Journal of Child Neurology
2023, Vol. 38(5) 329-335

DOI: 10.1177/08830738231176672
journals.sagepub.com/home/jcn


The Experience of Parents of Children With Genetically Determined Leukoencephalopathies With the Health Care System: A Qualitative Study

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Abstract

Parents of children with genetically determined leukoencephalopathies play a major role in their children's health care. We sought to gain a better understanding of their experience with the public health care system in Quebec, Canada, to obtain suggestions for improving their services, and to identify modifiable factors to improve their quality of life. We conducted interviews with 13 parents. Data was analyzed thematically. Five themes were identified: challenges of the diagnostic odyssey, limited access to services, excessive parental responsibilities, positive relationships with health care professionals as a facilitator of care, and benefits of a specialized leukodystrophy clinic. Parents felt like waiting for the diagnosis was extremely stressful, and they expressed their need for transparency during this period. They identified multiple gaps and barriers in the health care system, which burdened them with many responsibilities. Parents emphasized the importance of a positive relationship with their child's health care professionals. They also felt grateful for being followed at a specialized clinic as it improved the quality of care received.

Keywords

children, leukodystrophy, pediatric, quality of life

Date received: 4 December 2022; revised: 12 March 2023; accepted: 28 April 2023.

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In Canada, rare diseases are defined as disorders that affect fewer than 1 in 2000 people, whereas in the United States, they are defined as disorders affecting fewer than 200 000 Americans.[1] Although these conditions are individually rare, the ~8000 diseases are affecting more than 3 million Canadians and more than 30 million Americans.[1] Many of these diseases are complex and chronic, and they often cause physical, intellectual, and neurologic disabilities, entailing multiple health care needs. Because approximately 80% of rare diseases affect children, many families around the world are burdened by these diseases.[2]

Leukodystrophies and genetically determined leukoencephalopathies form a group of more than 100 heritable disorders with overlapping clinical manifestations.[3] These diseases affect the white matter of the brain with or without peripheral nervous system involvement.[3] Leukodystrophies are thought to primarily affect glial cells or myelin sheaths while the white matter abnormalities in genetically determined leukoencephalopathies are secondary to other pathological mechanisms that are neuronal, vascular, systemic, or else.[3] Here, we will use the term *genetically determined leukoencephalopathies* to represent all genetically determined white matter diseases including both leukodystrophies and genetically determined leukoencephalopathies.

Children with genetically determined leukoencephalopathies spend their whole lives heavily integrated into the health care system, as disease presentation is usually multisystemic, requiring continuous complex care at a tertiary/quaternary health care facility. Patients with genetically determined leukoencephalopathies commonly experience a progressive course and typically present motor (pyramidal, extrapyramidal, and cerebellar), cognitive, speech and language, hearing, and visual impairments.[4,5] No known curative therapy is available for most genetically determined leukoencephalopathies. Patients' life spans therefore rely on measures established to prevent secondary complications.[4] Their require regular appointments at the hospital with numerous medical specialists, including pediatric neurologists, geneticists, pediatricians, physiatrists, and others, as well as frequent appointments with occupational, physical, and speech therapists.[4,6]

Past studies have revealed that parents of children with rare diseases often struggle with having to constantly navigate the health care system. The many gaps in health care services force parents to compensate by assuming multiple roles such as being their child's advocate, case manager, medical navigator, therapist, as well as being a general expert of both the disease and their child's particularities and special needs.[7]

More specifically, quantitative research conducted with parents of children with metachromatic leukodystrophy showed the disease had a significant impact on the quality of life of patients' families.[8] Moreover, studies looking at cohorts of patients and parents of children with a genetically determined leukoencephalopathy showed that patients have a lower quality of life while their parents experience significantly higher levels of stress.[9,10] However, the experience of these parents with the health care system has never been specifically studied. Therefore, this study aims to better understand the stressors associated with their experience in the health care system, identify modifiable factors to improve their quality of life, and obtain suggestions for improving their services.

Qualitative studies are the method of choice to obtain comprehensive data regarding the rich experience of a subject, and individual in-depth interviews are frequently used to portray experiences associated with health care settings.[11,12] We employed a qualitative method to reach our objectives as we believe hearing parents' impressions and insights firsthand through interviews is crucial to understand their experiences.

Patients and Methods

Study Design

We conducted in depth semistructured interviews through a qualitative approach to provide a certain frame to interviews, while allowing parents to freely share their insights. The consolidated criteria for reporting qualitative research (COREQ) was used as a reference for designing this study.[13]

Participants

Parents who were eligible to participate in this study were identified through the Leukodystrophies and Neurometabolic Disorders Clinic at the Montreal Children's Hospital (MCH) of the McGill University Health Centre in Quebec, Canada, which is led by a single pediatric neurologist (GB). Inclusion criteria included being a parent of a child with a genetically determined leukoencephalopathy and being able to speak French or English. Sampling was purposeful, and we selected a diversified sample in terms of diagnosis, age, and functional limitations of the child.[14,15] We chose patients with the greatest number of medical visits and health care services, since we believed these families were the most affected by the different challenges of having to navigate through the health care system. The study was conducted with the same sample that participated in a study regarding the adapted health care services during the COVID-19 pandemic.[16] Both research topics were discussed in the same interview. Recruitment of participants was ongoing during the project until data saturation was met. Emails containing basic information about the study and its implications for participants were sent to eligible parents. When parents showed interest, researchers (PAY, MS, LTT) contacted them on the phone to provide additional details about the project and to answer their questions. No further contact was made to parents who declined participation. Written informed consent was obtained from parents who agreed to participate, and an interview was scheduled and performed.

Data Collection

The research team developed a semistructured interview guide containing open-ended questions to facilitate the discussion and cover all research topics (Box 1). Participants also filled a sociodemographic form. A parent, who was excluded from the final sample, pilot tested the interview guide, and they did not suggest any changes.

Box 1. Questions from the Interview Guide.

1. How do you feel about knowing your child's diagnosis?
2. What was your experience during the diagnostic odyssey?
3. How did you feel about the support you have been offered during this period?
4. How would you define good health care and exceptional health care?
5. What is the most important to you with respect to your child's health care?
6. How do you feel about the services offered to you?
7. Can you describe your experience with your child's health care professionals?
8. What changes would you like to see in the health care system?
9. How would you describe the communication between you and your child's physicians?
10. How would you describe the collaboration between the different health providers?
11. How do you feel about the different needs of your family? Do you feel like they are all satisfied?

PAY or MS conducted semistructured telephone interviews with parents between June and August of 2020. Depending on parents' preferences, interviews were conducted with both parents at the same time, in 2 separate interviews, or with one of the parents only. Interviews lasted between 42 and 108 minutes. They were recorded with an audio recorder and transcribed verbatim by the interviewer. Then, a different researcher read the transcript while listening to the recording to ensure accuracy. No more interviews were performed when data saturation was achieved, as it indicated that no new information was added to the data.[17]

Data Analysis

An inductive approach was used to thematically analyze the interviews, leading to the identification of different themes and subthemes.[18] PAY and MS independently analyzed and coded the transcripts by using NVivo, a qualitative data management software.[19,20] Discrepancies were discussed between researchers until they reached a consensus.[21] A final code structure was created once data saturation was met. All transcripts were then reanalyzed with the final codes.

Results

Twelve interviews with 13 parents (9 mothers and 4 fathers, including 2 couples) were included in the analysis, representing 11 families of 13 patients aged between 3 and 23 years. Two of the families had 2 affected children. Demographic data and the children's diagnoses are presented in Table 1. Family annual revenues ranged from CAD$20 000 to CAD$24 999 to higher than CAD$149 999.

Five main themes were identified through the analysis: (1) challenges of the diagnostic odyssey, (2) difficult access to services, (3) excessive parental responsibilities, (4) positive relationships with health care professionals as a facilitator of care, and (5) benefits of a specialized leukodystrophy clinic.

Theme 1: Challenges of the Diagnostic Odyssey

Parents expressed how emotionally difficult the diagnostic odyssey was for them. The wait before obtaining the diagnosis was long, ranging from a few months to a few years and some parents expressed how "eternal" this period felt. Moreover, not having a diagnosis limited the services offered to families, as certain public programs are only accessible for a specific subset of diagnoses.

> It was long, it took more than 6 months before getting the official diagnosis. In my head, it felt like it took 10 years. (participant QI41)

Parents identified health care professionals' transparency as an important factor to facilitate their experience during this stressful period. Indeed, parents deplored the lack of information provided by the physicians regarding the differential diagnosis, as it

Table 1. Participants and Patients Demographics.

Patient 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 genetically determined leukoencephalopathy

incited some parents to look online where they saw the worst possible outcomes. They believed that more transparency from their physician could have helped them cope better by preparing themselves for the different possibilities, as well as by having a clearer idea of what could be ahead.

Finally, although most of these children have diseases with a poor prognosis, parents clearly expressed their satisfaction of having received the diagnosis, because it allowed them to grieve their situation and finally be able to focus entirely on their child's needs.

Theme 2: Limited Access to Services

Next, parents reported facing many barriers to obtaining the necessary services for their child's needs including rehabilitation and at-home nursing services, as well as financial supplements. Excessive bureaucracy in the public health care system was a major source of parental stress since parents had to spend hours making phone calls and filling out forms to obtain important services. Additionally, parents often had to repeat some of these same procedures periodically, even though their children were known to have neurodegenerative diseases with no expected health improvement.

> I have always said that it is not really about having to live with the diagnosis and the disease, but more about being left to ourselves, to fight for all the services. That's what's really difficult. (participant QI9)

Moreover, even when children were recognized as eligible, services were sometimes insufficient, poorly adapted to their specific needs, or provided after a long delay. Indeed, parents deplored the generalized overcrowding and lack of funding and workforce in the public health system. Some parents even had to turn to private services to ensure proper care, causing economic strain on their families. This strain was further exacerbated by high costs for gas and parking associated with having to regularly attend appointments at the hospital. Furthermore, parents living in remote areas explained how difficult it was for them to balance their work and personal responsibilities with the burden of traveling long distances for medical appointments.

On a more positive note, parents greatly valued the availability of different health care professionals to schedule regular or last-minute appointments with their child. Parents also appreciated professionals who made themselves easily reachable by phone or e-mail.

Theme 3: Excessive Parental Responsibilities

According to participants, having a child with a genetically determined leukoencephalopathy entails many responsibilities as they felt obliged to take on multiple roles. For example, they had to become the coordinator of their child's numerous appointments and health care needs. Moreover, parents felt that many health care professionals were not familiar with genetically determined leukoencephalopathies, forcing them to be the medical expert of their child's condition to ensure proper care. Parents also felt burdened by the responsibility to keep every health care professional informed about the evolution of their child's condition at each medical visit, wishing for more interprofessional collaboration.

Finally, at-home services were reported as being insufficient. Children with genetically determined leukoencephalopathies often have limited autonomy, and therefore parents were left to constantly provide care to their children. For example, in addition to feeding, bathing, and supervising their child, parents also had to cognitively stimulate them and perform rehabilitation exercises.

> It often feels like I am his medical secretary. It really is a full-time job to have a handicapped child. (participant QI15)

All these different roles made it difficult for parents to have a proper work-life balance, often leading them to abandon their career dreams. These excessive responsibilities also limited the quality time they could spend with their children and their loved ones, affecting their mental health.

Theme 4: Positive Relationships With Health Care Professionals as a Facilitator of Care

Parents expressed that their relationships with health care professionals played a major role in their experience with the health care system. Parents valued physicians who were great listeners and who were sensitive to their particular family situation. They also expressed how comforting it was to have physicians who showed great investment in their child's case. Parents understood that the rarity of their child's condition made it unlikely for all health care professionals to be experts in this field, but they still greatly appreciated professionals who would make an effort to inform themselves before meeting them. Finally, parents knew there were a lot of uncertainties and saddening news involved in the evolution of their children's diseases, but these times were less difficult if they had a positive trusting relationship with the professionals.

> Good health care services start by listening to what the family wants and taking the time to understand their unique situation. There are so many different ways to see the disease, which makes it important to listen to the families' priorities. (participant QI41)

Theme 5: Benefits of Being Followed at a Specialized Leukodystrophy Clinic

Finally, parents expressed their great appreciation of being followed in a specialized leukodystrophy clinic. Indeed, parents saw numerous advantages of having access to this resource as it provided them with valuable additional expertise on their child's specific condition. Indeed, this clinic rapidly became

their primary reference for all their health-related questions and it helped them understand their child's disease better through more elaborate explanations. They also reported better organization and coordination of their care through this clinic. Moreover, parents valued the different research opportunities offered, such as access to new clinical trials, and the increased accessibility to international experts when necessary.

> Being referred to a specialized clinic is absolutely priceless. It's comforting for families. (participant QI5)

Discussion

This study highlights the challenges that parents of children with genetically determined leukoencephalopathies must cope with, starting at the very beginning of the diagnostic odyssey. They first have to face the uncertainty linked to the medical evolution of their child, which is then quickly followed by the emotional burden of caring for a child with a life-limiting disease. At the same time, these parents have to quickly figure out how to navigate the health care system as their child's numerous medical appointments rapidly become an integrative part of their everyday lives. Because of their extensive involvement within the health care system, having health care providers who can better understand their experience forms a crucial aspect of their frequent visits. Therefore, this study aimed to identify modifiable factors linked to their services that would improve their quality of life and quality of care.

Participants identified several shortcomings in the health care system that increase their parental responsibilities on a daily basis such as excessive bureaucracy, insufficient home care support, and long waiting lists. These excessive responsibilities cause emotional distress and exhaustion, making them feel abandoned and alone in their situation. This perception has been reported before in studies involving parents of children with other rare diseases. Indeed, in an online survey filled by parents in Australia and New Zealand, 45% of responders felt unsatisfied by the level of support they had overall received, and as much as 53% had felt anxiety and fear during the week prior to the survey, shedding light on parents' and families' multiple unmet needs.[22] The lack of services for families has also been discussed in a qualitative study with parents of children with Rett syndrome. Indeed, parents in this study deplored the difficulties of trying to obtain specialized equipment for their child through public social aids.[23] This was also the case for parents in our study, some of whom had to turn to private health care services to obtain services, adding financial strain on their families. In another Australian survey, 77% of the participating parents of children with rare diseases reported having received financial assistance but 52% believed it was still insufficient for their family situation.[24]

All of these responsibilities forced many parents to either significantly reduce their work hours or quit their jobs altogether. In families interviewed, mothers seemed more affected by this, and they expressed their dissatisfaction of not being able to pursue their career goals. They believed that if they had access to more services, they would be able to thrive in their professional lives. This finding is in line with other studies on pediatric rare diseases highlighting the challenges of thriving in work environments as a parent of a child with a rare disease. Furthermore, being a mother has also been found as a factor decreasing parental quality of life.[25,26]

Parents highlighted how the excessive bureaucracy and the narrow diagnostic criteria surrounding access to services often excludes children with genetically determined leukoencephalopathies and those still waiting for their diagnosis. This is a common concern for children living with rare diseases. In Currie and Szabo's study, children with rare neurodevelopmental diseases were also often excluded from services as they would not correspond to specific medical and behavioral manifestations.[27] Public health services seem more designed for periodic health incidents than for complex disease courses such as genetically determined leukoencephalopathies.[28,29] A suggestion for increasing access to services is to modify criteria to base them more on functional needs than on diagnostic labels.[25]

Our study sheds light on parents' satisfaction of being followed at a specialized leukodystrophy clinic. Indeed, being followed in a specialized clinic seems to have strengthened parental trust toward the management of their child's case, and also toward the medical information provided. This is an important finding as parents of children with rare diseases often reported having to find information online to properly understand their children's diseases.[25] Other studies on patients with rare diseases have also demonstrated great satisfaction linked to being followed in specialized centers, because of the same perceived advantages such as ease of access to medical staff, regular checkups, and better explanations.[30] Moreover, the rise of telemedicine following the COVID-19 pandemic could facilitate the access to these type of clinics for patients living in remote areas.

Parents in this study also highlighted the importance of their relationship with the different health care providers. Previous literature has undeniably established the importance of communication skills and sensitivity from practitioners in health care settings for increasing patients' satisfaction, and those are skills that could be found irrespective of the level of knowledge about a rare condition.[29,31,32] Our parents valued health care workers who viewed them as experts of their child's condition, because it helped foster a healthier partnership of care to reach the best standard of care. Our results are also in line with the findings of a qualitative study with parents of children with rare diseases where they felt like they were being silenced within interactions with care providers who often had limited knowledge about the experience of caring for an affected child at home.[7]

Finally, the majority of the shortcomings families perceived as essential to address to improve their experience in navigating the health care system were related to services received outside of the specialized leukodystrophy clinic. An important suggestion provided by families was to integrate a social worker or a nurse coordinator in their standard of care. This would provide support for organizing their child's appointments and optimizing their care, which could lessen the burden that was described by families. This service could be integrated into the

leukodystrophy clinic, which parents have already described as the central pillar of their care. Implementing this additional service may however be challenging within the framework of Quebec's public health care system.

Limitations

Several limitations of this work can be discussed. First, only families followed by the Leukodystrophies and Neurometabolic Disorders Clinic at the Montreal Children's Hospital were contacted for this research. It would have been valuable to obtain the points of view of families followed in other centers. Second, the relatively small sample size arising from the data saturation point limited the sample diversity from a cultural point of view. Although efforts were made to include parents of different ethnicities, only Caucasian families participated in this study. Furthermore, our sampling method, which prioritized parents of children with frequent health appointments, might have affected the results. Also, we noticed that parents who agreed to participate in this study were generally very proactive and invested in their child's health care. This might have affected the results as their experience can differ from the ones of less available parents, especially those living further away from our center, who could have been less inclined to participate. Finally, although similar results were reported in other qualitative projects around the world with parents of children with rare diseases, Quebec's health care system is public and the findings might not be translated to other health care systems, such as private health care systems.

Conclusion

In conclusion, this is the first study evaluating the experience of parents of children with a genetically determined leukoencephalopathy with the health care system. We identified 5 themes among which a few modifiable shortcomings were revealed that can help guide future clinical practices to improve families' quality of life. Excessive bureaucracy constitutes an important barrier to obtaining the necessary services for their children. Moreover, the multiple burdens linked to all the roles parents have to assume constantly weigh on their shoulders. By focusing on these burdens and on the relationships with the physicians, it would be possible to improve parents' quality of life. Indeed, whether at the time of diagnosis or throughout the medical follow-up, parents place a great deal of importance on transparency and personal investment of health care professionals in their child's case in order to generate a climate of trust that facilitates their experience. Being referred to a specialized leukodystrophy clinic also contributed to improving their care. This project adds to the growing literature on parents' experience of caring for children with rare diseases, and it will help to better address the specific needs of this unique subgroup of diseases.

Acknowledgements

The authors would like to thank all families involved in this study. The time and dedication they devoted to participating in this study is invaluable and greatly appreciated.

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, MDD, CTEN: acquisition of data, editing the manuscript for intellectual content; MM, GB: design and conceptualization of the study, acquisition of data, analysis andinterpretation of the data, editing the manuscript for intellectual content; all authors: review of the manuscript and final approval. PAY and MLSJ contributed equally to thisarticle. MM and GB contributed equally to the supervision of this work.

Declaration of Conflicting Interests

The author(s) declared the following potential conflicts of interest with respect to the research, authorship, and/or publication of this article: LTT currently manages sponsored clinical trials at the site level for Ionis Pharmaceuticals (Alexander disease clinical trial 2021-present), Passage Bio (Krabbe disease and GM1 gangliosidosis clinical trials, 2021-present), and Teva Pharmaceuticals (chronic and episodic migraine clinical trials, 2022-present). He also manages a GM1 gangliosidosis natural history study sponsored by the University of Pennsylvania with funding from Passage Bio. GB is/was a consultant for Passage Bio Inc (2020-2022) and Ionis Pharmaceuticals (2019). She is/was a site investigator for the Alexander's disease trial of Ionis Pharmaceuticals (2021-present), Metachromatic leukodystrophy of Shire/Takeda (2020-2021), Krabbe and GM1 gene therapy trials of Passage Bio (2021-present), GM1 natural history study sponsored by the University of Pennsylvania with funding from Passage Bio (2021-present) and Adrenoleukodystrophy/Hematopoietic stem cell transplantation natural history study of Bluebird Bio (2019), a site subinvestigator for the MPS II gene therapy trial of Regenxbio (2021-present) and the MPS II clinical trial of Denali (2022-present). She has received unrestricted educational grants from Takeda (2021-2022). She serves on the scientific advisory board of the Pelizaeus-Merzbacher Foundation, the Yaya Foundation Scientific and Clinical Advisory Council, and is the Chair of the Medical and Scientific Advisory Board of the United Leukodystrophy Foundation. She is a member of the Vanishing White Matter Consortium, the MLC Consortium, the H-ABC Clinical Advisory Board, and the Chair of the POLR3-related (4H) Leukodystrophy Consortium. She is on the editorial boards of *Neurology Genetics*, *Frontiers in Neurology—Neurogenetics*, and *Journal of Medical Genetics*.

Ethical Approval

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

Funding

The author(s) disclosed receipt of the following financial support for the research, authorship, and/or publication of this article: This project was funded by the McGill University Health Center Department of Medicine, the Estate of Daphne Joan Townsend Dale, and the Montreal Children's Hospital Foundation. AS has received funding from the Desjardins Studentship in Child Health Research through the Research Institute of McGill University Health Centre, the Healthy Brains for Healthy Lives Graduate Student Fellowship, as well as a Graduate Excellence Award from the Integrated Program in Neuroscience at McGill University. GB has received the Clinical Research Scholar Junior 1 award from the Fonds de

Recherche du Quebec–Santé (FRQS) (2012-2016), the New Investigator Salary Award from the Canadian Institutes of Health Research (2017-2022), and the Clinical Research Scholar Senior award from the FRQS (2022-2025).

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