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Pediatric Neurometabolic Disorders: Medical Needs, Service Use, and Impact of Disease on the Family

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Pediatric Neurometabolic Disorders: 
Medical Needs, Service Use, and Impact of Disease on the Family

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Abstract

Background: Inborn errors of metabolism include many rare genetic disorders with significant morbidity and mortality. A subtype of these disorders, neurometabolic diseases have a variable presentation with both neurologic and non-neurologic symptoms. Presently, few evidence-based guidelines exist to standardize patient care across centers and providers. Due to the absence of such recommendations, the purpose of this study was to characterize the medical needs, service use, and family impact of pediatric neurometabolic disease. Methods: An online survey was created to assess these areas. The study population was comprised of primary caregivers of children (ages 0-17) managed in a neurometabolic clinic at Cincinnati Children’s Hospital. Study invitations were sent to 132 eligible caregivers. Results: In total, 25 surveys were included in analysis resulting in a participation rate of 19%. Known diagnoses were reported for 60% of children with the most common being mitochondrial disorders, metachromatic leukodystrophy, and Batten disease. The most frequent medical problems were due to neurological (88%) and musculoskeletal symptoms (80%). Health concerns and functional limitations resulted in increased needs for services such as therapies (100%), educational supports (85.7%), and specialty medical care (75%). Financial stress (67%) was the most commonly reported factor impacting caregiver stress level. Many support services were desired by the cohort with 60% indicating they would like to be involved in support groups. Conclusions: This study provides evidence to better inform the care and genetic counseling needs of children with neurometabolic disorders and their families. The data delineates the medical needs and service use of this population, supporting the idea that multidisciplinary clinics may help to comprehensively meet their complex needs. Caregivers of children with neurometabolic disorders face a significant burden of care and could benefit from increased availability of support services.

Keywords: primary caregiver survey, pediatric neurometabolic disorders, medical needs, service use, family impact of disease
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INTRODUCTION:

Improvements in the medical care of children with complex health care needs in the last half of the century has resulted in a growing demand for specialized pediatric care [1, 2]. In 1998, the Maternal Child Health Bureau proposed a definition of this group of children, referred to as “children with special healthcare needs (CSHCN)—those who have or are at increased risk for a chronic physical, developmental, behavioral, or emotional condition and who also require health and related services of a type or amount beyond that required by children generally” [3]. Prior research indicates that up to 18% of children in the United States are considered CSHCN [4]. The term “children with medical complexity (CMC)” is a sub-classification of CSHCN coined by Cohen et al (2011) to describe children with marked intensive care needs and substantial medical fragility [5]. The researchers proposed that these pediatric patients likely have similar patterns of needs, chronic conditions, functional limitations, and health care use.

Given the fact that many metabolic conditions have the onset of disease symptoms by childhood and account for a significant proportion of overall pediatric illness, it is possible many of these children could fit the definition of CMC [5, 6]. As there is currently limited research into the needs of children with biochemical disorders and their families, using previous data on CSHCN and CMC can help to guide specific research and elucidate similar patterns of needs.

Inborn Errors of Metabolism

First described by Sir Archibald Garrod in 1908, inborn errors of metabolism (IEM) comprise hundreds of genetic and biochemical disorders [7]. While each disease classified as an IEM is individually rare, their collective numbers and impact on pediatric morbidity and mortality are significant. Several studies looking at the incidence of IEM in the population have concluded that these disorders affect 15.7 – 40 per 100,000 births [8, 9].
Much research has focused on the impact of and treatment for pediatric patients with IEM, however, these conditions can affect individuals throughout the entire lifespan [10, 11]. The clinical presentation of IEM is highly variable with symptoms manifesting either acutely or progressively, depending on the disease. While every body system can be affected by IEM, there are certain organ system manifestations that are highly associated with this group of conditions, including problems with the central nervous system, skin, eyes, and musculoskeletal system [6, 12, 13]. Due to the relatively small number of patients diagnosed with each unique IEM, few evidence-based practice guidelines exist to standardize patient care across centers and providers [14, 15]. Multidisciplinary approaches to treating these patients can therefore be beneficial to help address the diverse medical and psychosocial needs for both patients and their families in the absence of such clinical guidelines [16, 17].

**Neurometabolic Disorders**

Neurometabolic disorders comprise a subset of inborn errors of metabolism in which metabolic disease primarily affects the central and peripheral nervous system. These disorders include aminoacidopathies, organic acidurias, mitochondrial, lysosomal, polyglucosan, and peroxisomal diseases [18]. Because many neurometabolic disorders are neurodegenerative, a significant number of children presenting with progressive neurological and intellectual decline do have an underlying neurometabolic disease. Two studies looking at the epidemiology of progressive intellectual and neurological decline in children identified a metabolic cause of symptoms in 60% and 68% of children, respectively [19, 20]. One of the studies, conducted by Verity et al (2009), concluded that the three most common diagnostic groups in their study were: leukoencephalopathies, neuronal ceroid lipofuscinoses, and mitochondrial diseases [21].

Symptoms of neurometabolic disorders are highly variable and often non-specific in their clinical presentation. These symptoms can imitate more common disorders if there is not a high index of
suspicion for an IEM [22]. Workup for a neurometabolic disorder may be indicated with a complex disease presentation including neurological and non-neurological symptoms [12, 13, 20, 22, 23]. Non-exhaustive lists of possible neurologic and non-neurologic symptoms of neurometabolic disease are outlined in boxes 1 and 2.

Diagnostic testing and treatment options for neurometabolic conditions have grown demonstrably in the last few decades [9, 13]. However, these conditions are still associated with high rates of both morbidity and mortality, supporting the need for early diagnosis, genetic counseling, and appropriate medical management for patients and their families [20, 24]. As an example, a study by Bonkowsky et al (2010) looked at the burden of pediatric inherited leukodystrophies. Using retrospective data, an overall mortality rate of 34% was determined with the average age of death at 8.2 years. Although a final diagnosis was unable to be identified for 51% of enrolled pediatric patients, there was similarity in presentation and needs across the study cohort, including: epilepsy (49%), gastrostomy tube use (43%), and developmental regression (32%). Additionally, average yearly medical costs per patient reached $22,579 in 2008 [25]. Given this high burden of neurometabolic disease, it is suggested that multidisciplinary management and counseling for children with neurometabolic disorders and their families may help to address their unique circumstances [16, 17, 23].

The Neurometabolic Clinic at Cincinnati Children’s Hospital Medical Center (CCHMC) sees patients with suspected or diagnosed metabolic diseases of the nervous system. Notably, many children with mitochondrial disorders and lysosomal storage disorders are managed in different clinics despite their diagnoses fitting into the category of neurometabolic diseases. This multidisciplinary clinic currently includes five specialties: genetics, neurology, social work, nutrition, and physical medicine. Children seen in this clinic have a high diversity of needs with significant variability in their health and neurodevelopmental outcomes.
Despite the increasing testing and therapeutic options for children with known and suspected neurometabolic diseases, there continues to be a paucity of literature on the care needs of this specific population of children. The purpose of our study was to survey primary caregivers of children managed in the Neurometabolic Clinic at CCHMC in order to help describe the medical needs, service use, and family impact of pediatric neurometabolic disease.
Materials and Methods:

Study Participants

Participants were identified via a query of the CCHMC electronic medical records, yielding a preliminary group of all patients seen in the Neurometabolic Clinic since October 2010. Primary caregivers of adult patients, deceased patients, and patients seen for consultation but not managed in the clinic were not eligible to participate. The remaining caregivers of 132 children were invited to complete an online survey about their child’s medical needs, service use, and the impact of neurometabolic disease on the family. These individuals were asked to verify their eligibility based on four inclusion criteria listed on the first page of the survey, including: (1) the participant was the primary caregiver (defined as the individual who provides the majority of the care) of a child receiving medical management at the Neurometabolic Clinic at CCHMC, (2) the child was between 0-17 years of age at the time of survey completion, (3) the primary caregiver was at least 18 years of age and (4) the primary caregiver was able to read and write in English.

Recruitment and Data Collection

Multiple recruitment methods were used to optimize the sample size, including: (1) up to three paper invitations mailed to the child’s home address, (2) invitations given directly to families by the Neurometabolic Clinic staff at their follow-up appointments, and (3) up to three electronic invitations emailed to the child’s caregiver. All invitations provided to families contained a link to the online survey. Each family’s contact information was obtained from their child’s electronic medical records. Due to inaccurate or incomplete contact information, some families did not receive invitations through all three recruitment methods. However, all 132 families invited to participate in the study did receive at least one invitation and some families may have received up to seven total invitations. It was possible eligible
participants may have received additional invitations after completing the survey since the study was anonymous and we were not able to limit follow-up invitations to non-responders.

Data collection occurred between September 2013 and December 2013. The survey data was collected and managed through the secure online database REDCap (Research Electronic Data Capture) version 5.5.8, an electronic data capture tool hosted through CCHMC [26]. An information sheet on the first page of the online survey explained the purpose of the study and indicated that completion of the survey served as implied consent. This study was approved by the CCHMC Institutional Review Board. The requirement to document signed, informed consent was waived for this survey.

**Study Measures**

This research used a cross-sectional, descriptive study design. Following a review of the literature, the study team developed a custom survey tool to assess the medical needs, service use, and impact of pediatric neurometabolic disease on the family (see Appendix B). Many survey questions were adapted from the National Survey of Children with Special Healthcare Needs [27] and the Illinois State Waiver for Medically Fragile and Technology Dependent Children [28]. The survey went through several rounds of revision with input provided by specialists in clinical genetics, neurology, genetic counseling, statistics, and public health. The data collection tool was then pre-tested for readability among a small group of genetic counselors and students at CCHMC. The final survey consisted of 93 close and open ended questions with electronic skip-logic integrated to allow for participants to skip questions not applicable to their child or family. Survey questions were separated into the following eight sections:

1. Primary Caregiver and Child Demographics
2. Health Conditions and Concerns
3. Functional Needs
4. Education, Special Therapy, and Service Use
5. Access to Healthcare and Health Insurance Coverage
6. Care Coordination and Patient and Family Centered Care
7. Family Impact
8. Neurometabolic Clinic Experience

Data Analysis

Descriptive statistics were calculated for both continuous and categorical close-ended questions using Microsoft Excel 2010 [29]. Continuous variables were analyzed for means and standard deviations while categorical variables were analyzed for corresponding counts and frequencies. Open-ended questions were post-coded and analyzed for themes which were agreed upon by two members of the study team after analysis of all survey data.
RESULTS:

Study Participation

Of 132 families invited to participate in the study, 26 primary caregivers completed at least part of the online survey. One survey was only partially complete but the three completed sections (caregiver and child demographics, health conditions and concerns, and functional needs) were included in analysis. Additionally, one survey was completed but responses to open ended questions indicated that that family has not been seen in the Neurometabolic Clinic and thus the data was excluded from analysis for not meeting the study inclusion criteria. The overall participation rate for this study was 19%.

Section 1: Caregiver and Child Demographics

Table 1 and 2 provide summary statistics of the demographic characteristics of both primary caregivers and the child who is seen in the Neurometabolic Clinic. All but one of primary caregivers was a biological parent and all caregivers were mothers. The majority of caregivers (60%) indicated that their child has received a diagnosis with the three most frequent diagnoses being mitochondrial disorders (n=4), metachromatic leukodystrophy (n=3), and Batten disease (n=3).

Section 2: Health Conditions and Concerns

Caregivers identified the health conditions and concerns their child has experienced in the past 12 months (graph 1) indicating significant variability in the symptoms reported for each child (table 3). Of the top five body systems in which symptoms were reported (neurological, musculoskeletal, gastrointestinal, otolaryngological, and ophthalmologic) certain symptoms were more frequently experienced by the population (described in detail in table 4).

In addition to symptoms reported for each child, primary caregivers were also asked to indicate their primary concerns related to their child’s health in the last year. Primary caregiver responses were
coded into response themes and the top five areas of primary concern were: neurological (36%), musculoskeletal (28%), respiratory (20%), pain (20%), and gastrointestinal (16).

**Section 3: Functional Needs**

Due to the complex care needs of many of the children managed in the Neurometabolic Clinic, caregivers were asked to indicate if their child uses devices, equipment, or monitors to support their functional limitations (table 5). The most commonly reported needs by category included: vision, glasses/contacts; hearing, none; communication, electronic device; feeding, gastrostomy tube; mobility, wheelchair or braces; medical devices/monitors, suction pump. Table 5 also shows those functional areas in which caregivers believe their child’s needs are unmet. While unmet needs were reported for 12% or less of children in the areas of vision, hearing, feeding, and medical devices/monitors, a higher percentage of children were reported to have unmet needs in the areas of communication (24%) and mobility (32%).

**Section 4: Education, Therapy, and Service Use**

Twenty-four total caregivers reported their child’s enrollment in an educational program in the past year with 87.5% (21/24) indicating their child is currently in a program. Multiple program types were reported, including: early intervention, homebound education, mainstream classroom, special education, and other educational programs. Three caregivers reported that their child is not currently enrolled in any school program. Reasons cited for the child not being in a program were that the child is too young for school (n= 2) and the child’s diagnosis limits their ability to be in an educational program (n= 1). For individuals enrolled in an educational program, 85.7% (18/21) have an active individualized educational program (IEP), individualized family service plan (IFSP), or 504 plan. The majority of caregivers, 85.7% (18/21), believe their child’s needs are met and 4.8% (1/21) do not know if their
child’s needs are currently met. Caregivers who feel their child’s educational needs have not been appropriately met cited an unsafe school environment (n= 1) and not enough time provided in the homebound education program (n= 1).

All caregivers reported their child receives therapy services. Most children, 60% (15/24), have four or more therapy visits per month. A breakdown of the therapy types is included in graph 2. Additionally, the majority of children, 79.2% (19/24), receive three or more different therapy types currently. Most caregivers, 70.8% (17/24), believe that their child’s therapy needs are met by their current services while 12.5% (3/24) do not know if their child’s needs are met. Caregivers who feel that their child’s therapy needs are not met cited that the child does not receive enough therapy (n= 2), the caregiver is unable to obtain necessary equipment for their child (n= 1), and difficulty finding appropriate therapists (n= 1).

Section 5: Access to Healthcare and Health Insurance

The majority of the children in the study population received routine healthcare at a physician’s office and 90.9% (20/22) of caregivers report that their child’s primary care needs are currently met. For needs beyond that of routine healthcare, 75% (18/24) of children have been referred to or received care from a specialty healthcare provider (graph 3). Half of these children have been referred to or seen 9 or more specialty healthcare providers. Specialists for which greater than 50% of children had seen or received referral to include: genetics, neurology, cardiology, gastroenterology, ophthalmology, otolaryngology, and surgery. It should be noted that genetics, neurology, and physical medicine and rehabilitation are all included in the multidisciplinary Neurometabolic Clinic and caregiver reported specialty healthcare use may refer to the Neurometabolic Clinic physicians or other physicians within the same specialties. Nearly all caregivers, 94.1% (16/17), believe their child receives all of the specialty healthcare they need.
Difficulties getting healthcare services (in any area) were reported by 45.8% (11/24) of primary caregivers. These difficulties included: child is ineligible for specific services (n= 4), services are not available in the area (n= 2), long waiting lists or difficulties scheduling appointments (n= 4), issues related to costs of services (n= 3), difficulty getting information or resources by the caregiver (n= 3), and other difficulties (n=5). Despite complications getting healthcare services, most children, 95.8% (23/24), do have some type of health insurance coverage. Additionally, 22 caregivers reported total out-of-pocket spending on their child’s healthcare in the past 12 months. Among our study population, 32% of caregivers reported spending $5,000 or more on the child’s healthcare costs annually (graph 4).

Section 6: Care Coordination and Patient and Family Centered Care

Half (12/24) of caregivers indicated that they have received help coordinating their child’s care from a health care provider in the past year with the most likely individual to help with care coordination being the child’s primary care physician. Satisfaction with the communication between all of their child’s care providers was reported by 87.5% (21/24) of caregivers. Likewise, patient and family-centered care was measured by how often caregivers felt their child’s healthcare providers engage with them in six different ways. These scenarios were adapted from the validated National Survey of Children with Special Healthcare Needs and were therefore believed to be good proxies for assessing patient and family-centered care in our study population. Over 90% of caregivers indicated that they usually or always experience these different elements of care (table 6).

Section 7: Family Impact

Half (12/24) of primary caregivers indicated that they usually or always feel overwhelmed by their child’s health care needs. There were a variety of factors which contributed to caregiver stress level (graph 5) with the most frequent factors being financial stress (67%), caregiver’s own emotional
health concerns (58%), difficulty getting respite from caregiving (50%), and other caregiving responsibilities (50%). Additionally, caregiver support services were assessed to determine services caregivers currently use and services caregivers want but do not currently use (graph 6). Notably, the largest deficits in services families want but do not have access to were for support groups (60%), caregiver education/conferences (53%), and counseling services (53%).

Section 8: Neurometabolic Clinic Experience

In order to assess how caregivers feel their child and family’s needs are currently being met by the Neurometabolic Clinic at CCHMC, one open-ended and three close-ended questions were included in the survey. Presently, 87.5% (21/24) of caregivers feel that the clinic has met their child’s needs in the past 12 months. One caregiver indicated that they do not know if their child’s needs are currently met. Due to the optional nature of the open-ended questions, not all caregivers provided answers. Areas of strength were reported by 13 caregivers and areas for improvement reported by 9 caregivers. Answers given were post coded and grouped into response themes (diagram 1). Themes were classified in three different categories: areas of strength, mixed areas of strength/improvement, and areas for improvement. Mixed areas of strength/improvement were rated as either a strength or improvement by different caregivers and thus did not fit into one discrete category.

Caregivers were also asked to identify what other services or specialists they would like to see included in clinic. While only 6 caregivers provided responses, a broad range of specialists and services were indicated fitting into three different categories: medical specialty providers, allied health professionals, and specific informational resources. The medical specialty providers included: pulmonology (n= 3), gastroenterology (n= 2), endocrinology (n= 1), otolaryngology (n= 1), and cardiology (n= 1). Allied health professionals included: social work (n= 1), occupational therapy (n= 1), and
phlebotomy (n= 1). Lastly, specific informational resources were requested regarding: condition specific resources (n= 2), financial assistance (n= 1), and equipment consultation (n= 1).
DISCUSSION:

This study presents data collected using an online survey of primary-caregiver’s of children with suspected and diagnosed neurometabolic disorders to assess the medical needs, service use, and family impact of pediatric neurometabolic disease. In this study only 60% of children were reported to have a known neurometabolic diagnosis. This finding is comparable to another study by Bonkowsky et al (2010) which looked at the burden of pediatric inherited leukodystrophies, finding that 51% of children had not received a definitive diagnosis despite a suspected leukodystrophy [25]. Furthermore, the top three most highly represented diagnoses in our study (mitochondrial disorders, metachromatic leukodystrophy, and Batten disease) aligned closely with the findings of a study by Verity et al (2009) which identified leukoencephalopathies, neuronal ceroid lipofuscinoses, and mitochondrial diseases as the most common identifiable cause of progressive intellectual and neurological deterioration in childhood [21].

Medical Needs

Variability in the medical needs of the children in our study population is likely influenced by the fact that many different diagnoses were included and participants were not asked to specify the subtype of a disorder their child had (for example, to specify if Batten disease was infantile, late-infantile, or juvenile). The ability to make a specific neurometabolic diagnosis can be complicated by the fact that many children often present with non-specific problems like autism, dystonia, developmental delay, epilepsy, growth faltering, and ataxia [20]. Therefore, it is not unexpected that 36% of our study population reportedly does not have a known diagnosis.

This study assessed medical needs in terms of symptoms reported and medical problems of greatest concern to the primary caregiver. Based on the study population, it is not surprising that neurological concerns are experienced by the greatest percentage of children (88%) and are also the
most frequent primary concern reported by caregivers (36%). Musculoskeletal problems were both the second most reported cause of health problems (80%) and the second most frequently cited area of primary concern for caregivers (28%). The high frequency of these concerns suggests that neurological and musculoskeletal problems account for the two greatest sources of morbidity associated with pediatric neurometabolic disorders in our patient population. Respiratory problems were the third most indicated area of primary concern (20%) but interestingly were reported for only a minority of children (24%). This low prevalence of symptoms but high indication for primary concern suggests that respiratory problems affect a smaller percentage of children but account for a relatively significant proportion of morbidity associated with neurometabolic disease in our study.

The term “technology dependent children” has previously been used to describe those children who require “both a medical device to compensate for the loss of a vital body function and significant and sustained care to avert death or further disability” [5]. Functional limitations necessitating technology dependence are highly variable and may be influenced by the specific context in which the child lives. Our study described the current use of equipment and assistive devices in all areas (vision, communication, feeding, mobility, and medical devices/monitors) with the exception of hearing. Despite the frequency of technology use, many functional needs remain unmet by caregiver report. In a study of CSHCN conducted by Dusing et al (2004), unmet needs for therapy services and assistive technology were associated with not having insurance coverage and increased severity of functional limitations [30]. While nearly 96% of the children in our study had insurance coverage, it is possible that such coverage is simply inadequate to meet the child’s functional needs. Additionally, because the highest areas of unmet functional need in our study were for mobility and communication, it is likely that these functional domains are the most challenging to meet for a child who has very severe limitations.
Service Use

Access to educational services for children with disabilities is a federally mandated right for all individuals in the United States from birth until 21 years of age through the Individuals with Disabilities Education Act (IDEA). This law requires that children participate in school programs in the least restrictive environment while ensuring appropriate support services to meet their needs [31]. In our study, 87.5% of children were reported to be involved in some type of an educational program. Although two children were reported to not be receiving any services because they are too young, one child was reported to not be receiving services due to the severity of their disease. For these children who are not receiving services, it is possible there are in fact specific services available that are simply not being utilized. Of parents who did report their child is in an educational program, only 87.5% feel their child’s needs are met by their current program. This suggests that either caregivers may have a different perspective on their child’s educational needs or the full extent of the law has not been fully realized for every child.

Additionally, our study results illustrate that many children receive therapy services that are associated with those areas of most significant functional limitations. Mobility equipment needs were reported for 76% of this population and occupational therapy and physical therapy were each received by 92% of the population. In a similar Australian study looking at the needs of children with mitochondrial diseases and lysosomal storage disorders, a much smaller percentage of children received occupational and physical therapy (50% and 41.7%, respectively) [32]. The difference in utilization of therapies could suggest that there are either more resources for therapy services in the Cincinnati area or there is a higher rate of practitioners who refer children for occupational and physical therapy intervention. Functional needs for communication devices showed a similar correlation with high rates of speech therapy service use in our patient population.
Previous data suggests that CSHCN are more likely to have unmet needs for specialty healthcare services compared to routine healthcare services (from a primary care physician, for example) [33]. Our study data did not identify a similar difference in unmet needs with 90.9% of routine healthcare needs met and 94.1% of specialty healthcare needs met. The slight inconsistency between our data and previous research data may be attributed to the fact that our study has been conducted from a major children’s hospital with abundant access to subspecialty healthcare, therefore likely reducing unmet needs in our study population.

Given the fact that the children reported in this study are seen in a combined genetics and neurology clinic, it is not surprising that genetics and neurology were the top two specialty care providers seen by the study group. The other specialists that are frequently seen by the study population (cardiology, gastroenterology, ophthalmology, otolaryngology, and surgery) help to demonstrate the high diversity of needs experienced by children seen in the Neurometabolic Clinic at CCHMC and support the use of multidisciplinary medical management for children with neurometabolic conditions [23, 32].

**Family Impact of Disease**

The American Academy of Pediatrics released a policy statement in 2012 which specifically defines patient and family-centered care as “an innovative approach to planning, delivery, and evaluation of health care that is grounded in a mutually beneficial partnership among patients, families, and providers that recognizes the importance of family in the patient’s life” [34]. Implementing this type of clinical care in a pediatric healthcare setting has been shown to result in “better health outcomes and wiser allocation of resources as well as greater patient and family satisfaction” [34]. In our study, patient and family-centered care was measured by the self-reported frequency with which primary caregivers have experienced six different scenarios (table 6). Over 90% of caregivers reported that they usually or
always feel that these specific measures are implemented in their child’s care. It is likely that these high levels of patient and family-centered care are correlated with the similarly high percentage of caregivers (87.5%) who feel that their child’s needs are currently met by the Neurometabolic Clinic at CCHMC.

Despite efforts to increase inclusion of families into pediatric medical care, high levels of stress continue to place a significant burden on primary caregivers. Significant medical needs are linked to a substantial time commitment for providing care, frequent provider appointments, the need for care coordination, and financial burden [5]. Prior studies have indicated that mothers are far more likely to be the primary caregiver for a child with disabilities and mothers are also more likely to report caregiver stress compared to fathers [35]. A study by Dellve et al (2006) further delineated the stress experienced by mothers as that related to perceived incompetence, role restriction, social isolation, spousal relationship problems, physical strain, and emotional strain. Fathers, on the other hand, reported only high levels of perceived incompetence related to the care of their child with disabilities [36].

Our study supports the finding that the majority of primary caregivers of children with special needs are mothers. Additionally, 50% of the mothers in our study indicated that they usually or always feel overwhelmed by their child’s healthcare needs. Graph 5 summarizes parent reported areas of caregiver stress resulting from caring for a child with a neurometabolic condition. Notably, our study found that the highest factor associated with stress level was financial stress (67%) which was not specifically assessed in the study by Dellve et al (2006). One possible explanation for this finding could be due to the fact that caring for a child with a neurometabolic condition may be far more financially burdensome than caring for a child with other types of disabilities. Graph 4 helps to further support this possible explanation, showing that 32% caregivers in our study had out-of-pocket healthcare costs for their child of $5,000 or more in the past year despite the majority of children (96%) having some form of health insurance.
Further findings in this study demonstrate that there is either a dearth or low-uptake of support services for primary caregivers of children with neurometabolic conditions. For three different support services, there was a significantly higher number of individuals who reported wanting to use the services but not currently using them. Although our study did not delineate why these services were not specifically being used, it does suggest that the availability of counseling services, support groups, and caregiver education or conferences may help to bridge the gap between used services and desired services. The previously described high levels of stress further support the need for implementation of caregiver support and education programs.

Previous studies indicating high levels of social isolation for families of children with complex needs strengthens the argument for identifying appropriate support groups for families of children with neurometabolic conditions [36, 37]. While access to in-person support groups may be limited by geographic location and prevalence of each unique diagnosis, several studies have suggested alternatives. Online resources may be a good alternative to in-person support groups while more generalized support groups (e.g. for caregivers of children with any developmental disabilities) may be more appropriate for families who would like to meet other caregivers in person but do not live near another caregiver of a child with the same diagnosis [16, 32]. Likewise, many of the children in our study currently have unknown diagnoses, further complicating a family’s ability to reach out to others who have children with the same genetic syndrome.

Lastly, our study also assessed each caregiver’s experience with their child’s care in the multidisciplinary Neurometabolic Clinic at CCHMC. Most caregivers (87.5%) reported they believe their child’s needs are met by the clinic, supporting the previously mentioned suggestion for multidisciplinary care of children with complex conditions like neurometabolic disorders. While relatively few caregivers gave suggestions for other specialties they would like to see incorporated into the clinic, both pulmonology and gastroenterology were suggested by more than one caregiver. Similarly small numbers
of caregivers reported specific informational resources they would like to see made available in clinic
with only condition specific resources being requested by more than one individual.

Areas of strength and areas for improvement provided interesting information about those
experiences unique to each domain and those overlapping between domains (diagram 1). Unique
strengths of the clinic included: supportive and caring providers, resources and advocacy, and patient
and family-centered care. These strengths were in contrast to those areas for improvement, including:
scheduling of appointments, lack of specific expertise, and lengthy appointment times. Three other
overlapping response themes between the domains illustrate that there is currently inconsistency in the
experiences of different caregivers related to: communication with the family, multidisciplinary care,
and care coordination. The areas for improvement and areas in which there is inconsistency provide
specific evidence for ways in which the Neurometabolic Clinic can better meet patient and family needs.

Of note, many changes have already been implemented in the neurometabolic clinic to better
meet patient and family needs prior to and since completing the present study. The physical medicine
and rehabilitation physician who has previously been a member of the multidisciplinary team has
expanded the availability of services to better meet the demand. Additionally, a speech language
pathologist will soon be involved in the clinic. Lastly, a program to provide iPads to children managed in
the clinic should assist in meeting the communication needs of this population. These and other changes
may help to increase the comprehensiveness of care that is delivered to this population.

Limitations

There are several important limitations of this study. All research participants were recruited
from a single children’s hospital and receive management of their known or suspected neurometabolic
conditions by the same group of providers. Many children with lysosomal storage disorders and
mitochondrial disorders (many of which fall into the spectrum of neurometabolic conditions) are seen at
this facility but not managed in this clinic and therefore their caregivers were not invited to participate in the study. It is possible that caregivers of children with more severe needs were more likely to participate in the study because they were more likely to want to see changes made in the clinic to better meet their child’s needs. Conversely, it is also possible that caregivers of children with more severe needs are far busier with caregiving responsibilities and were less likely to have the time to participate in this study. The fact that this study was voluntary makes it difficult to know if the 25 participating caregivers were a representative sample. The relatively homogenous demographic characteristics of the participants in the study also may make the data less generalizable across different genders of care providers, racial groups, or caregiver types.

We attempted to limit recall bias by specifying that responses to the study should relate only to the past 12 months; however it is still possible that caregivers could have inaccurately recalled information. Prior research on the accuracy of parent recall related to a child’s medical care has shown that parents (particularly mothers) are typically able to acceptably recall health related events for research purposes [38]. This supports our belief that recall bias likely played only a minimal role in our study.

The overall participation rate of 19% was relatively low although the study team attempted to increase the response rate by having multiple modes of inviting study participation. Despite these limitations, it is hoped that this study will help to highlight those areas for which further study can elucidate novel care strategies and interventions for children with neurometabolic conditions and their families.
CONCLUSIONS:

There is currently a paucity of literature specifically investigating pediatric neurometabolic diseases and the associated medical needs, service use, and impact of disease on the family. Our results clearly describe children with neurometabolic conditions as a population with very complex care needs due to variable health conditions and functional limitations. Service needs are numerous with many children requiring ongoing specialty medical management, therapeutic intervention, and educational supports. Primary caregivers of children with neurometabolic conditions face a significant burden of care provision along with markedly high levels of parental stress. Patient and family-centered care strategies may help to meet the needs of these families. Caregiver support services and multidisciplinary clinics may also be of benefit but further research into how to comprehensively address these areas is necessary. Lastly, this research also creates evidence for specific changes that can be made within the Neurometabolic Clinic at CCHMC to better serve this unique population, including expansion of multidisciplinary services, continued improvements in communication (with families and providers), and streamlining of scheduling and appointment times.
REFERENCES:


APPENDIX A: Figures

Boxes:

<table>
<thead>
<tr>
<th>Box 1. Possible Neurologic Features of Neurometabolic Disease</th>
<th>Box 2. Possible Non-neurologic Features of Neurometabolic Disease</th>
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<tbody>
<tr>
<td>Encephalopathy</td>
<td>Abnormal respiration</td>
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<td>Hepatosplenomegaly</td>
</tr>
<tr>
<td>Microcephaly/macrocephaly</td>
<td>Liver dysfunction/failure</td>
</tr>
<tr>
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<td>Renal tubular dysfunction</td>
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<td>Cardiac problems</td>
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<td>Hypotonia</td>
<td>Feeding/growth problems</td>
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<td>Skin/hair abnormalities</td>
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<td>Exercise intolerance</td>
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<td>Polyneuropathy</td>
<td>Vision changes</td>
</tr>
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<td>Ataxia</td>
<td>Bone crisis</td>
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<td>Psychiatric manifestations</td>
<td>Stroke</td>
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<td>Pancreatitis</td>
</tr>
<tr>
<td></td>
<td>Recurrent emesis/diarrhea</td>
</tr>
<tr>
<td></td>
<td>Hearing loss</td>
</tr>
<tr>
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<td>Dysmorphic/coarsened features</td>
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## Table 1. Caregiver Demographics (n= 25)

<table>
<thead>
<tr>
<th>Category</th>
<th>Total (%)</th>
</tr>
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<tbody>
<tr>
<td><strong>Mean (SD)</strong></td>
<td></td>
</tr>
<tr>
<td>Age (years)</td>
<td>37.28 (5.04)</td>
</tr>
<tr>
<td><strong>Sex</strong></td>
<td></td>
</tr>
<tr>
<td>Female</td>
<td>25(100%)</td>
</tr>
<tr>
<td><strong>Relationship to child</strong></td>
<td></td>
</tr>
<tr>
<td>Biological parent</td>
<td>24(96%)</td>
</tr>
<tr>
<td>Adoptive parent</td>
<td>1(4%)</td>
</tr>
<tr>
<td><strong>Education</strong></td>
<td></td>
</tr>
<tr>
<td>High school graduate/GED</td>
<td>3(12%)</td>
</tr>
<tr>
<td>Some college/technical school</td>
<td>10(40%)</td>
</tr>
<tr>
<td>College degree</td>
<td>8(32%)</td>
</tr>
<tr>
<td>Some graduate school</td>
<td>2(8%)</td>
</tr>
<tr>
<td>Graduate school</td>
<td>2(8%)</td>
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<tr>
<td><strong>Employment</strong></td>
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<tr>
<td>Employed outside of home</td>
<td>10(40%)</td>
</tr>
<tr>
<td>Not employed outside of home</td>
<td>15(60%)</td>
</tr>
<tr>
<td><strong>Annual household income</strong></td>
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</tr>
<tr>
<td>Less than $30k</td>
<td>9(36%)</td>
</tr>
<tr>
<td>$30k - less than $60k</td>
<td>3(12%)</td>
</tr>
<tr>
<td>$60k - less than $90k</td>
<td>5(20%)</td>
</tr>
<tr>
<td>$90k or more</td>
<td>7(28%)</td>
</tr>
<tr>
<td>Declined</td>
<td>1(4%)</td>
</tr>
<tr>
<td><strong>State of residence (n= 24)</strong></td>
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<tr>
<td>Kentucky</td>
<td>12(50%)</td>
</tr>
<tr>
<td>Ohio</td>
<td>10(41.67%)</td>
</tr>
<tr>
<td>Indiana</td>
<td>1(4.17%)</td>
</tr>
<tr>
<td>Tennessee</td>
<td>1(4.17%)</td>
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</table>

## Table 2. Child Demographics (n= 25)

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<th>Category</th>
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</tr>
</thead>
<tbody>
<tr>
<td><strong>Mean (SD)</strong></td>
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</tr>
<tr>
<td>Age (years)</td>
<td>6.56 (3.56)</td>
</tr>
<tr>
<td><strong>Sex</strong></td>
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<tr>
<td>Female</td>
<td>13(52%)</td>
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<tr>
<td>Male</td>
<td>12(48%)</td>
</tr>
<tr>
<td><strong>Race</strong></td>
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<tr>
<td>White</td>
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<tr>
<td><strong>Ethnicity</strong></td>
<td></td>
</tr>
<tr>
<td>Non-Hispanic</td>
<td>25(100%)</td>
</tr>
<tr>
<td><strong>Has diagnosis</strong></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>15(60%)</td>
</tr>
<tr>
<td>No</td>
<td>9(36%)</td>
</tr>
<tr>
<td>Declined</td>
<td>1(4%)</td>
</tr>
<tr>
<td><strong>Known diagnosis (n= 15)</strong></td>
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<tr>
<td>Mitochondrial disorder*</td>
<td>4(26.67%)</td>
</tr>
<tr>
<td>Metachromatic leukodystrophy</td>
<td>3(20%)</td>
</tr>
<tr>
<td>Batten disease</td>
<td>3(20%)</td>
</tr>
<tr>
<td>Krabbe disease</td>
<td>1(6.67%)</td>
</tr>
<tr>
<td>KIF1A mutation</td>
<td>1(6.67%)</td>
</tr>
<tr>
<td>Hyperekplexia</td>
<td>1(6.67%)</td>
</tr>
<tr>
<td>Microcephaly</td>
<td>1(6.67%)</td>
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<tr>
<td>Macrocephaly</td>
<td>1(6.67%)</td>
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*one child with a mitochondrial disorder
also has Cowden syndrome
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<tr>
<th>DIAGNOSIS</th>
<th>Neurological problems</th>
<th>Musculoskeletal problems</th>
<th>Gastrointestinal problems</th>
<th>Otolaryngological problems</th>
<th>Ophthalmologic problems</th>
<th>Immunologic problems</th>
<th>Cardiovascular/hematologic problems</th>
<th>Endocrine/weight gain problems</th>
<th>Psychological/psychiatric problems</th>
<th>Respiratory problems</th>
<th>Urogenital problems</th>
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<tr>
<td>Response Percentage</td>
<td>88%</td>
<td>80%</td>
<td>64%</td>
<td>64%</td>
<td>58%</td>
<td>32%</td>
<td>28%</td>
<td>28%</td>
<td>24%</td>
<td>24%</td>
<td>20%</td>
<td>20%</td>
</tr>
</tbody>
</table>

● = Has problems
○ = Do not know if there are problems
Table 4. Most Frequently Reported Symptoms by Affected Body System

<table>
<thead>
<tr>
<th>Top 5 affected body systems</th>
<th>Highest reported symptoms</th>
<th>% of children with reported symptom</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neurological (&lt;n= 22/25&gt;</td>
<td>Developmental delay</td>
<td>95.5%</td>
</tr>
<tr>
<td></td>
<td>Hypotonia</td>
<td>77.3%</td>
</tr>
<tr>
<td></td>
<td>Movement disorder</td>
<td>72.7%</td>
</tr>
<tr>
<td></td>
<td>Poor coordination</td>
<td>68.2%</td>
</tr>
<tr>
<td></td>
<td>Intellectual disability</td>
<td>54.5%</td>
</tr>
<tr>
<td></td>
<td>Seizures</td>
<td>52.0%</td>
</tr>
<tr>
<td></td>
<td>Learning disability</td>
<td>45.5%</td>
</tr>
<tr>
<td></td>
<td>Brain structural abnormality</td>
<td>45.5%</td>
</tr>
<tr>
<td></td>
<td>Spasticity</td>
<td>45.5%</td>
</tr>
<tr>
<td></td>
<td>Sleep problems</td>
<td>45.5%</td>
</tr>
<tr>
<td></td>
<td>Temperature Instability</td>
<td>45.5%</td>
</tr>
<tr>
<td>Musculoskeletal (&lt;n= 20/25&gt;</td>
<td>Muscle weakness</td>
<td>90.0%</td>
</tr>
<tr>
<td></td>
<td>Scoliosis</td>
<td>45.0%</td>
</tr>
<tr>
<td></td>
<td>Muscle pain</td>
<td>35.0%</td>
</tr>
<tr>
<td>Gastrointestinal (&lt;n= 16/25&gt;</td>
<td>Frequent constipation</td>
<td>75.0%</td>
</tr>
<tr>
<td></td>
<td>Reflux</td>
<td>56.3%</td>
</tr>
<tr>
<td>Otolaryngological (&lt;n= 16/25&gt;</td>
<td>Frequent ear infections</td>
<td>56.3%</td>
</tr>
<tr>
<td></td>
<td>Snoring at night</td>
<td>56.3%</td>
</tr>
<tr>
<td></td>
<td>Speech problems</td>
<td>56.3%</td>
</tr>
<tr>
<td></td>
<td>Mouth breathing</td>
<td>50.0%</td>
</tr>
<tr>
<td>Ophthalmologic (&lt;n= 14/24&gt;</td>
<td>Nearsighted</td>
<td>42.9%</td>
</tr>
<tr>
<td></td>
<td>Farsighted</td>
<td>35.7%</td>
</tr>
<tr>
<td></td>
<td>Blind</td>
<td>35.7%</td>
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</tbody>
</table>
Table 5. Use of Equipment, Devices, and Monitors to Meet Functional Needs

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<tr>
<th></th>
<th>VISION</th>
<th>HEARING</th>
<th>COMMUNICATION</th>
<th>FEEDING</th>
<th>MOBILITY</th>
<th>MEDICAL DEVICES/ MONITORS</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Current Use</strong></td>
<td>48%</td>
<td>0%</td>
<td>56%</td>
<td>36%</td>
<td>76%</td>
<td>40.9%</td>
</tr>
<tr>
<td><strong>Unmet need</strong></td>
<td>12%</td>
<td>4%</td>
<td>24%</td>
<td>12%</td>
<td>32%</td>
<td>12%</td>
</tr>
</tbody>
</table>
Table 6. Experiences with Patient and Family-Centered Care (PFCC): Caregivers who usually or always experience 6 elements of PFCC

<table>
<thead>
<tr>
<th>&quot;Healthcare provider...&quot;</th>
<th>Percentage of Caregivers</th>
</tr>
</thead>
<tbody>
<tr>
<td>Listens carefully to the caregiver</td>
<td>95.8%</td>
</tr>
<tr>
<td>Encourages the caregiver to raise concerns</td>
<td>95.8%</td>
</tr>
<tr>
<td>Encourages the caregiver to ask questions</td>
<td>95.8%</td>
</tr>
<tr>
<td>Provides the caregiver with the specific information they need</td>
<td>91.7%</td>
</tr>
<tr>
<td>Considers the caregivers opinion about options for the child’s care</td>
<td>91.7%</td>
</tr>
<tr>
<td>Spends enough time with their child</td>
<td>91.7%</td>
</tr>
</tbody>
</table>
Graphs:

Graph 1. Medical Problems by Body System

- Neurological
- Musculoskeletal
- Gastrointestinal
- Otolaryngological
- Ophthalmologic
- Immunologic
- Cardiovascular/hematologic
- Endocrine/weight gain
- Psychological/psychiatric
- Respiratory
- Urogenital
- Dermatologic

Percentage of Children with Symptoms by Caregiver Report
Graph 2. Therapy Services Received
Graph 3. Use of Specialty Healthcare Services

- Genetics
- Neurology
- Cardiology
- Gastroenterology
- Ophthalmology
- Otolaryngology/ENT
- Surgery
- Nutrition
- Audiology
- Emergency Medicine
- Feeding Team
- Developmental and Behavioral Pediatrics
- Endocrinology
- Urology
- Nephrology
- Physical medicine
- Psychology/Psychiatry
- Immunology
- Pulmonology
- Dermatology
- Hematology

Percentage of Children Receiving Specialty Care
Graph 4. Out-of-pocket Spending on Child's Healthcare in the past 12 Months

- Less than $1,000: 23%
- $1,000 to less than $2,000: 18%
- $2,000 to less than $3,000: 18%
- $3,000 to less than $4,000: 5%
- $4,000 to less than $5,000: 5%
- $5,000 or more: 32%
Graph 5. Factors Contributing to Caregiver Stress Level

- Financial stress
- Personal emotional health concerns
- Difficulty getting breaks from caregiving
- Other caregiving responsibilities
- Issues with health insurance
- Employment concerns
- Difficult relationships with other care providers
- Personal physical health concerns
- A lack of training/skills needed to provide care
- Child's diagnosis is unknown

Percentage of Caregivers Reporting Stress Factor
Graph 7. Caregiver Support Service Utilization

Support service used  Support service NOT used but wanted

Percentage of Caregivers

- Respite care services
- Counseling
- Advocacy organization/association
- Support groups
- Religious groups
- Caregiver education/conferences
- One-on-one training
- Other services
Diagrams:

Diagram 1. Neurometabolic Clinic Experience

Caregiver reported areas of strength (green), mixed areas of strength and improvement (yellow), and areas for improvement (red) in the Neurometabolic Clinic at CCHMC

#### Areas of Strength
- Supportive and Caring Providers (n= 7)
- Resources and Advocacy (n= 4)
- Patient and Family Centered Care (n= 3)

#### Areas of Strength / Improvement
- Communication with the Family (n= 5) / (n= 2)
- Multidisciplinary Care (n= 4) / (n= 2)
- Care Coordination (n= 2) / (n= 2)

#### Areas for Improvement
- Scheduling Appointments (n= 3)
- Lack of Specific Expertise (n= 3)
- Lengthy Appointments (n= 1)
APPENDIX B: Online Survey

Comprehensive Assessment of Children Managed in a Neurometabolic Clinic:
Medical Needs, Service Use, and Impact of Disease on the Family

Study Title: Pediatric Neurometabolic Disorders: Medical Needs, Service Use, and Impact of Disease on the Family

Study Information and Purpose: The Neurometabolic Clinic at Cincinnati Children’s Hospital Medical Center (CCHMC) is doing a research study about the special health care concerns of children managed in the clinic. Primary caregivers of children are being asked to complete this survey to describe their child’s medical needs, service use, and the impact of his or her condition on the family in the past 12 months. In order to take part in this survey, you must be the primary caregiver of a child between 0-17 years of age who receives care at the Neurometabolic Clinic at CCHMC. Please fill out only one survey per child. Caregivers with more than one child between 0-17 years old who is seen in the Neurometabolic Clinic may fill out one for each child. The survey should take about 20 minutes to complete. There are no right or wrong answers to these questions and your responses will be kept confidential.

How the Data will be Used: There will be no direct benefits for caregivers who respond to this research study; however, the results of this survey will be used to find ways to improve patient care in the Neurometabolic Clinic. Please note, no changes to the clinic are promised as a result of taking this survey.

Study Participation: This study is voluntary and you may choose not to answer any question or stop taking the survey at any time. Choosing not to take part in this research study will not affect the care your child receives in the Neurometabolic Clinic. Clinic staff will not have access to your individual survey responses. This is a minimal risk study and no identifiable information (such as your name or phone number) will be asked for in the survey. Your child’s name and address will only be used to send out study invitations. The loss of your confidentiality is a small risk with this study; however, the study investigator will take measures to protect your contact information. Your information will be kept in a password protected database, separate from your survey answers. The data from this study may be published; however, results will not contain any identifiable information. By completing this survey, you are giving your consent to take part in the study.

Contact Information: If you have any questions or concerns about this study, please contact the study investigator, Jullianne Diaz, at Jullianne.Diaz@cchmc.org or the Neurometabolic Clinic genetic counselor, Christine Spaeth, at Christine.Spaeth@cchmc.org or 513-636-9861.

Thank you for considering taking part in this research.

NOTE: SURVEY RESPONSES SHOULD BE GIVEN FOR MEDICAL NEEDS, SERVICE USE, AND IMPACT ON THE FAMILY IN THE PAST 12 MONTHS ONLY (A note at the top of each page will serve as a reminder)
Survey Eligibility
Are you the primary caregiver of a child seen in the Neurometabolic Clinic at Cincinnati Children's Hospital Medical Center? (Primary caregivers are those individuals who provide the majority of the care to a child.)

☐ Yes
☐ No

Are you currently 18 years of age or older? (Only primary caregivers who are 18 years of age or older are eligible to complete this survey.)

☐ Yes
☐ No

Is your child currently 17 years of age or younger? (Only primary caregivers of children ages 17 years and younger are eligible to complete this survey.)

☐ Yes
☐ No
SECTION1: Demographics-- Primary Caregiver

What is your age in years?

☐ DROP DOWN LIST (AGES: 18 – 90 OR OLDER; DECLINE TO ANSWER)

What is your sex?

☐ Male
☐ Female
☐ Decline to answer

What is your relationship to the child seen in the Neurometabolic Clinic?

☐ Biological parent
☐ Step-parent
☐ Adoptive parent
☐ Foster parent
☐ Other adult relative
☐ Other adult non-relative
☐ Decline to answer

What is the highest amount of schooling you have completed?

☐ Less than high school
☐ Some high school
☐ High school graduate or GED
☐ Some college or technical school
☐ College degree
☐ Some graduate school
☐ Graduate degree
☐ Decline to answer

Are you currently employed outside of your home, either part-time or full-time?

☐ Yes
☐ No
☐ Decline to answer

What is your annual household income?

☐ Less than $10,000
☐ $10,000 to less than $20,000
☐ $20,000 to less than $30,000
☐ $30,000 to less than $40,000
☐ $40,000 to less than $50,000
☐ $50,000 to less than $60,000
☐ $60,000 to less than $70,000
☐ $70,000 to less than $80,000
☐ $80,000 to less than $90,000
☐ $90,000 to less than $100,000
☐ $100,000 or more
☐ Don’t know
☐ Decline to answer

In which state do you currently live?

☐ DROP DOWN LIST (ALL 50 STATES; DECLINE TO ANSWER; DO NOT LIVE IN THE US)
SECTION1: Demographics-- Child

What is the age (in years) of your child who is seen in the Neurometabolic Clinic?

□ DROP DOWN LIST (AGES: LESS THAN 1 – 17; DECLINE TO ANSWER)

What is your child's sex?

□ Male
□ Female
□ Decline to answer

What is your child's race? (SELECT ALL THAT APPLY)

□ American Indian or Alaska Native
□ Asian
□ Black or African American
□ Native Hawaiian or Pacific Islander
□ White
□ Other race
□ Decline to answer

What is your child's ethnicity?

□ Non-Hispanic
□ Hispanic
□ Decline to answer

Have doctors told you what your child’s suspected or diagnosed condition is?

□ Yes
□ No
□ Decline to answer

What is the condition doctors told you? (This question is voluntary; you may decline to answer by skipping the question.)
SECTION 2: Health Conditions and Concerns

NOTE: The following questions refer to the past 12 months ONLY

Has your child had any eye problems?
- Yes
- No
- Don't know
- Decline to answer

Please indicate your child's eye problems: (SELECT ALL THAT APPLY)
- Nearsighted (problems seeing far away)
- Farsighted (problems seeing up close)
- Strabismus (eyes point in different directions)
- Nystagmus (uncontrolled eye movements)
- Blind
- Light sensitivity
- Lens dislocation
- Cataracts
- Retina problems
- Other eye problems [Please specify your child's other eye problems: ___________ ]

Has your child had any ear, nose, mouth, or throat problems?
- Yes
- No
- Don't know
- Decline to answer

Please indicate your child's ear, nose, mouth, or throat problems: (SELECT ALL THAT APPLY)
- Frequent ear infections
- Frequent sinus infections
- Hearing loss
- Snoring at night
- Mouth breathing
- Speech problems
- Dental problems
- Other ear, nose, mouth, or throat problems [Please specify your child's other ear, nose, mouth, or throat problems: ___________ ]

Has your child had any cardiovascular, blood, or circulation problems?
- Yes
- No
- Don't know
- Decline to answer

Please indicate your child's cardiovascular, blood, or circulation problems: (SELECT ALL THAT APPLY)
- Heart murmur
- Congenital heart defect (born with a heart defect)
- Cardiomyopathy (disease of the heart muscle)
- Chest pain
- Anemia
- High blood pressure
- Cyanosis (such as bluish hands or feet)
Easy bruising
Lymphedema (such as swelling of the arms and legs)
Other cardiovascular, blood, or circulation problems [Please specify your child's other cardiovascular, blood, or circulation problems: ___________ ]

Has your child had any breathing problems?
- Yes
- No
- Don't know
- Decline to answer

Please indicate your child's breathing problems: (SELECT ALL THAT APPLY)
- Trouble breathing/shortness of breath
- Choking episodes
- Frequent cough
- Wheezing
- Asthma
- Apnea (pauses in breathing)
- Other breathing problems [Please specify your child's other breathing problems: ___________ ]

Has your child had any genital or urinary problems?
- Yes
- No
- Don't know
- Decline to answer

Please indicate your child's genital or urinary problems: (SELECT ALL THAT APPLY)
- Frequent urinary tract infections
- Other genital or urinary problems [Please specify your child's other genital or urinary problems: ___________ ]

Has your child had any stomach or intestinal problems?
- Yes
- No
- Don't know
- Decline to answer

Please indicate your child's stomach or intestinal problems: (SELECT ALL THAT APPLY)
- Frequent constipation
- Frequent diarrhea
- Stomach/abdominal pain
- Reflux
- Frequent vomiting
- Decreased appetite
- Increased appetite
- Other stomach or intestinal problems [Please specify your child's other stomach or intestinal problems: ___________ ]

Has your child had any muscle or skeletal problems?
- Yes
- No
- Don't know
☐ Decline to answer

Please indicate your child's muscle or skeletal problems: (SELECT ALL THAT APPLY)
☐ Muscle pain
☐ Muscle weakness
☐ Joint pain
☐ Arthritis
☐ Bone fractures
☐ Scoliosis (curve in the spine)
☐ Osteopenia/osteoporosis (loss of bone density)
☐ Other muscle or skeletal problems [Please specify your child's other muscle or skeletal problems: ___________ ]

Has your child had any skin problems?
☐ Yes
☐ No
☐ Don't know
☐ Decline to answer

Please indicate your child's skin problems: (SELECT ALL THAT APPLY)
☐ Eczema
☐ Skin growths
☐ Sores that do not heal
☐ Other skin problems [Please specify your child's other skin problems: ___________ ]

Has your child had any endocrine (hormonal) or weight gain problems?
☐ Yes
☐ No
☐ Don't know
☐ Decline to answer

Please indicate your child's endocrine (hormonal) or weight gain problems: (SELECT ALL THAT APPLY)
☐ Diabetes
☐ Thyroid problems
☐ Poor weight gain
☐ Excess weight gain
☐ Other endocrine or hormonal problems [Please specify your child's other endocrine (hormonal) or weight gain problems: ___________ ]

Has your child had any immune system problems?
☐ Yes
☐ No
☐ Don't know
☐ Decline to answer

Please indicate your child's immune system problems: (SELECT ALL THAT APPLY)
☐ Immune system deficiency
☐ Frequent infections
☐ Allergic reactions
☐ Other immune system problems [Please specify your child's other immune system problems: ___________ ]

Has your child had any neurological problems?
☐ Yes
☐ No
☐ Don’t know
☐ Decline to answer

Please indicate your child's neurological problems: (SELECT ALL THAT APPLY)
☐ Developmental delay
☐ Intellectual disability
☐ Learning disability
☐ Autism
☐ Seizures
☐ Syncope (fainting spells)
☐ Dizziness
☐ Frequent headaches/migraines
☐ Brain structure abnormalities
☐ Stroke
☐ Memory problems
☐ Hypotonia (low muscle tone)
☐ Movement disorder (such as tremors/ataxia or dystonia)
☐ Spasticity (such as muscle tightness or exaggerated reflexes)
☐ Poor coordination
☐ Sleep problems
☐ Temperature instability
☐ Other neurological problems Please specify your child's other neurological problems: ___________

Has your child had any psychological or psychiatric problems?
☐ Yes
☐ No
☐ Don’t know
☐ Decline to answer

Please indicate your child's psychological or psychiatric problems: (SELECT ALL THAT APPLY)
☐ Anxiety
☐ Depression
☐ Aggression
☐ Hyperactivity
☐ Impulsivity
☐ Short attention span
☐ ADD or ADHD
☐ Other psychological or psychiatric problems [Please specify your child's other psychological or psychiatric problems: ___________

What have been your primary concerns related to your child's health in the past 12 months? (This question is voluntary; you may decline to answer by skipping the question.)
SECTION 3: Functional Needs

NOTE: The following questions refer to the past 12 months ONLY

Has your child used any of the following devices to help with vision needs? (SELECT ALL THAT APPLY)
- Glasses/contacts
- Assistive devices (i.e. books on tape, magnifiers, projection devices, reading rectangle, etc.)
- Other vision devices [Please specify what other vision devices you child used: ___________ ]
- Has not used vision devices

Has your child used any of the following devices to help with hearing needs? (SELECT ALL THAT APPLY)
- Hearing aid(s)
- Cochlear implant(s)
- Assistive listening devices (i.e. FM sound system, alerting devices, etc.)
- Other hearing devices [Please specify what other hearing devices your child used: ___________ ]
- Has not used hearing devices

Has your child used any of the following devices to help with communication needs? (SELECT ALL THAT APPLY)
- Switches/buttons
- Electronic communication device
- Picture exchange communication system (PECS)
- Other communication devices [Please specify what other communication devices your child used: ___________ ]
- Has not used communication devices

Has your child used any of the following devices to help with feeding needs? (SELECT ALL THAT APPLY)
- NG-tube
- G-tube
- J-tube
- Central line/IV
- Other feeding devices [Please specify what other feeding devices your child used: ___________ ]
- Has not used feeding devices

Has your child used any of the following equipment to help with mobility (moving around) needs? (SELECT ALL THAT APPLY)
- Walker
- Wheelchair
- Braces
- Canes/crutches
- Gait trainer
- Stander
- Lift
- Other mobility equipment [Please specify what other mobility equipment your child used: ___________ ]
- Has not used mobility equipment

Has your child used any of the following medical devices or monitors? (SELECT ALL THAT APPLY)
- Apnea monitor
- Ventilator
- CPAP
- BiPAP
- Tracheostomy
- Oxygen tank
- Suction pump
- Oximeter
- Insulin pump
- Blood glucose monitor
- Blood ketone monitor
- Other medical devices or monitors [Please specify what other medical devices or monitors your child used: ____________]
- Has not used medical devices or monitors

Do you believe your child's functional needs are met in the following areas:

<table>
<thead>
<tr>
<th>Area</th>
<th>YES</th>
<th>NO</th>
<th>DON'T KNOW</th>
</tr>
</thead>
<tbody>
<tr>
<td>VISION</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>HEARING</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>COMMUNICATION</td>
<td>☐</td>
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<tr>
<td>FEEDING</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>MOBILITY</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>MEDICAL DEVICES/MONITORS</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
</tbody>
</table>
SECTION 4: Education, Special Therapy, and Service Use

NOTE: The following questions refer to the past 12 months ONLY

What kind of educational program was your child in? (SELECT ALL THAT APPLY)
- ☐ Early intervention
- ☐ Homebound (instruction provided by the school system in the home)
- ☐ Homeschool (instruction provided by a caregiver in the home)
- ☐ Mainstream/regular classroom
- ☐ Special education
- ☐ Other educational program [Please specify what other educational program your child was in: ___________ ]
- ☐ Not in an educational program [Please indicate why your child was not in an educational program: ___________] 

Does your child have an Individualized Family Service Plan (IFSP), Individualized Education Plan (IEP), or 504 plan?
- ☐ Yes
- ☐ No
- ☐ Don't know

Do you believe your child's educational needs are being met by his or her educational program?
- ☐ Yes
- ☐ No [Please indicate why your child's needs are not being met by his or her educational program: ___________ ]
- ☐ Don't know

What type of therapy has your child received? (SELECT ALL THAT APPLY)
- ☐ Occupational therapy
- ☐ Physical therapy
- ☐ Speech therapy
- ☐ Vision therapy
- ☐ Other therapy [Please specify what other therapy your child received: ___________ ]
- ☐ Has not received therapy

Do you believe your child's needs are being met by his or her therapy services?
- ☐ Yes
- ☐ No [Please indicate why your child's needs are not being met by his or her therapy services: ___________ ]
- ☐ Don't know

On average, how many total therapy visits does your child have per month?
- ☐ 1 or fewer visits
- ☐ 2-3 visits
- ☐ 4-5 visits
- ☐ 6-7 visits
- ☐ 8-9 visits
- ☐ 10 or greater visits
SECTION 5: Access to Healthcare and Health Insurance Coverage
NOTE: The following questions refer to the past 12 months ONLY

Where does your child usually go when he or she needs routine health care, such as well-child check-ups?

- Physician's office
- Urgent care center
- Hospital emergency room
- Other place [Please specify what other place your child usually goes to for routine health care: ___________]
- Does not receive routine health care

On average, how many total routine health care visits does your child have per month?

- 1 or fewer visits
- 2-3 visits
- 4-5 visits
- 6 or greater visits

Does your child receive all of the routine health care that he or she needs?
- Yes
- No
- Don't know

Does your child receive all of the routine dental care that he or she needs?
- Yes
- No
- Don't know

Has your child seen or received a referral to any specialty health care provider?
- Yes
- No
- Don't know

Please SELECT ALL of the specialty health care providers your child has seen or received a referral to:

- Audiology
- Cardiology
- Developmental-Behavioral Pediatrics
- Emergency Medicine
- Endocrinology
- Feeding Team
- Gastroenterology
- Genetics
- Hematology
- Immunology
- Nephrology
- Neurology
- Nutrition
- Oncology
- Ophthalmology
- Otolaryngology/ENT
- Physical medicine
- Psychology/Psychiatry
- Pulmonology
- Rheumatology
Surgery
Urology
Other specialty health care provider [Please specify what other specialty health care provider your child has seen or been referred to: ___________ ]

On average, how many total specialty health care visits does your child have per month?
- 1 or fewer visits
- 2-3 visits
- 4-5 visits
- 6 or greater visits

Does your child receive all of the specialty health care that he or she needs?
- Yes
- No
- Don’t know

Does your child use home nursing services (i.e. care provided by a nurse in your home)?
- Yes
- No
- Don’t know

Does your child receive all of the home nursing services that he or she needs?
- Yes
- No
- Don’t know

Does your child take prescription medications?
- Yes
- No
- Don’t know

Please indicate the reasons you experience difficulties getting health care services for your child: (SELECT ALL THAT APPLY)
- Child is not eligible for specific services
- Services are not available in your area
- Long waiting lists or other problems getting appointments
- Issues related to the cost of services
- Difficulty getting information you need
- Other difficulties [Please specify what other difficulties you experience getting health care services for your child: ___________ ]
- No difficulties getting health care services

Please indicate the types of health insurance your child is covered by: (SELECT ALL THAT APPLY)
- Medicaid
- Medicare
- State Children's Health Insurance Program (SCHIP)
- Medigap
- Military
- Indian Health Services
- Private insurance
- Single service plan (dental, vision, prescriptions, etc.)
- Other health insurance [Please specify what other health insurance your child is covered by: ___________ ]
☐ Does not have health insurance

Does your child's health insurance allow him or her to receive the services he or she needs?
☐ Always
☐ Usually
☐ Sometimes
☐ Never

Does your child's health insurance allow him or her to receive the testing he or she needs?
☐ Always
☐ Usually
☐ Sometimes
☐ Never

What would you estimate the out-of-pocket costs of your child's healthcare were in the past 12 months?
☐ Less than $1,000
☐ $1,000 to less than $2,000
☐ $2,000 to less than $3,000
☐ $3,000 to less than $4,000
☐ $4,000 to less than $5,000
☐ $5,000 or more
☐ Decline to answer
SECTION 6: Care Coordination and Patient and Family Centered Care

NOTE: The following questions refer to the past 12 months ONLY

Do any of your child's health care providers help coordinate your child's care?

☐ Yes
☐ No
☐ Don't know

Please indicate those health care providers who help coordinate your child's care: (SELECT ALL THAT APPLY)

☐ Physician (primary care)
☐ Physician (specialty care)
☐ Genetic counselor
☐ Nurse
☐ Therapist
☐ Social worker
☐ Other health care provider [Please specify what other health care provider helps coordinate your child's care: ___________]

Overall, how satisfied are you with the communication between your child's health care providers?

☐ Very satisfied
☐ Somewhat satisfied
☐ Somewhat dissatisfied
☐ Very dissatisfied
☐ No communication needed

How often do your child’s doctors do the following:

<table>
<thead>
<tr>
<th></th>
<th>ALWAYS</th>
<th>USUALLY</th>
<th>SOMETIMES</th>
<th>NEVER</th>
<th>NOT APPLICABLE</th>
</tr>
</thead>
<tbody>
<tr>
<td>Spend enough time with him or her?</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Encourage your to ask questions?</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Encourage your to raise concerns?</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Consider your opinion about options for his or her care?</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Listen carefully to you?</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
<tr>
<td>Provide you with the specific information you need?</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
<td>☐</td>
</tr>
</tbody>
</table>
SECTION 7: Family Impact

NOTE: The following questions refer to the past 12 months ONLY

Did your child's health care needs cause financial problems for your family?
☐ Yes
☐ No
☐ Don't know

How often did you feel overwhelmed by your child's health care needs?
☐ Always
☐ Usually
☐ Sometimes
☐ Never

What factors added to your stress level as a caregiver? (SELECT ALL THAT APPLY)
☐ Financial stress
☐ Personal emotional health concerns
☐ Personal physical health concerns
☐ A lack of training/skills needed to provide care to your child
☐ Employment concerns (such as difficulty getting time off of work or needing to reduce work hours)
☐ Issues with health insurance
☐ Other caregiving responsibilities
☐ Difficulty getting breaks from caregiving
☐ Difficult relationships with other family members or care providers
☐ Other factors [Please specify what other factors added to your stress level as a caregiver: ___________ ]
☐ No factors added to stress level

Please indicate the caregiver support services your family used: (SELECT ALL THAT APPLY)
☐ Caregiver education/conferences
☐ Counseling
☐ One-on-one training
☐ Respite care services
☐ Support groups
☐ Religious groups
☐ Advocacy organization or association
☐ Other caregiver support services [Please specify what other caregiver support services your family used: ___________ ]
☐ No caregiver support services used

Please indicate the caregiver support services your family did NOT use but would WANT to use, if available: (SELECT ALL THAT APPLY)
☐ Caregiver education/conferences
☐ Counseling
☐ One-on-one training
☐ Respite care services
☐ Support groups
☐ Religious groups
☐ Advocacy organization or association
☐ Other caregiver support services [Please specify what other caregiver support services you would want to use: ___________ ]
☐ No caregiver support services wanted
SECTION 8: Neurometabolic Clinic Experience

Do you believe your child’s needs are being met by the Neurometabolic Clinic at CCHMC?

☐ Yes
☐ No
☐ Don’t know

What are the areas of strength at the Neurometabolic Clinic? (This question is voluntary; you may decline to answer by skipping the question.)

What are the areas that need improvement at the Neurometabolic Clinic? (This question is voluntary; you may decline to answer by skipping the question.)

What additional services or specialists would you like to be made available to your child at the Neurometabolic Clinic? (This question is voluntary; you may decline to answer by skipping the question.)