
Families' Experiences of Newborn Screening for MPS-I, Pompe Disease, and X-ALD

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Project Team

Lisa Anderson
Ashley Johnson
Mariyam Naadha

Enterprise Director, MAD

Ryan Church

Assistant Director

Beth Bibus

Contact Information

Telephone: 651-259-3800
Email: Management.Analysis@state.mn.us
Fax: 651-797-1311
Website: mn.gov/mmb/mad

Address:
658 Cedar Street
Centennial Office Building
Room 300
Saint Paul, Minnesota 55155

Management Analysis and Development

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Executive summary

Overview of project

The Minnesota Department of Health (MDH) began newborn screening (NBS) for mucopolysaccharidosis type I (MPS I), Pompe disease, and X-linked adrenoleukodystrophy (X-ALD) in 2017. MDH was interested in understanding the needs and experiences of families and caregivers of children/youth with MPS I, Pompe disease, and X-ALD to inform public health follow-up protocols. MDH asked Management Analysis and Development (MAD) to conduct in-depth, qualitative interviews and analyze the findings.

Methodology

MAD and MDH collaborated to carry out the project. MDH recruited participants through various networks and mediums, including social media. MDH provided MAD with the contact information of 26 families from across the United States that volunteered to participate in the project. MAD completed in-depth qualitative interviews with 24 families from May to June 2018. Interviewees consisted of 11 families with children/youth who had been diagnosed with Pompe, seven families with children/youth who had been diagnosed with MPS I, and six families with children/youth who had been diagnosed with X-ALD.

MAD asked families to discuss their experience related to:

- receiving the diagnosis and how they obtained further information about the condition,
- facilitators and barriers to families in attending to the health care, social, and educational needs of their children,
- the impact of diagnosis and care on family and personal life,
- supports accessed by families and family-identified resources/services, and
- advocacy roles families have taken on since their child's diagnosis.

Findings

The findings presented in this report are solely based on the opinions expressed by the families that were interviewed. MAD transcribed, reviewed, and coded the interviews to identify key themes and insights, using the main question categories as organizing principles. In the analysis of interviews, MAD has attempted to strike a balance between shielding individual identities and providing the maximum amount of useful information in this report.

Diagnosis and condition education

- Across all three diagnoses, several families discussed challenges with a lack of primary care provider (PCP) knowledge of their child's condition (i.e., choosing to defer to specialists for treatment decision and family education). Several families, however, discussed positive experiences with PCPs, such as

when their PCP was not necessarily an expert on their child's condition but provided referrals, when they were willing to treat their child for basic medical needs, or when they tried to learn more about the diagnosis and condition.

- Several families described interactions with specialists or specialty teams that were perceived by families as insensitive.
- A majority of families identified a lack of accurate and up-to-date information available on the internet as a significant gap in their experience of learning more about the condition, especially after receiving a positive NBS or while waiting for initial specialist consult.
- Several families described educating themselves about the condition through societal or professional organizations, conferences, and their medical/specialist teams. Several families also discussed learning more about the condition through other families with whom they had connected.
- Families highlighted a variety of information that was missing since their child received a positive screen or diagnosis, ranging from information about confirming a diagnosis, information about which specialists they needed to see, on-going medical and monitoring needs as children get older, treatment options, and updated information about medical advances. Some families discussed the need for prompt, condition-specific information that should be available when children receive a positive NBS screen or diagnosis.

Day-to-day needs

- A majority of families discussed the complexities of navigating systems and processes such as insurance and support services. Nearly half of the families noted that care coordination (i.e., managing appointments, treatments, therapies, and communication between medical teams) takes a significant amount of work on their part.
- Common challenges in meeting day-to-day needs included a lack of local resources (e.g., medical, social, educational), and the need to take time off work or obtain flexible work schedules to attend to the needs of their child.
- While several families reported good or fairly good insurance coverage, close to half of the families discussed challenges with insurance coverage, including the continual need for specialist referrals or prior authorization for procedures, and difficulty getting coverage for tests, procedures, or treatments.
- Half of the families discussed their satisfaction with accessing early intervention, highlighting the value of obtaining services in the home. Nearly all of these families had children who were symptomatic.
- While several families found schools and child care providers to be accommodating and responsive to their needs, a few families with older children also discussed the need to advocate for more accommodations as their children aged.

Impact on families

- A majority of families noted making choices around employment (i.e., quitting a job, working more hours, or working fewer hours). Most commonly, the primary caregiver quit their job or went down to part-time/reduced hours or flexible schedules to take on the role of coordinating care and attending to the needs of the child.

- A few families also discussed relocation (permanent and temporary) for treatment or proximity to a specialized medical facility.
- Several families discussed the effect the child's condition had on family and personal life, including hardship on their other children due to demands on parents' schedule and time, strain on marriage, personal and familial mental health, as well as the financial impact and burden, including debt and bankruptcy.

Support

- A majority of families noted the day-to-day support they receive from (and provide to) other families with children who have the same condition, including consulting with other families in identifying care teams, and developing treatment plans.
- A majority of families said that they had been able to connect with other families through social media groups, and a few families noted that their specialist, genetic counselor, or pharmaceutical company had also connected them.
- Other common support systems included societies or professional organizations, medical teams, and friends and family.
- Families discussed a variety of additional supports that would be helpful, including mental health support for siblings, post-partum mothers, and spouses or partners.
- A few families discussed the need for condition-specific information and guidelines for care as additional support they needed.

Advocacy and hope

- Most commonly, families discussed having to advocate for consistency in care across providers as well as coordinating messages and follow up across providers.
- A few families discussed having to advocate for accommodation for their children to have inclusive environments in their schools and social life.
- Several families discussed either themselves or their child taking on an advocacy role in their community or nationally, including advocating for inclusion of the condition in the NBS panel in their state, or advocating for a particular treatment.
- Most commonly, families discussed hopes for their children having the best quality of life possible, and for more or alternative treatments or cures to become available.

Recommendations

MAD developed the following recommendations based on the information received from the interviews with families. Though families identified many support gaps in their journeys with MPS I, Pompe disease, and X-ALD, or system gaps that were not specific to Minnesota, MAD's recommendations below focus on gaps that MDH can play a role in supporting or public health systems can address. MDH should explore these possibilities in the broader context of organizational goals and other priorities.

- **Develop or compile verified and up-to-date, condition-specific information:** Families expressed the need to have early and easy access to accurate, condition-specific information and to be connected to the right resources and supports as soon as possible. MDH should develop and share information about the condition (including clinical guidelines, when available) and resources to support PCPs and families when children screen positive for MPS I, Pompe disease, or X-ALD and after the diagnosis is confirmed. Information should include an overview of the condition and connections to commonly used or needed resources (e.g., financial assistance, early intervention, insurance options such as Medical Assistance, mental health services, and respite care). In addition, MDH should provide information on social and medical supports, such as social media groups, professional societies, and networks of hospitals and clinics that specialize in caring for children with the condition.
- **Support health care providers in providing information to families:** Families discussed challenges with communication between them and PCPs, specialists, and genetic teams (i.e., lack of provider knowledge about the condition, insensitive and poor messaging, etc.). MDH should provide support to PCPs, specialists, and genetic teams in providing information and education to families once a child receives a positive NBS result. This could include support to improve parent-provider communication and to strengthen the role of providers in addressing the concerns of families who have received a positive NBS result or diagnosis.
- **Support families in care coordination:** Families also discussed that there is often not much support for care coordination and management (e.g., medical appointments, insurance navigation) as well as other day-to-day needs as children age (e.g., education). Given these gaps, MDH should support capacity building efforts to strengthen systems to address care coordination needs of families. This could range from supporting families in navigating complicated systems in the short-term (e.g., families receive information that is comprehensive, up-to-date, and helpful), to long-term support to stakeholders to build their capacity to address care coordination needs.
- **Facilitate peer-to-peer and networking opportunities:** Families discussed direct connections with other families as one of the most important forms of support they received. MDH should consider facilitating connections with peers for support, matched when possible, on families' specific needs, phenotype, severity of symptoms, and similar experiences.
- **Convene and collaborate with stakeholders to address larger gaps:** Families discussed gaps in systems such as insurance, and care coordination within specific hospitals or clinics as major challenges in the management of care needs of their child. MDH should explore internal and external partnerships and convene stakeholder groups who can address these larger system gaps. This can include working with legislative partners or Minnesota Department of Human Services to close gaps in insurance systems.
- **Further evaluation of the NBS system:** Given that these conditions are relatively new to the NBS panel, MDH should consider further exploration of supports at a later stage of the program. This should include conversations with stakeholders such as PCPs, specialists, and families. MDH should improve community engagement efforts to reach traditionally under-represented families such as people of color and families that speak another language to gain an understanding of their needs and perspectives. It should also include an evaluation of the larger NBS system at MDH to assess the effectiveness of the program in addressing the needs of children identified with these conditions.

Full report: Introduction

The Minnesota Department of Health (MDH) administers the newborn screening (NBS) program in Minnesota. The program works closely with hospitals, laboratories, and medical professionals across the state, and screens newborns shortly after birth for over 60 disorders. Minnesota began screening for mucopolysaccharidosis type I (MPS I), Pompe disease, and X-linked adrenoleukodystrophy (X-ALD) in 2017.

As MDH moves through rolling out follow-up protocols for NBS conditions, the need to understand the experiences of families and children who have MPS I, Pompe disease, and X-ALD has arisen. MDH asked MAD to conduct in-depth phone interviews with families across the United States to understand families' perspectives on information, services, and support needed once their child receives a positive newborn screen or diagnosis for these three conditions.

Methodology

MAD and MDH collaborated in conducting the interviews. MDH invited families to participate in the project through various networks and mediums, including social media¹. Interested families then contacted MDH project staff to provide their contact information to MAD. MAD contacted 26 families over the phone and completed in-depth interviews with 24 families from 14 states in May and June 2018.

The interview topics included:

- Families' experiences receiving the diagnosis and obtaining further information about the condition.
- Facilitators and barriers to families in attending to the health care/social/educational needs of their children.
- Families' decision making after their child's diagnosis.
- Impact of diagnosis and care on family and personal life.
- Supports accessed by families and family-identified resource/service gaps.
- Advocacy roles families have taken on since their child's diagnosis and families' hopes about the future of their children.

The findings presented in this report are solely based on the opinions expressed by the families interviewed. MAD transcribed, reviewed, and coded the interviews to identify key themes and insights, using the main question categories as organizing principles. In the analysis of interviews, MAD has attempted to strike a balance between shielding individual identities and providing the maximum amount of useful information in this report. MAD has adopted the following conventions in this summary:

¹ MDH sent recruitment flyers to condition-specific advocacy organizations (e.g. MPS society), national NBS clearing house (Baby's First Test), and health care providers, in addition to posting flyers on MDH's Facebook page and condition-specific social media pages.

- General terms like *many*, *several*, or *a few* are used instead of reporting frequencies or percentages of responses.
- General descriptors such as *specialty hospital*, *condition-specific organization*, or *academic health center* are used as substitutes to protect individual identities of families who participated.
- To provide more concrete qualitative information, paraphrased statements from interviewees are included *in italics*. Though the statements accurately reflect the sentiment and content of interviewee comments, they should not be viewed as direct quotations attributable to individuals.

Demographic background of families

MAD completed interviews with 24 English-speaking families; 11 families with a child/youth with Pompe, 7 families with a child/youth with MPS I, and 6 families with a child/youth with X-ALD. Five families had more than 1 child who had a positive diagnosis for the condition. Nine families indicated their children had been identified by newborn screening while 15 families said they had received a diagnosis through clinical presentation. The children of families that MAD spoke to range from 3 months old to 20 years in age. Nine of the interviewees had post-graduate degrees, while seven had 4-year college degrees, three had 2-year degrees, two had attended some college, and two had high school diplomas or a GED. The families that participated reside in 14 states located across all seven Regional Genetics Networks². All respondents identified as white/ Caucasian.

Findings

Diagnosis and treatment

Participating families were asked to discuss their experience with diagnosis and treatment. Often, they discussed how their child was diagnosed (i.e., through newborn screening or clinical presentation), the role of their pediatrician, and the role of specialists or specialty teams.

Diagnosis

Many families learned about their child's condition through clinical presentation. That is, the child began exhibiting signs and symptoms of the condition, which often led to a series of tests, lasting variable amounts of time. Several families learned about their child's condition through NBS. A few of the families that received a positive NBS test received multiple borderline results, leading to a brief period of unknown diagnosis. Additionally, a few NBS families experienced lengthy waiting periods in identifying their child's phenotype or the right treatment option.

"I wish I knew more about Lorenzo's oil—there are a lot of different answers about that. I've gotten iffy answers about different treatments and what's available from providers. I have been getting a lot of

² The Regional Genetic Networks are an initiative established with support from the Maternal and Child Health Bureau of the Health Resources and Services Administration, Genetic Services Branch, in an effort to improve health of medically underserved communities and to bring genetic services closer to local communities. For more information visit: <http://www.nccrcg.org>.

information from a Facebook group for parents of children with this condition. However, everyone seems to have a different case, so it's hard to just trust that. Even the doctors have different things to say, so I'm really just going day-by-day."

Support for NBS

Several families were supportive of NBS in identifying their child's condition. In each case, children had early-onset symptoms.

- *"I really wish that this condition was something that could be in newborn screening because it's such a severe disease even though it's rare. If we had known earlier, she wouldn't have had any developmental delays. I fought for the first year of her life trying to figure out what was wrong with her."*
- *"The main thing is the importance of how early diagnosis and treatment is. Even not knowing for the first 6 months of her life, it damaged her muscles to the point she can't stand."*

However, a few families whose children had a late-onset condition or whose children were asymptomatic expressed mixed feelings about NBS.

- *"This is a really grey area and I think when late onset is part of newborn screening, it's outside of the scope of newborn screening—I feel like I should have been given a choice. I chose not to do genetic testing before or during pregnancy but then I get this information and now I'm being told by experts that with her phenotype she probably won't even have any symptoms. I think they neglected to think about long term consequences. Now she has a pre-existing condition, so even though she's fine, she's disqualified for life insurance, disability, and possibly health insurance depending on where that goes, so that's concerning to us. Also, we're going to have to tell her about her diagnosis at some point because her siblings don't have all of these appointments—how will this information change the choices she'll make in her life? I think if we all had gene sequencing we'd all find things we might not want to know. With something like this, informed consent needs to be initiated—more than just: here sign this form—it needs to actually be a discussion."*
- *"I really do feel like I wonder what would have happened if I'd known that at 2 weeks—it's a hard time for parents and mothers with postpartum depression and routines being shaken up. I got almost a year for him to be my buddy and for me to be in ignorant bliss—I was so attached to him at that point and I knew nothing would change about how I felt about him and I'd just fight harder."*

Primary care provider interaction

Several families across all diagnoses discussed challenges regarding a lack of primary care provider (PCP) or pediatrician knowledge of their child's condition. These families mentioned that their PCP often deferred to specialists for treatment decisions and family education.

- *"I've been taking her to this pediatrician since she was born and I always had concerns about her breathing because it sounded like she was snoring all the time—but he kept saying it's normal, even though she wasn't sitting up on her own or she wasn't crawling."*

- *“Our own pediatrician couldn’t tell us what the condition was. We were kind of in the dark for these couple of weeks and if you google it, it tells you your child is going to die between 4 and 10 years.”*
- *“Our pediatrician told us. I didn’t take the call, my husband did. And she told us he tested positive for something and we had to go to the hospital and a genetic counselor or nurse practitioner would explain it. It’s a devastating call for any new parent to get. To this day, she maintains she’s not the expert on this. For condition-specific questions, I would go to the specialist.”*

A few families also noted that non-primary pediatricians were hesitant to treat their children for certain medical needs, such as ear infections, due to their condition.

- *“His overall health got better which made the physician partners be a little more open to treating him for generic things like tubes in his ears—because nobody would before—or a tonsillectomy, or fix his hernia—basic medical care—nobody would do it because they considered him too sick.”*
- *“When my son had ear infections—if we can’t see our pediatrician for basic things like that, it’s hard because she knows about his condition and is fine with prescribing him antibiotics because she knows it won’t affect his other treatments—a lot of doctors aren’t comfortable prescribing because they don’t know about the diagnosis so I have to spend 30 minutes of the appointment explaining it and I end up having to go through [the specialty hospital] anyway. So sometimes, rather than just going to a walk-in clinic, we go to the emergency room at [the specialty hospital] just to get approval for the antibiotic.”*

Several families, however, discussed positive experiences with their PCP. Though these families acknowledged that their pediatrician was not necessarily an expert on their child’s condition, they expressed gratitude for the provider’s willingness to make referrals, treat their child for basic medical needs, and learn about the diagnosis.

- *“We had a great pediatrician who we love—and she loves him and goes above and beyond in caring for him in terms of allowing us to do things for him at home, but she needed a backup person for her to treat him so that she wasn’t walking out on a plank by herself and there just wasn’t one around. It wasn’t easy for her to get in touch with people or other providers—everyone is really busy and overworked and their world revolves around more than just one child. I wish there had been greater access for local providers to have at least a support system because there are a lot of things that can be done for these kids within their medical home that we don’t get access to, mainly because there is not a lot of support for providers who are occasionally willing to do things without sending them to major medical universities.”*
- *“Initially, we talked to our doctor, we trusted him enough and he spent hours on the phone to research and walk us through it.”*
- *“Our pediatrician has been wonderful. My son had a fever a couple months ago inexplicably. Our standard pediatrician wasn’t on call but there was another one and when we walked in he knew about his adrenals which was like—oh my God, thank you. I don’t know what is written in our file, but whatever she did is amazing because they’re very in tune with him and they’re on top of it.”*

Specialist interaction

In addition to discussing support from pediatricians during initial diagnosis, families discussed interactions with geneticists or other specialists/specialty teams during this time. While families described the information and

support they received from specialists or specialty teams, several families interviewed described interactions with specialists or specialty teams that were perceived by parents as insensitive or that used poor messaging. These interactions, in part, led most of these families to change care teams, seeking a team of providers that would be supportive and a better fit for their treatment goals.

- *“One of the other things I did was take him to a specialist. He had heard of the condition, so he was a little familiar with it but even he said to me ‘why is this kid here?’ And I said ‘because we were willing to take him.’ He said ‘they should have never placed him in your care; they should have placed him somewhere where he would have had access to a major university.’ And at that point I was too timid to speak up—now I would say a lot of things to a provider that would say something like that to me. I can’t imagine how people feel when they hear things like that.”*
- *“We asked them about future children and if we would have to use in vitro and they said that there was a 1 in 4 chance that the child would have the condition, so we could go ahead and try and have an abortion if it came up. Really? That’s your suggestion?”*
- *“In our first appointment with the specialist, once we got the diagnosis I said ‘so basically I’ll never sleep again.’ The doctor said ‘he’s 10 months old so you’ve had 10 months to sleep.’ I thought ‘I’m done with you, we can wrap this up and go now.’”*
- *“The last thing he said to us was—by the way, you probably need to reconsider being his foster parents because over 50% of marriages of children with disabilities end up in divorce. But we’d been married 20-25 years at that point so we just said we aren’t going anywhere.”*
- *“We got the call for my son and it was a geneticist—she was rude and told us we had a better chance of winning the lottery.”*

Condition education

When asked about families’ experience getting information about their child’s diagnosis, a majority of families discussed a lack of information available once their child received a positive NBS result or had a confirmed diagnosis. Almost all families, irrespective of condition or how they received the diagnosis, discussed the lack of accurate and reliable information on the internet. Several families recommended not searching the internet for education, saying that information available online was outdated or too new to be reliable. Across diagnoses, families described how the little information available caused fear and anxiety.

- *“The first thing our provider said was, don’t google it. But obviously no one is not going to google that. Yes, that was horrifying. The problem is that the information that comes up when you do a web search is the horror stories, you read about the slow progression and you assume that’s what’s going to happen to my son.”*
- *“I went to Google and it was the worst thing I could have done. Don’t ever do that.”*
- *“I wish I had not looked up everything on the internet. Information is old on the internet. I was devastated when I looked it up. But once I found out there’s a treatment, it made me feel better. In the Facebook community group, we always get these new people who are devastated. The biggest piece of advice is not to google it. There’s some stuff that’s more accurate, but a lot of it says with infant-onset—*

it's fatal and they'll die and mostly its horrible news. But if you read medical journals, you get better information. But there isn't a lot of positive information on Google."

Several families described educating themselves about the condition through societal or professional organizations, conferences, and their medical/specialist teams.

- *"[Condition-specific organization] and the [academic health center] have been the best support. [The academic health center] is cutting edge in everything for his disease. Everything we need to know, we can get from there. Everybody there knows everything. [Condition-specific organization] has a lot of knowledge and resources. And handbooks. I'm not able to go to all the conferences, I can only go when it's driving distance."*
- *"Prior to going to [academic health center], we didn't know all these things we were supposed to be doing. Once we went to the [academic health center], we got a whole list of things we needed to do. Going to [the academic health center] was probably the best thing for him and for us. His care here would have been, not lacking, but they just didn't know."*
- *"I personally, I don't search because I get discouraged. I wait for the conference every year and that provides us with more concrete information. So I'd rather just wait a year to get that from a source of information that I can trust."*

Several families also mentioned learning more about the condition from interactions with other families including through various social media forums.

- *"I think the information on Google is outdated so definitely updated information would be more useful. The best information and best guidance has come from other families and having conversations with them and the conferences. I don't think I have googled any information in over a year and half. It's not accurate anyways, but I can have a conversation with another mom who's going through it and can give me accurate and concrete information about their child."*
- *"Through other families, so they are big source of information. That's how I found out about the [academic health center] conference and my doctor didn't know about it."*

Families were also asked about what information they felt was missing or lacking since their child received either a positive NBS result or diagnosis for the condition. They discussed a variety of information, ranging from information about confirming a diagnosis, information about specialists that they needed to see, treatment options, on-going medical needs and monitoring needs as children age, and updated information about medical advances.

- *"Information needed was about on-going medical treatment, connecting to other specialists, and what symptoms and signs we needed to look for. Also looking for a community of other parents. I wish I had had more information about how you get diagnosed, obviously. We went through a couple of weeks of wrong diagnosis. I wish geneticists knew the proper tests to diagnose."*
- *"At that point we just wanted answers, answers on his diagnosis, from someone, doesn't matter who. They were saying, 5-8 year life span, but nobody had talked to us about treatment. So there was just an information gap. He was about a month old when we finally met with the hematologist. We were in*

limbo for about a month, not knowing what was going on or what the next steps were. Even with transplanting so young, and we didn't know what to expect. The [academic health center] gave us more realistic information to manage our expectations. Even if it wasn't what we wanted to hear, it was good to know the facts and know what we're dealing with. I just wish we had known more about the treatment, and what to expect. We were just getting lots of conflicting information. I'm someone who likes to know, even if it's bad. Straight information that was accurate was probably the most important."

- *"I want more information about experience of other families, more about how other families are doing—that's what's nice about the [condition-specific forum] and the Facebook pages. More examples of experiences with what they were dealing with, what treatments they were and weren't doing. I want to hear from parents and the doctors that have actually dealt with this condition."*
- *"Getting connected to doctors who are knowledgeable is important. But it's going to be hard to find doctors who are knowledgeable right at newborn screening. Getting doctors who are familiar with the condition would be helpful. But we have to share about the condition, and everyone asks what is that? —and very rarely are they familiar with it. We're educating doctors, and I'm not as educated as many parents. We haven't had the onset of symptoms, but I know enough to share a little. Getting connected to knowledgeable doctors and also getting that emotional support from other families is important."*
- *"The hospital is great with paying bills and insurance, but is not very good with giving you information on next steps when you go home, such as treatment or therapy. There needs to be more out there."*
- *"Now our focus is on trying to find out what research is going on, status of studies, and if trials have been successful. Difference between trial drugs and what she's getting now. We're learning about what's next."*

Several families, irrespective of how they received the diagnosis, highlighted the need for prompt, condition-specific information that was in plain language when children receive a positive NBS screen or diagnosis.

- *"What we want is for when families get diagnosed they should get a brochure to get information, to get the right resources, so they are not scared to death. They are not getting that information. Pediatricians have zero information, so they don't know anything about the disease. They don't have literature we put together. They do a Google search, which gives them bad information. Those poor families come out scared to death."*
- *I don't know if there's a way for information to be more readily available, because Google shows the worst-case scenario. We found out that Google shows that information because newborn screening for the condition is so new, so before when people found out they were too far along to do anything about it. So there's not a lot for people who find out early and treat it right away. It's important for there to be better information, like a website for people to look at while they're waiting for the appointment."*

A few families emphasized the importance of guidelines or standard of care.

- *"It would be beneficial for new parents to come in with some guidelines. Our counselor did provide something like that—a protocol that mapped treatment out at different ages—so we could keep up with it all, and that helped."*
- *"I hope that they do come up with a standard of care in terms of monitoring for newborn boys."*

Day-to-day needs

Families discussed a variety of day-to-day needs, such as insurance coverage and navigation, accessing services, child care or school, and care coordination.

Insurance coverage

A majority of the families reported having private insurance as their primary insurance coverage with only a few families indicating they had insurance through the state as their primary insurance. One family noted having private insurance as their primary coverage, with secondary insurance through the state that covered payments not covered under private insurance.

Responses regarding insurance coverage were mixed with no discernible difference based on type of primary insurance. Several families reported good or fairly good coverage in terms of access to providers, approval of tests, procedures, or treatments, and the costs related to medical needs being fairly low. Nearly half of the families, however, reported challenges with insurance coverage. Challenges included the continual need for specialist referrals or prior authorizations for procedures, and fighting for coverage of tests, procedures, or treatments.

- *“In the insurance world, they just fight. It’s always a fighting battle trying to get what you need. We’ve never had anything declined, we’ve never been denied or said no, but it’s always a fight. Home infusions have been the biggest fight. The hospital is 1.5 hours away—that’s a long day, 3 hours driving, with infusions taking 6 hours. So I wanted her to be home, because that would open avenues in the future for schooling, instead of missing a whole day of school every week. It was a fight to get that approved, it took almost a year before it was approved.”*
- *“Our insurance has been a nightmare. They flat out tell you that it’s your responsibility to make sure everything is covered and approved. We got referred to our treatment center, and we thought everything was sorted out and approved for infusion, medication, and physician visits. Then I started getting bills in the mail and we found out nothing was covered.”*

A few families reported reaching out to medical teams for assistance with appeals to denied medical services, often in the form of physician letters to private insurance companies.

Half of the families discussed high deductibles and high out-of-pocket maximums, though only about half of those families discussed the costs as problematic. There were no significant differences between families that discussed these costs as problematic and those that did not. Many families mentioned seeking other means of payment for medical expenses. Most commonly, families with private insurance discussed applying for secondary insurance coverage through their state’s Medicaid program. Several families also discussed fundraisers (i.e., organized by families or friends) and patient assistance organizations as means for covering costs. A few families of children with a Pompe diagnosis noted receiving copay assistance through pharmaceutical companies. A couple of families reported being in significant debt or filing for bankruptcy due to an inability to pay for medical bills.

Several families discussed the complexities of navigating the insurance system, most of whom reported that they received support from genetic counselors, case managers, or pharmaceutical companies for insurance needs. Challenges related to navigating insurance systems varied, but included issues such as a loss of coverage or hitting life-time maximum coverage, denial of out-of-state treatment, and a lack of plain language within the system.

- *“Over the years, we have hit all the insurances. Initially we had private insurance but we had to switch to a different one because she hit her life-time maximum coverage. We switched again, but we knew we needed to go on state health care because we were hitting her maximum coverage in all of them. But getting state care was hard, and it took a lot of research to figure it out. We ended up talking to different case workers. The pharmaceutical company also helped a lot – they had different case workers that helped navigate insurance.”*
- *“The other problem is traveling outside of our state to get specialty care—they don’t want you to do any of that. We got approval a couple of times to go to out of state for treatment and procedures. However, when we needed to go out of state for a follow-up due to a procedure complication, we found out that our state’s Medicaid had not paid the other state’s Medicaid for our original trip, so they wouldn’t let us go back to see the surgeon. The other hurdle is getting to out of state providers that we might need to see sometimes to get some knowledge and get some stuff done and bring that knowledge back to our local providers where we want to receive care. Getting to those people is almost impossible in our state.”*
- *“Our insurance has assigned us one person so that every time I call, I can talk to the same person, but she speaks insurance language and I don’t, so I don’t find that very helpful. So if there was a way to simplify and understand that better, because I would like to get more resources for my daughter and advocate for her more, but I don’t know what else to ask for or what she’s eligible for. The insurance company doesn’t broadcast every resource, and I don’t know how to ask.”*

Early intervention

Families also discussed accessing other services, such as early intervention. Half of the families highlighted their satisfaction with accessing early intervention services, or noted their importance. The most commonly mentioned early intervention services were occupational therapy (OT), physical therapy (PT), and speech therapy (ST). Nearly all families accessing services had private insurance as their primary carrier, had children who were symptomatic, and had MPS-I or Pompe diagnoses. Only a few families reported not accessing early intervention services, however, these families had children who were asymptomatic; therefore, families did not feel that early intervention services were necessary or had not had a need to access these services at the time of the interviews.

Among the families that did access early intervention services, several discussed the value of obtaining services in the home, noting that it allowed them to spend more time together as a family, provided breaks for family caregivers, and helped their diagnosed children miss less school. Only one family that originally received services in the home shifted some of their services to the outpatient setting.

School/daycare

Responses on the supportiveness of schools or child care providers were mixed, though only about half of the families had enrolled their child in child care or school. While several families found schools and child care providers to be accommodating and responsive to their needs, a few families reported more mixed experiences. These families noted that, overall, their education system had been supportive, but that there were cases where they had to advocate more strongly on behalf of their children.

“All of his therapy is provided through the school system and if they didn’t do it we would have to travel 120 miles round trip to get it. The flip side is that he is the only kid that has ever come through the school system that has been so medically complex. There just isn’t another one like him and there probably won’t be another one like him for a long time. I’ve had to educate myself and basically be the team leader and tell the school what they need to be doing. And if I didn’t do that it wouldn’t get done. They love him and they care for him but as far as being able to help him, they don’t know what to do for him so I’ve had to do all of that research and give them that information. They’re still not great at implementing it.”

A few families with older children also discussed the need to advocate more for accommodations as their children aged and wanted to participate in day-to-day activities in the school, or in school outings.

“I definitely play an advocacy role. For example, there is a class retreat. They are looking at her IEP and all the issues and problems she has, and they said she can’t go unless her parents go. She’s in scouting, she goes camping – all with injectable medication. So they are saying, let’s put it on hold. She has a lot of problems, but she has overcome a lot of them and adapts and does things in her own way.”

Care coordination

Nearly half of the families discussed care coordination. These families noted that it takes a significant amount of work on their part to manage appointments, treatments, therapies, and communication between medical teams.

“It is absolutely crazy. She has an appointment every day of the week. It’s a lot, and you’re a parent who has to deal with that. Doctor appointments, therapy appointments, keeping up on the follow up—it’s a lot of work. Sometimes I feel like I’m on the phone more with doctors or therapists than I am playing with her. The last thing I want to do is fight with people who don’t know my daughter. I want to enjoy my time with her, because I don’t know how much time I have. I have tried to get services to help me with those things. With case managers, they are so overworked themselves that stuff falls through the cracks, so I tend to do a lot of it anyways. If you were put in the situation...you do what you have to do. No one is going to be her voice to stand up for her, so I have to do it.”

In order to ease the burden of care coordination, several families reiterated the importance of supportive care providers in attending to their child’s day-to-day needs, such as determining treatment plans, appealing denied insurance claims, and coordinating more convenient care.

“At our hospital, if they know we’re having an infusion at a certain time, they’ll stay late and they will see her after or during her infusion. Or, they will see her first thing the next morning if they really can’t do it during or after. They work around our infusion dates, rather than when it’s convenient for their schedules.”

A few families reported that they received support from pharmaceutical companies related to navigating insurance and other services.

“We had people who were trying to help. We had a transplant team, nurses, case managers—but they would all tell you something different. The case manager for the pharmaceutical company was the one who sorted everything out.”

The two most common challenges in meeting day-to-day needs included a lack of local resources (e.g., medical, social, educational) and the need to take time off of work or obtain a flexible working schedule. Flexible scheduling and work absences were discussed by parents of both children who were symptomatic and asymptomatic.

Other needs identified by families

A few families also discussed accessing services to assist with transportation or housing. Most commonly, these families discussed transportation services that provided financial assistance for mileage or plane tickets. However, a few families noted that the need to track mileage made the use of these services challenging. One family reported connecting with a service for mortgage assistance.

Several families mentioned a variety of additional supports that were helpful or that they would recommend for families. Most commonly, they discussed mental health support or grief counseling for a variety of audiences such as siblings, post-partum mothers, and spouses or partners.

- *“I think I was trying to do too much and not asking anyone for help and I fell apart for a little bit and had to get my way back to a good place. I got counselors and therapist. I checked myself in to a rehab and outpatient. I needed the kids and the kids needed me. And then I started going to meetings, and there was some support there. And then the therapist I talked to for a few months. There was help that way and I knew how to reach out to get help if I needed it.”*
- *“I wish there was support for parents to help them accept and deal and cope with it. This didn’t happen until two and a half years ago for my husband, who denied the diagnosis—and because of that, there were a lot of times that dealing with the burden of organizing care while working was on me. And there is so much travel time for treatment. So I gave up my job, which I do not regret, but there was no support and nobody educating my husband. Someone from the pharmaceutical company spoke one-on-one with my husband for an hour and a half and he could ask any questions and get answers. And that was very critical because that was a changing point in our family because finally the family was all on the same page. Once my husband was on board it was easier to manage the family because he would take some of the burden for me. I understand it’s a grieving process and we all grieve differently and it takes time. But we were never offered a counselor or to speak with another family.”*

- *“I think just in terms of family therapy that would be really important past the short-term because I don’t know when to tell my daughter about our son. Our pediatrician has been great and has helped us and told us that the condition is not him being sick because we don’t want her to be sick with the common cold and have her think she has this condition because she’s sick. We also don’t talk about the outcomes with her. We’re all aware of what the statistics are for each phenotype so we don’t talk about it anymore. But direction in short- and long-term on how to navigate conversations with family members would be helpful.”*

A few families also mentioned needing respite or nursing care to help with meeting the day-to-day needs of their child.

“Neither my husband nor I have slept in a year because she doesn’t sleep through the night, I think because we had to feed her overnight using the NG tube. I was doing sleep training with my other daughter at this age and she was sleeping through the night at this point. But with my daughter, we have to get up and feed her. I’ve been starting to look to see if there is any type of overnight babysitter because we’re so tired. When she had her feeding pump, every three hours I was hooking her up to the feeding pump and doing that all day and all night every three hours.”

Impact on families

Families discussed a variety of impact the diagnosis and their child’s condition has had on their personal and family life. Many families mentioned the impact it had on their work life, including the need to quit their job, or reduce hours, or in a few cases needing to work more hours. Most commonly, the primary caregiver (or mother in majority of the instances) quit their job, or went down to part-time/reduced hours or flexible schedules to take on the role of coordinating care and attending to needs of the child.

- *“I work 75 percent, which has been the biggest factor in managing his care. Beyond the infusions, there are many doctor’s appointments that he has to go to, once a year, and 20 different doctors he has to see. So I work reduced hours. After he was diagnosed I went down in hours to manage that.”*
- *“My work schedule has always been very helpful. When I had her, and early on after she was born, I only worked weekends. And that allowed me to go to appointments, or stay home if she’s sick. Later on I found a job that would let me work from home so that’s what I do now. The most time is spent on her care, medical appointments, treatments, which is why my schedule has been really helpful.”*
- *“I don’t know how anyone will get any stuff done, I’m on maternity leave and there are days when I’m on the phone all day. In terms of coping, I’ll be taking unpaid leave for all this [treatment].”*

Even in instances where the child had a late-onset condition or was asymptomatic, caregivers discussed the time and effort required to gather information about the condition or to monitor their child’s health and its impact on their work life.

“I worked up until my due date. We could afford it for me to stay home. If he hadn’t been born with it, I would have probably gone back to work. Me staying home has really helped us figure it out. I don’t think anyone has especially helped me navigate. I don’t know how I would do this if I was working full-time.”

A couple of caregivers expressed sadness about the lack of career progress or loss of job to take on the role of care coordination and monitoring their child's health.

"Reducing time at work is the biggest one. I haven't progressed in my career because its part time-ish. I have also had subsequent children. Just my working hours and having to deal with all the doctors and service providers- it's a lot of work. I say I have three jobs—I am a mom, I have a regular job, and I'm his care coordinator/advisor/nurse. It's a huge commitment. Does the medical benefit keep me at my job? Maybe. I might have left for a higher pay if I didn't risk losing my insurance."

Several families discussed the effect the child's condition had on other children in the family, balancing the needs of all the children in the family, and impact on social needs of the family.

- *"You can't forget your healthy child and she has her own issues, like anxiety. She worries about her brothers, she knows they have this condition. She worries about that. I take her to see someone to talk these things out. I just want her to be able to enjoy her life as well."*
- *"It's also hard because we have another kid, an 8 year old. Having all of these appointments, it's difficult with my other son and he's in school and we have to balance that. So nothing can be during the evenings. Trying to adjust to the needs of both kids has been really hard."*
- *"We've had to make sacrifices to our social lives too. We don't get to go out as much, or she has an older sister and we may not be able to do a lot of things like go on vacation because she's sick. So it does impact family's social needs."*
- *"It's always hard on our other child. Our youngest is the center of attention. We have nurses in the house all the time, and if he's sick, mom and dad go to the hospital or he's got a doctor's appointment. We haven't taken family vacations or things like that. You're trying to make sure that your family isn't just defined by this condition. And you want them to interact like brothers. But he can't rough-house with his brother. So it's kind of hard. We're always telling our oldest "don't do that" or "don't touch him like that" and it's hard when you want them to be friends."*

A few families mentioned other children in the family were affected in terms of placement (or not) in daycare or school settings because of condition specific issues.

"She is immune compromised in the sense that if she gets sick, her body cannot fight that well. So something another kid might get over in 24 hours takes her 2 weeks. So we stopped putting my older daughter in daycare because if she gets sick there and brings it home, that's going to affect my other daughter a lot more. That type of decision happens all the time. For example, there was a big cultural event that we usually go to and we didn't this year because we didn't want to expose her to germs."

Several families also discussed temporary and permanent relocation to be closer to treatment or specialty teams, with some families stressing the impact those decisions have on the rest of the family.

- *"Deciding where to have treatment was a big thing. We struggled because we have an older child, and a support system here, and it's more convenient to stay here. But after visiting the [academic health center], it was obvious this is where we need to be. I knew right away that it's where we needed to go."*

Just learning about how all of their specialists are familiar with this condition, and they had a program dedicated to it, it was a no brainer for me.”

- *“My daughter and I moved to another state for 6 months for treatment and my husband and son stayed behind so my son could have somewhat of a normal life and go to school.”*
- *“We moved mainly because my husband moved for a job. But when he was looking for a new job, something we considered was whether it was close to a large hospital. That way, my son would have treatment if needed.”*

A few families also discussed strain on marriage, mental health, as well as financial impact and burdens such as debt and bankruptcy.

- *“Me and my husband had different philosophies. He wanted to enjoy life and not really focus on too much of the medical stuff, so it was kind of on me to research, make appointments, and take her to appointments and that’s probably why our marriage broke down. I started using alcohol because I was so stressed, which was not a good thing. I got help for that. He convinced me to file for bankruptcy because of all the medical bills.”*
- *“Primary insurance picks up most of it, but we’re in a huge debt right now. We have copays and certain items that primary insurance doesn’t pay for, such as the supplement that she takes, horse therapy, swimming... Right now we’re putting it on a credit card and hoping Medicaid picks up soon.”*

A few families highlighted the decisions they made about whether or not to have future children or explore reproductive technology.

- *“We’ve talked with our geneticists and team about wanting more kids. One of the questions they ask is about future kids. Part of why we waited so long was fear of it. During our last appointment we asked to get confirmation from them that it’s okay. It looks like it’s more recessive, he’s our worst-case scenario and he’s doing so well so it’s encouraging.”*
- *“Our family made the decision. We would not allow this condition to proliferate. From our son’s diagnosis, we knew our daughter would need assistance to have children. So my husband and I decided to start a reproductive technology fund—so that if she wanted to have children, we would have finances to help her have a condition-free baby.”*
- *“We had planned on having a larger family and have chosen not to. When we discovered the genetic component and the chance of having another kid with this condition or a kid that would be a carrier, we looked into IVF and, given the difficulties we had getting pregnant and the cost of IVF, it was better for our family to focus on our son and his care as an only child. We grieved over this—the vision we had for our family has changed. We decided as a couple that that wasn’t in the cards for us.”*

Support

The support system described as most helpful by families was other families with children with the same diagnosis. Many families mentioned the day-to-day support they received and provided to other families.

- *“I think when it happened to us, only one other state had included the condition on newborn screening, so now I could talk to other mothers with a similar experience. I met one in-person and it was nice to connect with someone that went through the same thing. So now I try to do the same thing for other people.”*
- *“It helps a lot to have other parents or caregivers. My daughter is older now and we’ve been doing this for so much longer—I feel like I’m answering more questions of parents who are brand new, than me seeking answers. It’s not that I know it all, it’s just that I’ve been there.”*

Several families also discussed consulting with other families on identifying care teams and developing treatment plans.

In connecting with other families, two caveats were noted. First, several families across diagnoses, timing of onset, and method of condition identification mentioned that it is important to match families based on phenotype, severity of symptoms, or similar experiences to avoid negative interactions.

- *“I would caution you that there are some people whose children have the milder form of the disease and speak about what is or isn’t helpful. Their experiences aren’t as helpful and their situation is so different because their children are not getting it as babies.”*
- *“For me, having a child with late onset, there are many people living with late onset in the group, so you see pictures and learn that they have a full-time job and have kids and grandkids and a life outside of the infusions. That really calmed me down because the stuff on the internet about infantile onset says it’s fatal. So to really see that people are dealing with it was helpful.”*

Second, a few families with children with early onset symptoms noted that the timing of connecting newly diagnosed families to other families should be considered.

- *“It took me a while to find other families. I don’t think I was ready to talk to people. I was depressed. I wanted to enjoy my time with my baby, and it took me a little bit, and when she was stable, I thought maybe I could talk to other families. After I realized she’s stable, she’s doing ok, I started reaching out to other families.”*
- *“Our genetics counselor did give me some contact information for another parent who was a couple hours away and for the longest time I just wasn’t ready for that—to make that connection. Everything was so negative with her diagnosis and she was falling behind and I was afraid of hearing any more negatives from another parent, so for a long time I didn’t use that resource at all. Eventually I did reach out to that parent once, so I know that that is out there.”*

In addition to discussing appropriate connections, families also discussed the ways in which they connected with other families. Most commonly families connected through social media, such as Facebook groups, in order to learn about others’ experiences, educate themselves, and develop a support network. A few families also discussed connecting with other families through specialists, genetic counselors, or pharmaceutical companies. Many of these families mentioned that engaging in social media had been a positive experience for them.

- *“I would recommend joining one of the two Facebook groups. Prior to this I would have said: forget it, support groups are ridiculous—but for whatever reason this community is very kind. The people are really positive and just want to help people.”*
- *“I got a lot of hope from our group online—because, there, kids range in how they are doing—some kids are walking and talking and other kids are in wheelchairs, so you have somebody to compare with and someone who has gone through what you’re going through. It’s nice to see when you see kids doing real well. And you don’t feel so alone when you’re going through something horrible, and you have other people who you can reach out to.”*

However, a few families reported negative experiences on social media, while a few discussed mixed experiences.

- *“There are Facebook groups, Instagram groups, Twitter groups. I can get ahold of the people in those groups if I want, but sometimes it’s not even necessary because there are so many different messages and I want to avoid misinformation. I kind of have to filter out the fake news, which is hard to do.”*
- *“Social media can also be scary because you can see some of the worst kids. However, you get to know the families and it’s just nice to talk to people and know they’ve been through that. I’ve really got a lot out of the social media groups on Facebook.”*
- *“The Facebook page has been helpful for certain—for the most part parents can be good resources and you can share your experiences. I’m careful about what I say because our family is not typical so we’re scrutinized. I think it’s better to private message people with questions.”*

Other common support systems included societies or professional organizations, medical teams, and friends or family, who often provided support through fundraising and care provision.

“My mom especially helped me with my son—she stayed here to help with him and make sure he got to school and was taken care of—not that his dad wouldn’t have done this, but his dad also worked many hours to keep our house and pay our bills. They were very supportive in all of my decision making. Even aunts and uncles helped. It wasn’t easy for everyone either, but we all made it through together. My brother came to visit me when we sought care out of state, and my mom came with my other son to visit, which helped too. You need your family through this whole journey.”

A few families mentioned feeling like outsiders among families of children without developmental delays.

- *“I definitely think within our circle we get hall passes. Everyone knows that this is a lot of work to manage, it’s a lot of time, and is emotionally taxing. It’s overall supportive. People do look at him differently—the family takes more pictures of him and they’re more outward with their feelings toward him, which is great. Going outward of our circle, people definitely have a hard time talking to us about it. They don’t want to share if they’re having hard times because compared to us they think it seems insignificant—it’s done with positive intent but you can feel like you’re on the outside.”*
- *“It’s been isolating. I had a lot of close friends, but I have to be careful because I can’t take him out if anyone is sick. I don’t take him out in big crowds—I don’t usually leave him either. I’ve seen some of my friends, but for the most part, it’s been isolating. We’ve started seeing less and less of people.”*

- *“It’s hard to relate to normal moms—I call us hospital moms. I recently ran into an old friend whose daughter also has a rare condition. We went for a drink and discussed how hospital moms are a different thing. You go to the lowest of the low—except that I haven’t had to bury my child yet. You don’t get to be a normal mom anymore so it’s hard to relate to other moms who want to make sure their kid goes to the best preschool, while you just want your child to live. It changes how you prioritize.”*

Advocacy

The majority of caregivers identified themselves as strong advocates for their children and their care. Most commonly, they discussed advocating for consistency in care across providers as well as coordinating messages and follow up across providers.

- *“I’ve been very involved with everything, in the medical side anyways. So I feel like I’ve been more involved than a lot of people. I’ve had issues, due to medication errors with his infusion. They weren’t doing the math correctly, they started it twice as fast, which puts him at risk for reaction. And that happened 4 times, so I had to contact the patient advocacy board and at that point didn’t feel my kid was safe, didn’t feel comfortable and was on my toes, and I switched at that point.”*
- *“I do find that I basically advocate for consistency. The nurses know what they’re doing but they all do it differently and some ways work better for my daughter than other ways so I find myself saying “the other nurse did this way, and that seemed to work okay—are you willing to do that too?” So not correcting them, but making adjustments. We also don’t see the doctor every time she gets her infusion so I find myself calling and emailing the doctors to ask questions and follow-up. I contact them all the time, usually via email, which is fantastic.”*
- *“Also, in terms of primary care, they don’t seem to know or want to know about the disability. It’s a big place where I have to be an advocate—if I’m not on it, nothing would get done. The doctors don’t communicate with each other.”*

Some families noted having to advocate for accommodations for their children to have inclusive environments in their schools and social life.

“We had some problems with the orchestra teacher, and she was not as accommodating as she needed to be. She was going to make my daughter stand the entire class, which wasn’t going to work. She also wanted my daughter to share a music stand. I had to go above her head and called the administrative building to ask if they had another stand. The teacher was not going to let my daughter play a violin in a concert, because she missed some of the practices, so I got the principal and teacher involved and she played at the concert.”

Several families discussed either themselves or their child taking on larger advocacy roles in their community or nationally. The particular issues noted ranged from inclusion of conditions into NBS panels in their state to advocating for a particular treatment.

- *“We started raising money for awareness and to donate for research to get better treatment. We’ve also made donations for travel assistance for kids for clinical evaluation. I think it’s important for me to not focus on the bad, to make a difference. And what can I do to help. And newborn screening is one I’ve had*

to advocate for. I've reached out to the Department of Health, and asked them why we don't have it on newborn screening. They finally approved and it hasn't started yet. It was nice to be able to hear that."

- *"I do a lot of advocacy around newborn screening. I believe in its importance. I wanted to years ago — you want to get started but you don't know where to go. I sent emails to lots of people. The doctors invited or provided opportunities to play a role. Even with the bill to get it on the state newborn screening, I got involved to represent the family side. We've done some work in her schools even, especially her elementary years. Every kid thought she was different. We knew that this was important so that she had a good experience there. We have also done some work on treatment and some work with a company developing newborn screening tools, helped them out to get them FDA approval. Most of these opportunities were presented by her care team. They invited us to participate as patient and family."*
- *"She also plays a role in advocating for herself. She is very good at telling people about what she has. She is not shy about educating people. She is going to be good at that."*

A majority of parents expressed feeling empowered about their role in advocating for their child or children's condition. While many also said it was a big role to play, and that they do sometimes feel burdened by it, they also said they felt empowered by taking on issues to support the health of their child or other children who have the same condition.

- *"My son is the catalyst. We've saved dozens of lives, and it will never be good enough for me. It will only be good enough if he was by my side. Tragedy is what spurs people into action. The bottom line is that my son is dead, and I don't want anyone to experience what we had to go through and I will do anything I can do to change that."*
- *"I enjoy it. I wish I could quit my job. I wish I could start a nursing company for this condition. Make sure the right tests are ordered, and their medical needs are being met. I know how important it is from the beginning. So many families who don't know about that. I wish I could do it every day."*
- *"I like this role a lot. I feel like I'm doing something good. I'm not one to be depressed. I want to make a difference. So if I can impact one life and help a kid get on a higher treatment then I feel like I've done something good."*
- *"I've spoken at a couple of events. I've spoken at a conference for geneticists, from a parents' perspective. Also at a genetic counselors' class at [the university]. Education is key and awareness is key and I will talk to anyone and anybody who will talk to me. Even a 7-year old because they could be the next genetic counselor. If anyone will listen to me, I will talk. It makes me feel good to know I'm helping spread awareness, to get people thinking about it and talking about it."*

Hopes

When asked to discuss hopes for their family's future, caregivers most commonly discussed two hopes—first, that their child would have the best quality of life possible for as long as possible; second, that more or alternative treatment options or cures would become available.

- *“My hopes are that medical developments can help my boys live a normal, healthy life. They can live a full life without being devastated or having too many disabilities. There’s going to be challenges for them, and I would hope they could embrace and accept them. I just hope to raise them as healthy and well-minded people, and that they go out there and try to make a difference in life.”*
- *“My hope is that all these new therapies and studies will do wonderful things and we’re going to get good information once studies are complete. I’m excited to hear what they are learning and finding. Hopefully there will be a cure and it’ll help her and other kids and adults. I hope that she’ll live a long good life, that she’ll surpass me and that she’ll be happy and healthy.”*
- *“Our hope is that we never have to see symptoms. But seeing all the research that’s happening, and hoping for better treatment—and we hope he continues to live doing normal life sort of things. I want to watch him grow up and have normal parent experiences. For example it was just tee ball at first. I remember crying after his first game, just to see him being able to run outside, and play and go to school and do normal childhood things – and someday getting married and having a job he loves and is talented at. Maybe he can help advocate or not be ashamed or fearful of his condition and take it head on and help other people.”*
- *“We’ve always wanted him to have the best quality of life that he can have, versus prolonging things for him. Before treatment failed, we were more about curative and now we’ve changed our perspective to being more about quality. Since we backed out of trying to “fix” him and started letting him be what he is, and just taking care of his needs as they arise, he’s been the healthiest and best person he can be. It was like when we got out of the way and just let God work in his life he’s been healthy. He’s not cured, he’s not healthy, but he has a great quality of life. It’s a different life than most adolescents but he enjoys his life and other people enjoy being around him. He has the best smile and you can’t not be around him and not be happy. I thought I knew love, but he has heightened my understanding of what it means to love someone. I don’t know that I would have learned that without him coming into my life. It’s been hard and there have been lots of times I wanted to give up but there’s been way more joy out of it. The lessons he’s taught us—you can’t get that knowledge anywhere else. When it’s time, it’ll be time. I look at my life more like that now—I enjoy the moment and when it’s time for me, it’ll be time.”*

A few families expressed hopes related to inclusion, such as the ability to educate other children or their community about their child’s condition.

“They look different, so educating children and others about people that are different would be helpful. A lot of us parents of children with disabilities hide them because we’re worried about what people think, but in reality we shouldn’t—we should put them out there, and just because they look and act different doesn’t mean that they are different from any other child—so pushing for awareness and helping make them feel confident in how they were born. I always tell my daughter that she’s great, beautiful, and smart and she views herself that way. She tells herself that she’s beautiful all the time. We’re all beautiful in our own ways—it’s not just what’s on the outside, so making them confident about themselves is very big—push your children and help them understand that there are people who look and are different, but we respect people no matter what and we respect them as we would want to be.”

Finally, a few families mentioned that they hope their child paves the way for children who are diagnosed in the future.

“My son will be participating in a [research study] shortly and I just want them to figure it out. I want to do whatever we can do to be a data point. We’re not going to be the miracle but I just want us and our family to be a data point and if there are 100,000 data points, then maybe they can piece it together to figure it out.”

Limitations

- As outlined on page 10, there was limited racial/ethnic and educational diversity amongst the families that participated and the findings do not represent the views and experiences of traditionally under-represented communities such as people of color or families that spoke another language.
- A majority of the families who had received a diagnosis through NBS had relatively young children. As more states add these conditions to their NBS panel and more children receive a diagnosis, their experience with health care providers may change.
- Nearly half of the families that participated in the project had children under school-age and could not provide input on experience with school and child care.

Recommendations

MAD developed the following recommendations based on the information received from the interviews with families. Though families identified many support gaps in their journeys with MPS I, Pompe disease, and X-ALD, or system gaps that were not specific to Minnesota, MAD’s recommendations below focus on gaps that MDH can play a role in supporting or public health systems can address. MDH should explore these possibilities in the broader context of organizational goals and other priorities.

- **Develop or compile verified and up-to-date, condition-specific information:** Families expressed the need to have early and easy access to accurate, condition-specific information and to be connected to the right resources and supports as soon as possible. MDH should develop and share information about the condition (including clinical guidelines, when available) and resources to support PCPs and families when children screen positive for MPS I, Pompe disease, or X-ALD and after the diagnosis is confirmed. Information should include an overview of the condition and connections to commonly used or needed resources (e.g., financial assistance, early intervention, insurance options such as Medical Assistance, mental health services, and respite care). In addition, MDH should provide information on social and medical supports, such as social media groups, professional societies, and networks of hospitals and clinics that specialize in caring for children with the condition.
- **Support health care providers in providing information to families:** Families discussed challenges with communication between them and PCPs, specialists, and genetic teams (i.e., lack of provider knowledge about the condition, insensitive and poor messaging, etc.). MDH should provide support to PCPs, specialists, and genetic teams in providing information and education to families once a child receives a positive NBS result. This could include support to improve parent-provider communication and to strengthen the role of providers in addressing the concerns of families who have received a positive NBS result or diagnosis.
- **Support families in care coordination:** Families also discussed that there is often not much support for care coordination and management (e.g., medical appointments, insurance navigation) as well as other

day-to-day needs as children age (e.g., education). Given these gaps, MDH should support capacity building efforts to strengthen systems to address care coordination needs of families. This could range from supporting families in navigating complicated systems in the short-term (e.g., families receive information that is comprehensive, up-to-date, and helpful), to long-term support to stakeholders to build their capacity to address care coordination needs.

- **Facilitate peer-to-peer and networking opportunities:** Families discussed direct connections with other families as one of the most important forms of support they received. MDH should consider facilitating connections with peers for support, matched when possible, on families' specific needs, phenotype, severity of symptoms, and similar experiences.
- **Convene and collaborate with stakeholders to address larger gaps:** Families discussed gaps in systems such as insurance, and care coordination within specific hospitals or clinics as major challenges in the management of care needs of their child. MDH should explore internal and external partnerships and convene stakeholder groups who can address these larger system gaps. This can include working with legislative partners or Minnesota Department of Human Services to close gaps in insurance systems.
- **Further evaluation of the NBS system:** Given that these conditions are relatively new to the NBS panel, MDH should consider further exploration of supports at a later stage of the program. This should include conversations with stakeholders such as PCPs, specialists, and families. MDH should improve community engagement efforts to reach traditionally under-represented families such as people of color and families that speak another language to gain an understanding of their needs and perspectives. It should also include an evaluation of the larger NBS system at MDH to assess the effectiveness of the program in addressing the needs of children identified with these conditions.

Appendix: Interview guide

Name of Interviewee:

Child(ren)'s condition:

Thank you for agreeing to speak with us today! We'd like to talk with you to understand the needs of caregivers, family members, and children with MPS I, Pompe disease and X-ALD.

Minnesota Department of Health (MDH) hopes to hear from families and caregivers of children with this condition as part of a Minnesota-based, nationwide project to understand information needs and experiences. MDH would like to hear your experience in accessing relevant information, managing care for your family member, and your input on resources and services needed to fill any gaps.

MDH has hired Management Analysis and Development (MAD), where I work, to contact families and ask these questions. MAD is a division within the State of Minnesota, and is a neutral third party that has helped the public sector for over 30 years.

The Minnesota Data Practices Act protects any information that you give MAD. MAD has a special section in state law that helps them keep your information private (Minnesota Statutes 13.64). MAD will make a report that sums up the interviews for MDH. MAD will leave out names or any other information that could identify specific people in our report. You do not have to take part in this interview- it's completely voluntary. Your feedback is valuable and will help us develop follow-up processes based, in part, on needs and experiences identified by the families. If there are any questions you do not want to answer, you are welcome to skip them.

Do you have any questions before we start?

1. Can you tell me about your child(ren)?

- How did you find out about your children's diagnosis? From whom? How old were they? (Examples; was diagnosis through newborn screening, family history, clinical signs/ symptoms, other?)
- What information was given at the time of diagnosis?
- Please describe for me how you learnt more about your child(ren)'s condition?
- Where did you go to find more information about the condition? (was it online? Organization?)
- Who did you want that additional information from?
- How did the information you needed change as your child got older?
- What did you wish you had known?

2. The next set of questions are about your child(ren)'s day-to-day needs and activities. Including health care, social services (early intervention, family home visiting, medical assistance, WIC, housing or transportation), and educational needs. Can you tell me about your experience navigating these different processes and systems? (*If child has late-onset; How has your experience been in these areas since your child(ren)'s diagnosis?)

- What has worked for you in attending to those needs? (*If child has late-onset; how has this diagnosis impacted your day-to-day activities? What about family life?)
 - Which day-to-day needs and responsibilities takes the most time/ effort?
 - What has been challenging?
- a) What, if any, challenges did you have in navigating the insurance system?
- What type of insurance do you have? (primary family insurance, secondary/ supplementary insurance)
 - Who has helped you navigate the insurance system? (Examples: pharma company, family/friends, organizations?)
 - Has insurance been able to cover all the services your child needs? If not, what was not covered?
 - Tell me about out-of-pocket expenses. Has it been burdensome? What has been some coping strategies? (Examples: changing jobs, participating in clinical trials?)
 - If insurance changed, tell me about what happened when insurance changed?
- b) Have you accessed any services or other forms of assistance? What did you find most useful? (Examples; early intervention, WIC, home visiting, medical assistance, transportation or housing?)
- What do you wish was different?
 - How did you learn about them?
 - If you did not access services, but needed them, why?
- c) How has your experience been with school or child care?
3. Now we're going to talk about some choices about your child's care as well as decisions about your family, you have had to make as a result of your child(ren)'s diagnosis. What were they? (Examples: having to move, switch jobs?)
- Who and what was helpful?
 - What made it more difficult?
 - Did you feel like you were in control? Why or why not?
 - If they have other children, "what has it been like for you in attending to the needs of your family, including other children? (Social needs, personal needs, and family needs such as vacations, dates, etc.)
4. What kind of support have you received as a caregiver that has been most valuable to you? What about support services for your child?
- How has social media, and broadly the internet helped you getting more support and information?

- Who was most helpful? How did you find out about this person/group?
 - Have you met other parents/caregivers of children with this condition? Have you found that to be helpful?
 - What has been your experience with families with children who don't have a medical condition/ or experience with the broader community? (Has it been isolating? Have they been accommodating?)
5. In your opinion, have you played any advocacy role since your child(ren)'s diagnosis? If so, can you tell me a little bit about your journey in becoming an advocate for your child(ren)?
- What support have you received in becoming an advocate for your child(ren)?
 - What are some key issues/ points you have had to advocate for?
 - What advice would you give to other parents of children with this condition on advocating for their child?
 - How do you feel about your role as an advocate? (Empowered? Burdened?)
 - *With older children- Has your child assumed or been in an advocacy role?
6. We would like to end by talking about your hopes. What are your hopes for your family's future?
Possible follow up:
- What about your child's future?
 - What do you want other people to know about children with this condition?
7. Is there anything else you'd like to share with us that we have not talked about?
8. Before we wrap up, I just have some quick questions about you and your family:
- a) Number of children affected with this condition (if not covered already)
 - b) Race/ethnicity of children
 - c) State of residence
 - d) Highest education level of caregiver
 - ☐ less than high school
 - ☐ high school diploma or GED
 - ☐ some college
 - ☐ 2-year college degree
 - ☐ 4-year college degree
 - ☐ post-graduate degree
 - ☐ other (specify)
 - e) Type of insurance (broad/standard, private/ public)

THANK YOU for your input. We appreciate that you took the time to speak with us. MAD will analyze the notes from these conversations and provide a summary report to MDH by the end of the summer.

The combined findings that will not identify individual families or caregivers will also be shared in the development of long-term follow up protocols and processes scheduled to take place this summer. Please don't hesitate to contact us or Elise Holmes at MDH if you have any further questions.