



Experience of Parents of Children with Genetically Determined Leukoencephalopathies Regarding the Adapted Health Care Services During the COVID-19 Pandemic

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Abstract

Parents of children with genetically determined leukoencephalopathies play a major role in their children's health care. Because of the COVID-19 pandemic, many health care services were suspended, delayed or delivered remotely with telemedicine. We sought to explore the experience of parents of children with genetically determined leukoencephalopathies during the pandemic given the adapted health care services. We conducted semistructured interviews with 13 parents of 13 affected children. Three main themes were identified using thematic analysis: perceived impact of COVID-19 on health care services, benefits and challenges of telemedicine, and expectations of health care after the pandemic. Parents perceived a loss/delay in health care services while having a positive response to telemedicine. Parents wished telemedicine would remain in their care after the pandemic. This is the first study assessing the impact of COVID-19 on health care services in this population. Our results suggest that parents experience a higher level of stress owing to the shortage of services and the children's vulnerability.

Keywords

leukodystrophy, COVID-19 pandemic, pediatrics, quality of life

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Since early 2020, the pandemic arising from the coronavirus disease 2019 (COVID-19) has placed considerable strain on many hospitals worldwide.^{1,2} Health care communities were forced to adapt to this unprecedented challenge, leading to major changes in the services provided to patients, for instance, the adoption of a telemedicine program over in-person examinations.³

In Quebec, Canada, like many places globally, public health authorities took additional measures to prevent further spread of the virus to patients. Many non-urgent medical appointments were cancelled or delayed. For some time, in-person appointments were only scheduled in urgent situations. To ensure continuity of care, there was a significant shift from in-person medical appointments toward telemedicine, which is defined as medical care delivered via electronic communication between a health care professional and a patient at different locations.⁴

The implementation of telemedicine has been crucial for the care of vulnerable populations, as complications of the severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2) are more common in these populations. Vulnerable populations include individuals with underlying medical conditions, compromised immune systems, and neurologic disabilities, as well as individuals who require special medical care and those who need to be supervised by others.⁵ Even though pediatric patients, including those with chronic and complex medical problems, are thought to be less at risk for complications compared to adult patients, significant concerns for complications exist for children with complex medical issues.⁶ In a study that aimed to describe the most severe forms of COVID-19 in children, 70% of the children admitted to an intensive and high-dependency care unit with a confirmed or highly suspected SARS-CoV-2 diagnosis had an underlying neurologic condition.⁷

Children with leukodystrophies and genetically determined leukoencephalopathies are among the vulnerable and are considered at risk of COVID-19–related complications.^{8,9} Leukodystrophies and genetically determined leukoencephalopathies are a heterogeneous group of more than 100 heritable disorders with different pathologic mechanisms and clinical manifestations.¹⁰ They affect the white matter of the brain with or without peripheral nervous system involvement.¹⁰ Leukodystrophies primarily affect glial cells or myelin sheaths whereas the white matter abnormalities in genetically determined leukoencephalopathies are thought to be secondary to other problems such as neuronal, vascular, systemic, and other disorders.¹⁰ For simplicity, we will use the terms *genetically determined leukoencephalopathy* to represent all genetically determined white matter diseases including both leukodystrophies and genetically determined leukoencephalopathies.

Disease manifestations in genetically determined leukoencephalopathies can be multisystemic, requiring continuous complex care at a tertiary/quaternary health care facility. In general, genetically determined leukoencephalopathies are progressive and commonly present with motor deficits; cognitive disability, and deterioration; dystonia and spasticity; and language, hearing, and visual impairments.^{8,11} Because there is

currently no known curative treatment for most genetically determined leukoencephalopathies, patients' life spans rely on measures established to prevent secondary complications.⁸

Follow-ups of patients with genetically determined leukoencephalopathies before COVID-19 usually consisted of regular appointments at the hospital with numerous medical specialists, including pediatric neurologists, endocrinologists, geneticists, pediatricians, physiatrists, otolaryngologists, and others as well as frequent appointments with occupational, physical, and speech therapists.^{8,12} This multidisciplinary approach optimizes patients' health and ensures that health care professionals can identify and address patients' evolving needs.

In the past, shifts to telemedicine have been deliberately performed in various pediatric contexts.^{13–15} These past transitions often involved research to assess families' satisfaction. In contrast, during the COVID-19 pandemic, changes to families' health care services were made drastically. The experience of parents with vulnerable children relating to the adapted health care systems implemented during the pandemic is yet to be studied. In this project, we sought to explore the extent to which parents think modifications of health care services have affected their families.

The experience of the parents can be defined as all perceptions and lived experiences throughout their health care trajectory.¹⁶ It is based primarily on individuals' subjective assessment regarding the services received.¹⁷ Qualitative studies are best suited to obtain comprehensive data accompanying the rich experiences of the individual.¹⁸ Health care researchers commonly use individual in-depth interviews to explore experiences related to health care settings.¹⁹ For this reason, we chose to conduct in-depth qualitative interviews with parents of children with genetically determined leukoencephalopathies to better understand their experience with the adapted health care services during the COVID-19 pandemic, their changes in quality of life, as well as their prevailing child care needs during this time.

Insights from this study can provide feedback regarding the implementation of telemedicine in pediatric neurology, can help guide future clinical care beyond the pandemic to improve families' quality of life, and can serve as a template for future pandemics.

Methodology

Study Design

To explore the parents' experience and needs during the COVID-19 pandemic, we employed a qualitative interview approach to obtain data that accurately reflects their subjective experience in all aspects of care.²⁰ We conducted in-depth semistructured interviews to cover relevant aspects of the research question while encouraging the parents to freely share their insights relating to these issues.²⁰ The consolidated criteria for reporting qualitative research (COREQ) was used as a reference for designing and reporting this study.²¹

Participants

Eligible parents were identified through the Leukodystrophies and Neurometabolic Disorders Clinic at the Montreal Children's Hospital of the McGill University Health Centre in Quebec, Canada. This clinic is held by a single pediatric neurologist (G.B.), ensuring a systematic and reproducible care delivery approach. Parents were eligible for inclusion if their child had received a diagnosis of a genetically determined leukoencephalopathy and if they were able to speak French or English. Sampling was purposeful, and not random.^{22,23} In order to obtain the most accurate representation of the changes perceived by parents, we made a list of patients followed at the clinic with the greatest number of medical visits and received health care services. We believed that these families were in the best position to provide insightful information because they had a rich experience with the medical system. Then, we contacted families, making sure to obtain a diversified sample with regard to age, diagnosis, and functional limitations of the child before reaching saturation point. Parents caring for young children and young adults, many different diagnoses, and a wide spectrum of functional limitations were represented. Parents were first contacted by email with basic information about the objectives of the study and a description of their contribution in the project if they agreed to participate. Researchers (P.A.Y., M.S., L.T.) then contacted these parents on the phone to provide additional details on the study's research goals and focus. If no response was forthcoming or the family declined participation, no further contact was made. If they agreed to participate, written informed consent was obtained, and an interview was scheduled. Eligible parents were contacted until data saturation was reached.

Data Collection

A semistructured interview guide was developed by the research team to help direct discussion toward research topics (Box 1). The questions were open-ended, not leading, and single-faceted. The interview guide was pilot tested on a parent and they did not suggest any changes. This parent was excluded from the final sample. Participants also filled a socio-demographic form.

BOX 1. Interview Guide (Translated from French)

1. Could you tell me about your experience with the health care system since the beginning of the pandemic?
2. How were your medical appointments conducted since the beginning of the lockdown?
3. How did you find that?
4. How did the pandemic otherwise affect your health care services?
5. Did you face any challenges during this period related to your child's care?
6. How do you feel these changes have affected your life during the pandemic?
7. How do you perceive the long-term care of your child beyond the pandemic?

Semistructured interviews were conducted over the phone between June and August 2020 by a member of the research team (P.A.Y. or M.S.). In families where both parents are highly invested in the care of their child, interviews were conducted with both parents at the same time, or in 2 separate interviews depending on their preferences. In families where one parent was significantly more involved in the child's health care than the other, we conducted the interview with the primary caregiver. Interviews lasted between 16 and 84 minutes and were recorded with an audio recorder and transcribed verbatim by the interviewer. After the initial transcription, a different researcher read the transcript while listening to the recording to ensure accuracy. All interviews were conducted and transcribed verbatim in French. Only selected citations for publication were translated to English. Interviews were conducted until no new information emerged from the data, indicating that data saturation was achieved.²⁴

To ensure trustworthiness of the data, P.A.Y. and M.S. were trained before conducting the interviews to develop their skills in leading semistructured discussions by an experienced qualitative researcher (M.M.) who also closely supervised the qualitative methodology throughout the research. Furthermore, P.A.Y. and M.S. asked clarifying questions throughout the interviews and often summarized the parents' answers back to them to ensure accurate representation of their experience.

Data Analysis

Parents' responses were analyzed thematically using an inductive approach and different themes and subthemes were identified. Thematic analysis is well suited to provide a detailed description of the data in its particular context.²⁵ Transcripts were uploaded to NVivo, a qualitative data management software,^{26,27} and analyzed and coded by P.A.Y. and M.S. independently. Discrepancies were discussed until a consensus was reached, as per standards in qualitative research.²⁸ This also helped refine the existing codes and identify new ones. Once no new concepts emerged from the interviews, a final code structure that reflected all interviews was created, and all transcripts were then reanalyzed with the final codes.

Results

Twelve interviews from 11 families were included in the analysis. Thirteen parents (4 fathers and 9 mothers, including 2 couples) of 13 patients aged from 3 to 23 years participated in the study. Two of the interviewed families had 2 affected children. Demographic data and the children's diagnoses are presented in Table 1. Family annual revenues are not included in the table as it provides sensitive information and, because of the small sample size, could lead to identification of the participants. Annual revenues ranged from CAD\$20 000-24 999 to higher than CAD\$149 999.

We identified 3 main themes during the analysis: (1) perceived impact of the pandemic on received health care services;

Table 1. Parents and Patients Demographics.

Serial number	Gender	Age	Child's age	Child's sex	Child's diagnosis
1	Female	42	17	Female	Alexander disease
2	Female	52	15	Female	Sialic acid storage disease
			22	Female	Sialic acid storage disease
3	Female	38	5	Male	Aicardi-Goutières syndrome
4	Male	39			
5	Male	56	23	Female	Aicardi-Goutières syndrome
6	Female	54			
7	Female	38	6	Male	X-Linked Adrenoleukodystrophy
8	Female	44	9	Female	Cockayne syndrome
9	Female	37	3	Male	X-linked adrenoleukodystrophy
10	Male	42	6	Female	Leukoencephalopathy with thalamus and brainstem involvement and high lactate (LTBL)
11	Male	45	13	Male	Allan-Herndon-Dudley syndrome
12	Female	38	13	Male	Peroxisome biogenesis disorder/Zellweger spectrum disorder-PEX16
			7	Female	Peroxisome biogenesis disorder/Zellweger spectrum disorder-PEX16
13	Female	36	7	Male	Undiagnosed gLE

gLE, genetically determined leukoencephalopathy.

(2) benefits and challenges of telemedicine and the use of technology in health care settings; (3) expectations of health care after the COVID-19 pandemic. All the themes and subthemes are listed in Table 2.

Theme 1: Perceived Impact of the Pandemic on Received Health Care Services

The main impact of the pandemic perceived by parents on their child's care was a loss or a delay in many services. Many of their non-urgent medical appointments including follow-ups with physicians, occupational therapists, physical therapists, etc., were cancelled or delayed. This delay in services caused stress to some parents, as they feared their child would lose some abilities. Motor abilities were a concern as some children previously had frequent appointments with physical and occupational therapists.

Her appointments used to keep her active, but seriously, walking around became really difficult for her. (Parent 12, mother of 2 affected children: a 13-year-old boy and a 7-year-old girl)

However, some parents were relieved by the cancellation of certain services. They considered that this potential loss of abilities was less concerning than the risk of their child being exposed to the virus. Even when services were reinstated, some families decided to further delay some appointments to limit the risks.

We could have seen the physical therapist, but because of COVID, and since our child has a medical condition that makes him at risk, we decided not to. We practice the confinement more strictly than a normal family. (Parent 11, father of a 13-year-old affected boy)

Some parents had to go to the hospital for essential in-person appointments during this period. Because only one person was allowed to accompany the child, parents felt going alone was an additional burden as they could not rely on someone else for support. Another important reported impact was the loss of received home care support services for several months. Home care support services are provided by governmental institutions in health and social services networks, or can be provided by a worker who is hired privately by the user or paid under the service employment paycheck arrangement or direct allowance.²⁹ Parents valued this help because these children often require a continuous and considerable amount of care. Home care was eventually offered back to them, but some families declined it as they feared for their child's safety. In fact, many of the home care employees worked in multiple homes, increasing the risk of spreading the virus. Moreover, parents who usually relied on school for some of their child's physical, intellectual, and social stimulation had to provide for these needs on their own.

For a long time, we were alone at home trying to manage everything and try new things, but it's only since May-June 2020 that help was slowly coming back, but it was since March 2020 that [child's name] lost help, services, everything. (Parent 11, Father of a 13-year-old affected boy)

Theme 2: Benefits and Challenges of Telemedicine and the Use of Technology in Health Care Settings

While many appointments were cancelled or delayed, some departments introduced telemedicine promptly to ensure continuity of care. Parents reported different methods of telemedicine. Some had video consultations with their physicians, and others had regular phone appointments. Telephone follow-ups were done with both parents, or with a single parent, with or without the child. Some telephone appointments were done

Table 2. Themes, Subthemes, and Codes.

Themes	Subthemes	Codes
Perceived impacts of the pandemic on received health care services	Delays and losses in services Impacts of delays and losses in services	Cancelled appointments; prolonged waiting time for services; loss of home care support; loss of school-related services; parental decision to delay services for the safety of their family Management of child's needs on their own; perceived amplification of child's symptoms
	Related feelings	Parental exhaustion; parental relief that appointments are cancelled; parental stress regarding child's potential regression
Benefits and challenges with telemedicine and technology in health care	Methods of telemedicine Advantages of telemedicine Concern about telemedicine	Appointment settings; caregivers involved; child's presence Being in the comfort of their home; saved time due to reduced travel; lessened emotional and physical burden of going to the hospital; more quality time spent with loved ones; saved costs associated with gas and parking; reduced work absenteeism; reduced child exhaustion; improved communication between parents and health care professionals
	Expectations of health care after the COVID-19 pandemic	Desire of implementing telemedicine in their child's long-term care Not all appointments should be conducted via telemedicine; telemedicine should be employed only when physical examination is not necessary; preference of video consultations over phone appointments; telemedicine should be employed only when the child is stable and well known to the physician

gL.E, genetically determined leukoencephalopathy.

when parents were at work, and others when parents were home. Parents felt like this transition of care did not entail major adjustments from their end.

Parents shared their detailed perceptions of the advantages and challenges associated with telemedicine. Telemedicine allowed families to save time and energy, which was the main benefit reported. Time management seemed to be a challenge in most parents' lives as they felt that, before the pandemic, they spent the majority of their time managing their child's numerous medical appointments. Parents highlighted that having appointments at home saved travel time to the hospital, which was particularly life-changing for families living far away. Parents spoke positively about the associated savings with parking and gas costs.

One good thing about COVID is that I had telephone and Zoom consultations, and it's just fantastic! For years, we had to go to the hospital for follow-ups every three months. It's a whole battle every time. (Parent 2, mother of 2 affected girls of ages 15 and 22)

Parents also believed telemedicine decreased the burden associated with work absenteeism. Some of them reported having to miss entire workdays every time they had to go to medical appointments. Telemedicine also saved families from the hardships of going to the hospital, which they reported to be very emotionally and physically draining. Caregivers not only apprehend receiving bad news but also face many logistical challenges such as traveling with a child with physical limitations, bringing appropriate food, simultaneously listening to the physicians while attending to their child's needs, etc. With both the child and the parents being in the comfort of

their home, parents said they could concentrate better during the appointments. They were also more comfortable engaging in the discussion and asking questions to the physicians.

When we have remote consultations, we are in the comfort of our home and our son is surrounded by his stuff, or in my wife's arms, or comfortably installed in his stroller. This makes it easier for my wife and I to concentrate on what is important during the appointment. (Parent 4, father of a 5-year-old affected boy)

The only concern that parents identified related to telemedicine is that physicians cannot physically examine their child. This was particularly a concern among parents with a medically unstable child because they were afraid physicians would miss important clinical features. However, parents felt they could still have access to in-person care in case of an emergency. Some of them had emergencies during this period, and they reported having received state-of-the-art care.

Theme 3: Expectations of Health Care After the COVID-19 Pandemic

The different methods of care employed during the COVID-19 pandemic presented parents with new options for the future of their child's care that they would never have thought possible before the pandemic.

With COVID, we saw the arrival of video and phone consultations, and we discovered that it works really well. When they don't need to physically see my child, having a video or

phone appointment would be miraculous. (Parent 3, mother of a 5-year-old affected boy)

Parents felt telemedicine offered more advantages than challenges, which led them to propose long-term changes in that direction. They thought that telemedicine would contribute greatly to adapt health care services to their unique needs. Parents specified that they did not wish to replace all their in-person appointments with remote ones because they considered the physical examinations to be an integral part of their child's care, especially when the child is unstable. According to them, telemedicine should thus only be encouraged when the main focus of the appointment is answering clinical questions. Parents were convinced that if telemedicine remained post-pandemic, it would increase their quality of life by giving them more time to spend with their child and their loved ones.

Discussion

Parents play a major role in the health care of children with genetically determined leukoencephalopathies. In this study, we sought to understand the experience of parents in response to health care changes that occurred because of the COVID-19 pandemic. This work underlines potential courses of action to improve these families' quality of life with adapted health care services.

Our study shows that parents experienced a delay or a loss of health care services during the pandemic. Despite the negative outcomes of this shortage of services, such as the potential regression of their child's physical abilities, some parents were relieved to reduce the risk of their child being exposed to the virus. In fact, some parents even refused in-person appointments when institutions offered them again. This is worrisome as children with genetically determined leukoencephalopathies require continuous complex care with numerous medical specialists to maximize their life span and quality of life.⁸ Therefore, these delays in health care services and the fear of parents constitute barriers for optimal prevention and management of children's symptoms.

One major change that dramatically impacted parents' day-to-day life was the loss of home care support. This service provided help with household tasks such as cooking and cleaning and/or help with the general care of the child. Parents had to compensate for this loss of service independently, resulting in significant added stress and exhaustion. This finding is concerning as parents who have a child with a rare disease have already reported being overwhelmed with their typical caregiving roles,³⁰ and have been shown to experience a higher level of stress than parents of healthy children.³¹ Higher parental stress was also documented in a study with parents of children with genetically determined leukoencephalopathies.³² Parents in our study reported similar feelings. Therefore, having to take on the responsibility resulting from the loss of these services has added to the perceived stress in the caregivers' lives.

During the COVID-19 pandemic, many institutions have implemented telemedicine to minimize health risks for patients and to ensure continuity of care. In this study, all parents had accessed some form of telemedical care since the start of the pandemic. In general, parents had a strongly positive experience with telemedicine. In fact, parents reported several advantages: improved work-life balance, facilitated time management, decreased costs, increased flexibility, facilitated access for individuals living in rural areas, decreased stress regarding child care, and improved communication with physicians, among others.

Parents, especially mothers,^{30,33} of children with a rare disease face many challenges daily, including changes in work patterns, income, and domestic duties.³⁴ In an Australian study, 38% of parents had to reduce their working hours or had to quit working altogether to attend to their child's needs.³⁴ Difficulties balancing work and caregiver obligations create a significant financial burden on these families.³⁰ Parents we interviewed faced these challenges, and irrespective of the pandemic, some of them had to quit their job to ensure proper care for their child. Telemedicine helped lessen this burden by providing more flexibility for the parents. Being able to attend appointments from the location of their choice, parents save travel time and costs related to going to the hospital. This could make it easier for them to pursue a career and manage other tasks that are not related to their child's care.

Moreover, our interviews highlighted that telemedicine had the potential to contribute to better access to health care services for families. Previous research has indeed suggested that telemedicine improves accessibility to pediatric specialty care.³⁵ Individuals living in underserved, rural areas often have limited access to specialized health care, having to travel long distances for each visit with a specialist.³⁶ Considering that specialized clinics for the care of children with genetically determined leukoencephalopathies are exclusively present in urban areas, telemedicine would benefit families living far away from these institutions. Parents we interviewed who lived in remote areas were concerned by this issue and expressed their wish for increased accessibility of care in their region by maintaining telemedicine services.

The incorporation of telemedicine for routine medical care has perhaps been long overdue for many individuals seeking health care services. A certain unease toward telemedicine among some patients and physicians has perhaps delayed its implementation despite the advancements in technology and its accessibility. Our participants' main concern regarding telemedicine is their fear that physicians might not be able to accurately detect important clinical signs because a complete physical examination cannot be performed. This is also a concern for physicians themselves, as reported in a study that explored the challenges they faced working in a pediatric telemedicine service.³⁷ Because physical examinations are an integral part of the care of children with genetically determined leukoencephalopathies, missing consecutive clinical examinations could represent a threat to the optimal care of patients.⁸ Despite these concerns, parents still strongly expressed their desire to integrate telemedicine

into their child's long-term care. Therefore, efforts should be deployed to evaluate the benefits and the risks associated with the long-term implementation of telemedicine in the care of children with rare and chronic conditions, as it could contribute to enhancing families' quality of life.

Limitations

This study has some limitations. First, research was conducted only with families from a single site (The Leukodystrophies and Neurometabolic Disorders Clinic at the Montreal Children's Hospital). Second, despite the fact that we contacted families of different ethnicities, no caregivers identifying as non-Caucasian consented to participating in this study. Efforts were made to include participants of all ethnicities; however, the relatively small sample size needed for data saturation precluded approaching more minorities for consent to participate. Another limitation is that because the sampling method prioritized parents who regularly attended clinical appointments in the past, the views of those who did not were not acknowledged in the study. This could have led to bias as their experience might have been significantly different. Parents who had less contact with the health care system might have been even more interested in incorporating telemedicine to their long-term care as they were possibly limited by physical distance from tertiary centers, work, or other responsibilities. We noticed that parents had an overall positive experience at the specialized clinic, and many of them could be characterized as very proactive parents. These parents might have been more inclined to participate in the study than parents with a different experience. Therefore, this study offers some lines of thought that have to be reinforced by larger studies, including more diverse samples.

Conclusion

Our study is the first to look at the impact of the COVID-19 pandemic on the health care services received by patients with genetically determined leukoencephalopathies. We found three main themes that emphasize the need for health care institutions to be better prepared for future unexpected events. Parents indicated that telemedicine should remain integrated to the care of their child after the pandemic, as they believed it significantly improved their quality of life. Further efforts should be deployed to evaluate telemedicine's ability as a preventive and therapeutic tool and its impacts on potential related morbidity. Parents appear to have experienced a higher level of stress during the pandemic owing to the shortage of (in-person) services and to the children's vulnerability. However, telemedicine's numerous advantages that they experienced in the same period could be an important factor contributing to reducing their stress. Quantitative characterization of parents' levels of stress in these extraordinary circumstances would thus be necessary. Our findings add to the growing literature on the challenges that caregivers face during the pandemic, and they could guide long-term practices for other pediatric patient

populations, especially those affected by a chronic condition requiring multidisciplinary care.

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Author Contributions

PAY and MLSJ: acquisition of data, analysis and interpretation of the data, drafting the original version of the manuscript; SM, AS, LTT, RMB, CP, BO, MS, BR, SC, JFS, AML, NB, MDD, CTEN: acquisition of data, editing the manuscript for intellectual content; MM, GB: design and conceptualization of the study, acquisition of data, analysis and interpretation of the data, editing the manuscript for intellectual content; all authors: review of the manuscript and final approval. PAY, MLSJ, MM and GB contributed equally to the work described in this article.

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Ethical Approval

This study was approved by the Research Ethics Board of the McGill University Health Centre (project number 2020-6222).

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Supplemental material

Supplemental material for this article is available online.

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