child needed it or what factors contribute to your resilience as a family caregiver? e.g. help from other supports*)
 - a. What factor is most significant and why?**
- 3. What factors have caused difficulties or have prevented you from accessing care?** (*e.g. transportation, care for other family members, getting time off work?*)
 - a. What factor is most significant and why?**
- 4. How has income been a factor in accessing care for your child?** (*i.e. how does it relate to the factors that have helped you access care or have acted as barriers to care*)
- 5. How has the COVID-19 pandemic and related restrictions changed your experience of accessing care for your child?** (*e.g. did it impact decisions to seek care or travel plans for appointments, did it change access to virtual care through Telehealth or phone appointments?*)

Is there anything else that you would like to tell me about today?

I want to ensure that you have access to the results of this study when they are published. How would you like to receive information about the results of this study? (*e.g. infographic, audio message, news article, brochure or newsletter, paper mail or handed out in clinic?*)

Would you like to be contacted to discuss some of the findings before they are published? (*e.g. to see how my interpretations of the interviews that I am conducting fit with your experience?*)

Topics discussed in these interviews have the potential to be distressing and I want to ensure you have access to supports as a family caregiver and so I would like to provide you with a list of resources today. [Ask social worker to provide links and phone numbers to resources such as crisis lines, food banks, respite services, metabolic support organizations, information for applying to tax benefits, etc.]

Thank you very much for your valuable participation in this interview. We will mail you a \$25 pre-paid credit card as appreciation for your time. What mailing or email address is best for us to use?

Probing Questions:

- *Tell me more about that. What did that mean to you? How has that impacted you? Can you provide me with an example of that?*