DEPARTMENT OF HEALTH

Considerations for Public Health Newborn Screening Follow-up for X-ALD, MPS I, and Pompe Disease

RECOMMENDATIONS BASED ON STAKEHOLDER COLLABORATION

October 2018

Considerations for Public Health Newborn Screening Follow-up Protocols for X-ALD, MPS I, and Pompe Disease: Recommendations Based on Stakeholder Collaboration

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This work is supported by the Association of Public Health Laboratories through the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under grant number # UG9MC30369 New Disorders Implementation Project for $4,000,000. This information or content and conclusions are those of the author and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the U.S. Government.

Contents

Background

In 2017, the Minnesota Department of Health (MDH) began newborn screening (NBS) for X-linked adrenoleukodystrophy (X-ALD), mucopolysaccharidosis type I (MPS I), and Pompe disease. Also in 2017, MDH received funding from the Association of Public Health Laboratories NewSTEPs program for a project aimed at maximizing public health (PH) NBS follow-up for these conditions. The project sought to answer the question, "What can MDH do to improve the health and well-being of individuals that screen positive for X-ALD, MPS I, or Pompe disease and their families?" MDH deployed a multi-pronged strategy to gather information from stakeholders and collaboratively create recommendations for future development of PH protocols and processes for PH NBS follow-up for X-ALD, MPS I, and Pompe disease. The project had four primary components:

Key Informant Interviews: MDH interviewed experts on X-ALD, MPS I, and Pompe disease including pediatric subspecialists, genetic counselors, and other health & community professionals. The interviews helped to better understand the conditions, existing medical resources, and inform further engagement (Summary in Appendix C).

Primary Care Provider Survey: MDH surveyed MN primary care providers (PCPs) to learn about their experiences and needs working with children/youth with inborn errors of metabolism (Summary in Appendix D).

Family Interviews: MDH also included the valued voices of families of children and youth across the U.S. with these conditions through telephone interviews. The goal was to better understand the needs and experiences of families, and what lessons learned could be incorporated into PH NBS follow-up (Summary in Appendix E).

Protocol Evaluation Workgroup: The project concluded by convening a workgroup (WG) of families, health care, community, and PH professionals. The group came together to make recommendations based on their expertise, and then evaluate a proposed plan for PH follow-up of children and youth with X-ALD, MPS I, and Pompe disease identified by NBS (Summary in Appendices A & B).

Abbreviations & Acronyms

BMT: Bone Marrow Transplant

CGC: Certified Genetic Counselor

ECLDS: Early Childhood Longitudinal Data System

ED: Emergency Department

EHDI: Early Hearing Detection & Intervention

ERT: Enzyme Replacement Therapy

HCP: Health Care Provider

HSCT: Hematopoietic Stem Cell Transplant

LPDR: Longitudinal Pediatric Data Resource

LTFU: Long-term Follow-up

MAD: Management Analysis and Development

MDH: Minnesota Department of Health

MN: Minnesota

MPS: Mucopolysaccharidosis

MRI: Magnetic Resonance Imaging

NBS: Newborn Screening

NICU: Neonatal Intensive Care Unit

OT: Occupational Therapy

PCP: Primary Care Provider

PH: Public Health

PT: Physical Therapy

ST: Speech Therapy

STFU: Short-term Follow-up

QI: Quality Improvement

WG: Workgroup

X-ALD: X-linked Adrenoleukodystrophy

Protocol Themes

The following section summarizes the four themes described by stakeholders as important for robust NBS PH follow-up for X-ALD, MPS I, and Pompe disease.

1. Supportive Follow-up after NBS Result Notification

The NBS result notification and follow-up to the point of (or ruled out) diagnosis (so called short-term follow-up (STFU)) of a child with a positive NBS result typically involves communication and resource support between the state NBS program staff, health care providers (HCPs), appropriate specialty care providers, and families.

What MDH heard from stakeholders:

- Too many and/or unproductive initial contacts for families
- Families need reliable, hopeful information up front (cannot wait until first comprehensive specialty visit)
  - Up-to-date, condition-specific information applicable to early identification through NBS
  - Crisis management counseling
  - Resource support (e.g., insurance, housing, transportation)
  - Options for specialty care
    - Some families do not have a strong opinion on where to refer, others want to be provided a choice
  - Concrete next steps, “action plan” for immediate future
- Coverage & timeliness for confirmatory & family testing
  - E.g., Confirmatory testing for Pompe disease needed to inform immediate treatment plan

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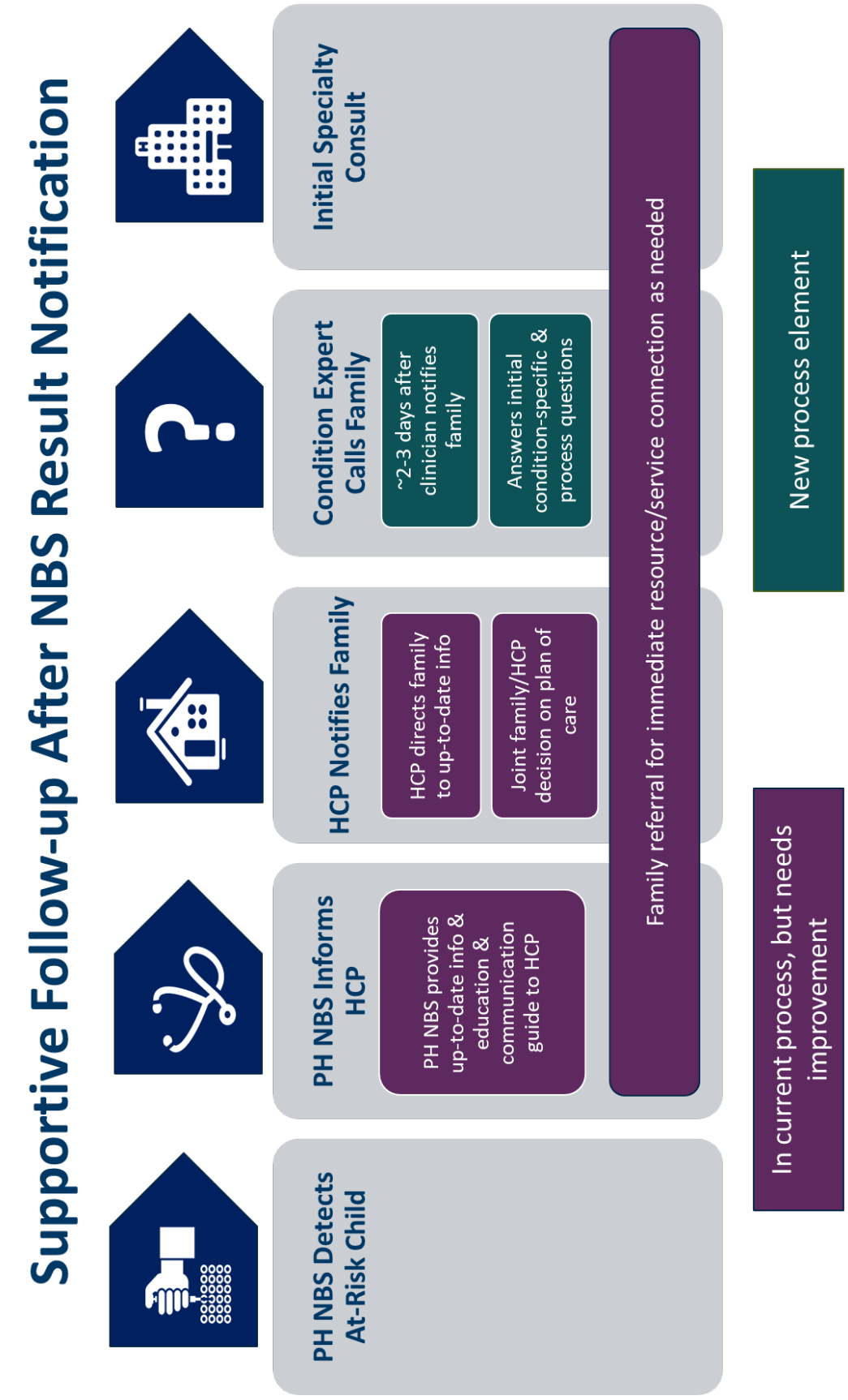
With that input, MDH developed the following proposed STFU process that improves upon the concept of family support after positive NBS result detection.


Supportive Follow-up After NBS Result Notification
PH NBS Detects At-Risk Child
PH NBS Informs HCP
PH NBS provides up-to-date info & education & communication guide to HCP
HCP Notifies Family
HCP directs family to up-to-date info
Joint family/HCP decision on plan of care
Condition Expert Calls Family
~2-3 days after clinician notifies family
Answers initial condition-specific & process questions
Initial Specialty Consult
Family referral for immediate resource/service connection as needed
In current process, but needs improvement
New process element

PUBLIC HEALTH NBS INFORMS HEALTH CARE PROVIDER

The specific timing of notification for NBS results of X-ALD, MPS I, and Pompe disease was discussed briefly by select clinical and family stakeholders. There was concern that notifying HCPs and families before a 2nd tier/molecular result would be inefficient and distressing. However, it was also imperative that results for certain presumptive positive lysosomal conditions be called out without delay to afford the opportunity to most efficiently and effectively establish a newborn in care and treatment. Thus, programs need to weigh the benefit of enhanced specificity of 2nd tier/molecular testing with the timeliness needs of the condition.

The state PH NBS program typically contacts the PCP, neonatal intensive care unit (NICU) staff, midwife, or lead specialty clinician in the event of a positive result for X-ALD, MPS I, or Pompe disease. At the time of notification, state PH provides just-in-time, condition-related information to HCP and recommended next steps. Many PH NBS programs provide the child's HCP with a fact sheet or other materials, detailing basic condition information, next steps, specialist contact information, as well as information to be disseminated to families. It was noted that while materials are provided to the clinic (most commonly via fax), they are not always reaching families, and in some cases, also not reaching the desired HCP. To improve this process, PH must conduct additional quality improvement (QI) to assure the resources are reaching intended audiences. Families also suggested that PH provide HCPs with communication guidelines to aid in sensitive communication of unexpected news. For state PH NBS programs that rely on a PCP/non-specialty HCP to communicate results to families, specialty clinicians voiced their willingness and preference for the PCP/non-specialty HCP to contact them before informing the family. Specialists noted that this contact allowed HCPs to receive more condition education as well as an opportunity to establish a basic set of next steps to inform the family.

HEALTH CARE PROVIDER NOTIFIES FAMILY

At the time of family notification, PH expects the HCP to provide basic condition information, next steps, and provide emotional support to the family. Further QI is necessary to ensure providers convey up-to-date and accurate information to families, in a way that is respectful of the family's learning style preferences. It is imperative that HCPs not simply state, "Don't Google it!", but explain that much of the information online regarding newer NBS conditions is likely to be outdated, and/or not applicable to early-identification through NBS, then offer sources of relevant, reliable information available online. Multiple families mentioned the importance of conveying "evidence-based hope" in the initial notification. Evidence-based hope was described as a balance between the reality of having a serious, rare condition with the notion that early-identification through NBS allows an opportunity for early treatment initiation and increases the potential for better outcomes. Another element in the process is to promote joint decision-making between the family and HCP. This may include presenting families with available options for specialty center referral(s) in order for them to make informed decisions about their child's subspecialty providers, rather than have the HCP decide unilaterally. Some stakeholders engaged in this project desired having this choice made known to them at the time of the initial referral process.

CONDITION EXPERT CALLS FAMILY

Providers and families suggested that the initial notification of the NBS result to the family does not often cover sufficient condition-specific content and/or provides little hope and guidance on next steps for their child. Thus, it was proposed to add an additional family contact to allow the family to ask more specific questions that were not covered in the initial notification as well as reinforce an "evidence-based hope" message (See Appendices A & B). Additionally, the shock of the initial notification is not always the most appropriate time to provide and retain education, so this contact can also serve to assess the understanding and retention of information. In the proposed process, a "condition expert" (i.e., a Certified Genetic Counselor (CGC) or other professional with expertise in the condition, either through PH or the family's associated specialty center) calls the family to check in within a few days of receiving their child's NBS result to assess condition knowledge, the understanding of the follow-up process/next steps, and answer further questions. While a CGC may not be able to answer all questions about specific clinical care, they would serve to act as a bridge between the notification and the initial specialty visit. This bridge is especially important in instances where the initial specialist visit is weeks after the NBS result notification. If PH CGCs intend to take on the role of calling families after the initial notification, it is important to identify whether some specialty teams already engage in this practice so as to avoic duplicate efforts. It may also be necessary to explore the types of information, counseling, and resources provided by specialty center calls and PH calls to ensure families receive consistent messaging and services. It was noted by multiple stakeholders that an initial family notification of the NBS result that conveys a lack of hope, sensitivity or accurate information cannot entirely be rectified through future interactions. However, it will provide an opportunity to not only address the aforementioned concerns, but also serve as an assessment point for QI improvement in HCPs making the initial notification to families.

INITIAL SPECIALTY VISIT

Several families indicated that the waiting period between receiving the positive NBS result and their initial specialty visit was particularly distressing. This was exacerbated by families having insufficient and outdated information regarding the health outcomes of the condition identified early through NBS. Regardless of the infant's apparent physical health status (e.g., observed signs or symptoms or apparent lack thereof) at the time of the NBS result notification, parents expressed significant need from an emotional standpoint to connect with the specialty center as soon as possible after being notified of the NBS result. Families indicated it would be beneficial if the specialty center made every effort to connect with the family soon after receiving the NBS result to provide a hopeful and realistic picture of their child's health outcomes and, using joint decision making, develop a care plan. It is unclear if this need can be met through communications with a "condition expert" as mentioned above, therefore additional collaboration is necessary to further assess and improve this experience for families. Some specialty providers in this project mentioned their willingness to leave designated openings in their clinic schedules to accommodate newborns identified by screening for X-ALD or MPS I within one week of NBS result notification.

REFERRAL FOR IMMEDIATE RESOURCE/SERVICE CONNECTION

Families may require additional emotional, financial, or other supports at any time after the initial notification of the result. The initial HCP, condition expert, PH staff, and/or subspecialist should be cognizant of the families' level of distress and the potential need for additional resources such as crisis counseling, financial counseling, parent-to-parent support, transportation to and/or temporary housing close to the specialty center, or the like. The above stakeholders can facilitate referrals to the most appropriate source. The potential referrals could include local PH, a medical social worker or financial counselor, mental health professionals, the Health Information Center (www.pacer.org/health/) etc.

2. Timely Connection with Accurate Information & Resources

While condition-specific information exists in medical literature and select websites, the most up-to-date and relevant materials do not always reach the appropriate audience at the desired time and are too scientifically complex for a lay audience. Additionally, the need for ongoing connection with accurate information and resources does not end at the time of the NBS result notification or initial diagnosis, but extends into childhood and adulthood. Stakeholders stressed the importance of having these materials and services in a format that is culturally, linguistically, and otherwise appropriate and accessible to their intended audiences. One important function, and a potential barrier to achieving this goal, is sustainability and the need for regular updating of information and resource materials to keep pace with evolving knowledge and discovery. Large-scale collaborative efforts with PH are likely required to ensure sufficient engagement of condition experts and other stakeholders to both develop and then assure ongoing maintenance of a resource repository. A feasible role for state PH NBS follow-up is the unique ability to collect and disseminate materials and resources for use across the NBS system due to their connections across many stakeholders and systems as well as non-profit, neutral status in the health system. Families also desire education and resources on topics such as navigating health systems, availability and eligibility of local services, and communicating with and supporting their other children and extended family.

What MDH heard from stakeholders:

- Need for anticipatory guidance or "road map" for X-ALD, MPS I, and Pompe disease
  - Not necessarily detailed standards of care (although that is also desired)
  - General guidance for financial planning such as the approximate number of specialty visits annually and/or by age.
- Need for ongoing condition information/education relevant to the child's development, phenotype, life stage etc.
- Materials and supports available in multiple/requested languages and at an appropriate reading level for a lay audience
- Need for assistance in insurance navigation & financial planning

    - Some organizations & hospitals already providing this, but families are unaware and/or capacity is insufficient
- Need for effective care coordination with clearly defined roles for families, providers etc.
- Educational supports vary and require significant communication between schools, families, and providers
- Desire for a summary of possible educational needs by condition at the state level
- Families want to connect with similar families
    - Same condition and phenotype identified as important by families and providers
    - Unfiltered social media has many pros/cons

"ROADMAP" FOR FAMILIES

Families expressed a strong desire for a general roadmap to better prepare them for what they may encounter in the near and distant future. The roadmap would not provide directive or specific information related to their child's health status, but instead, would guide the family in asking questions, planning next steps, and seeking resources (example Appendix F). Some suggested content from families and other stakeholders included:

- Steps to confirming the diagnosis
- Potential care treatment options and their general timeline
- Potential signs/symptoms, especially those requiring an emergency department (ED) visit
- Expected number of times families may need to travel to/maintain an extended stay by the specialty center
- Types of subspecialty providers that individuals with these conditions may encounter
- Questions families may wish to ask providers
- Types of supports for which families may be eligible
- How to connect with similar families

CONDITION & LIFE STAGE INFORMATION

Similar to the "road map" mentioned previously, families, HCPs, and school staff all expressed a desire for relevant information on the child's condition in the context of their life stage. This is especially pertinent in times of significant transition such as starting child care or school, undergoing prolonged hospitalization, or approaching adolescence. Families reported examples related to the experiences of childhood such as playing sports, getting braces, finding a car seat that fits correctly, or going on vacation. Information currently online often describes medical milestones, rather than life transitions, and focuses primarily on non-NBS identified children or adults. The PCP survey reported that important pieces of information after diagnosis applicable to primary care are the potential signs/symptoms, schedules for continued screening/monitoring, adjustments to the immunization schedule, and contact information for specialty providers. PCPs also noted that a centralized source or clearinghouse was important in accessing this information on NBS conditions (See Appendix D).

INFORMATION & RESOURCES FOR CHILDREN/YOUTH

Children and youth with X-ALD, MPS I, and Pompe disease and their siblings have their own unique information and support needs. Even basic information, such as "Why do I (my sibling) need to come to the clinic/hospital so often?" can provide reassurance. Child and Family Life Specialists are especially skilled at adapting complex information, both medically and emotionally, to help children and youth understand the purpose of their medical encounters, treatments, and procedures. In addition to information and preparation, mental health services may also be supportive for children and youth with these conditions and their siblings.

INFORMATION FOR EXTENDED FAMILY

Confirmation of a genetic condition for one child could have implications for the health of the immediate and the extended family. For example, with conditions such as X-ALD, the mother is often receiving new information regarding her (potentially symptomatic) carrier status, as well as the implications for her child(ren), siblings, and parents. As reported by stakeholders, the emotional burden is often high for families, especially if they take on the role of contacting the extended family with health information and attempting to answer questions or coordinate testing. It would be optimal to have a form letter or other communication given to parents/guardians of the affected newborn to distribute to the extended family to address basic elements of condition heritability, how to be tested, etc. Such a form should also include contact information for condition or other resource experts so that the newborn's family is not required to act as a coordinator.

RESOURCES & SERVICES

There are various resources and services which may or may not be applicable or accessible to families with a child identified with X-ALD, MPS I, or Pompe disease by NBS. Families engaged in this project were often unaware of existing grants, services, organizations, or other sources of support that could meet their needs. Additionally, not all families will be eligible for some supports. It is imperative to provide opportunities to allow families to explore the potential programs for which they may be eligible to meet their individual needs. Additionally, families may require assistance determining their eligibility and completing their applications/paperwork.

Multiple stakeholders mentioned that families above the eligibility cut-off for financial and other services have unmet needs. It is important to acknowledge other avenues of assistance such as grants or private sources of funding that have broader eligibility criteria. Community organizations, HCPs, and specialty centers should be aware of these sources in addition to means-tested services.

FAMILY-TO-FAMILY SUPPORT

There is a well-established need for family-to-family support in the existing literature and this need was evident in this project as well. Increasingly acknowledged as evidence-based practice by the Maternal and Child Health Bureau, family-to-family support has shown to improve families' ability to cope and function, which in turn can affect child health outcomes (Ireys, 2001; Singer, 1999). Although many families first meet other families online, social media also provides an unfiltered amalgam of potentially

misleading information. Families and specialty center staff emphasized that meeting a family with a child who has a similar phenotype and has undergone similar care/treatment is vital to a successful connection. This particular expressed need related to pairing families of children/youth with the same phenotype (e.g., early vs. late-onset). It was also expressed that, due to the rarity of conditions, as well as scheduling constraints (e.g., travel, work, the time spent by parents on coordination of care, finding child care) for families, it would be optimal to have options for telephone, online forums, webinars, or other remote opportunities when in-person meetings cannot always be arranged.

EDUCATION & TRAININGS

Families noted the importance of training HCPs how to best communicate unexpected information to families in a sensitive, hopeful, accurate, and effective way. Education and trainings can be supported by PH, as well as accomplished through community organizations, health systems, or other partners.

While paper and online materials are important, stakeholders, particularly families, also mentioned the importance of trainings or other educational sessions. Proposed training topics included tips on navigating health systems and working with insurance companies, particularly the prior authorization, referral, and appeals processes. Families also asked for in-person support when searching and applying for services. Financial planning/counseling was another service that families desired, particularly from someone with enough knowledge about their child's condition and current/anticipated treatments or interventions to accurately help estimate direct and indirect costs of care. Multiple families mentioned the benefits of attending conferences with both families and HCPs as a means of education and networking.

MENTAL HEALTH/COUNSELING

In addition to the potential need for crisis counseling around the time of NBS result notification, families also mentioned the importance of attending to mental health needs after diagnosis. For some families, informal emotional support through friends and family can be sufficient; however, multiple families brought up the importance of access to formal/professional mental health services for themselves and/or their family members. Caregivers (including fathers & grandparents) may benefit from support relating to the needs of their child, partner, or accompanying life circumstances. Families, HCPs, and specialty center staff also mentioned the needs of siblings who may experience difficulties coping with their sibling's diagnosis and how the family may have adjusted to meet the other child's health-related needs. Finally, the child/youth with the condition may also have unmet mental health needs that would best be addressed through formal mental health support. HCPs and specialty centers should be cognizant of these potential needs of families and equipped to make appropriate referrals.

RESEARCH OPPORTUNITIES

Multiple families mentioned their interest in pursuing research opportunities, but were unsure how to proceed. Families were dedicated to improving future care and treatment, and saw research as a means of accomplishing that for their own children and others with the same condition. While a resource provided by the U.S. National Library of Medicine (https://clinicaltrials.gov/) is publicly available to search online, it is important for families to talk with their HCP(s) and learn about the risks and the

potential benefits of research participation. Stakeholders identified specialty providers and/or condition-specific organizations as the most equipped to disseminate and discuss this information.

3. Relevant Data Collection, Analysis, & Use

The rare nature of NBS conditions creates a heightened need for stakeholders to collect, analyze, and report data for the purpose of maximizing the health of individuals identified with a condition through NBS currently and into the future. Although public and private researchers devote time and resources to these topics, it is also imperative that PH utilize these data at the population level. PH is in a unique position to evaluate and improve NBS systems as they have access to the full denominator of individuals identified with conditions through state PH NBS programs. For example, by collecting the approximate age of treatment initiated (e.g., Enzyme Replacement Therapy (ERT), Bone Marrow Transplant (BMT)/Hematopoietic Stem Cell Transplant (HSCT)), NBS systems can evaluate whether early identification through NBS resulted in early treatment and improved outcomes. In some state NBS programs, the process of monitoring and analysis after diagnosis is accomplished through a component of the PH NBS program known as PH NBS long-term follow-up (LTFU).

What MDH heard from stakeholders:

- Need for longitudinal information on X-ALD, MPS I, and Pompe disease
  - We are *all* still learning
  - Lack of well-established natural history after early identification through NBS
  - Just knowing diagnosis & mortality isn't enough
- Need to assess the impact of NBS and effect on the PH and healthcare system upon adding new conditions
- Need to identify and address disparities in outcomes at the population level

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GENERAL OUTCOME INDICATORS

While special projects and in-depth analysis of particular topics can provide additional and important insight, it is imperative that programs also have a consistent, overarching set of indicators that apply across all conditions identified by NBS. There are examples of core data elements already developed and applicable to this population through the Maternal Child Health Bureau Core Outcomes for CYSHN (https://ncbi.nih.gov/books/NBK132163/) as well as collaborative efforts such as the Public Health Longitudinal Pediatric Data Resource (PH LPDR) (https://nbstrn.org/research-tools/longitudinal-pediatric-data-resource). Consistency in data collection (i.e., common data elements) across all state NBS programs would allow for a more robust analysis of national NBS systems and outcomes. However, states have their own agency in determining the number, specificity, and type of data collected. Examples of general outcome indicators discussed by key informants in this project included:

- Morbidity and mortality
- Developmental and educational outcomes and supports

- Connection/retention in primary and specialty care
- Insurance status and access to care
- Need for and access to care coordination
- Connection to community, financial/insurance, other resources & supports
- Successful transition to adult care

It is apparent through the input of stakeholder engagement that solely clinical data is not sufficient to determine the full scope of social determinants of health or other factors affecting families of children/youth with a condition identified through NBS. Health and developmental status can provide more quantitative measures of child/youth health while qualitative family experience elements help to understand the various successes and challenges families encounter while pursuing a fulfilling, healthy quality of life. Stakeholders mentioned the importance of collecting data with the purpose of identifying disparities at the population level and identifying and addressing system gaps. PH seeks to not just collect information, but to use that information for the purpose of protecting and promoting health and ultimately, improving systems of care. NBS programs would benefit from routinely providing information back to stakeholders regarding NBS system successes and challenges. This communication should include narrative accounts as well as information on how data has impacted policies and procedures.

CONDITION SPECIFIC INDICATORS

In addition to general outcome indicators applicable across all NBS conditions, there are additional morbidity indicators that may be unique to specific NBS conditions. Especially relevant to X-ALD, MPS I, and Pompe disease are characteristics of symptom onset and timeliness to various treatments, particularly for late-onset phenotypes. With the assertion that NBS provides the opportunity to mitigate the severity of the condition by early monitoring and intervention, it is important to evaluate that hypothesis and identify the potential disparities in achieving those improved outcomes. Although symptom onset and timeliness to treatment may already be collected for other NBS conditions, determining what constitutes symptom onset (e.g. Loes score) or following a standard of care will require more detailed planning. Such planning necessitates additional engagement and should include careful consideration of existing data elements such as those developed through extensive collaborative efforts like the LPDR.

For X-ALD, MPS I, and Pompe disease, the added complex nature of late onset disease necessitates a longer PH follow-up and consideration of impact through a life course perspective. Specialists engaged in this project voiced concerns about families discontinuing monitoring or treatment protocols if signs/symptoms are not apparent. Additionally, the possibility for later-onset phenotypes makes it more difficult to identify false positive NBS results, potentially leading to inaccurate condition incidence reporting and inability to make QI adjustments to laboratory algorithms or screening methods.

One distinction of LTFU for X-ALD, is the lack of expected signs/symptoms of the condition for females affected by X-ALD in infancy or childhood. For this reason, additional engagement is required to determine how females identified with X-ALD through NBS may interact with PH NBS LTFU. An important factor identified by specialists and families was the need for females identified with X-ALD through NBS to receive genetic counseling at or before puberty. While particularly salient for this

population, this recommendation does not discount the importance of genetic counseling for males identified with X-ALD or individuals with other NBS conditions.

One of the most noticeable PH impacts of implementing X-ALD screening was the cascading identification of extended family members with this condition. Assessing the impact and effects of adding a new condition to the NBS panel should not be limited to the individual newborn, but include a broader family and PH population assessment. The identification of additional family members in the household, the extended family, or both will affect a family's resource or other needs. Testing and identification of additional family members also increases the referrals and/or consultation and coordination activities of specialty centers, possibly leading to additional systemic effects.

TIMELINE FOR PH FOLLOW-UP

While a consensus on a specific PH NBS LTFU timeline for ongoing surveillance of X-ALD, MPS I, and Pompe disease was not reached, stakeholders identified key considerations for future timeline development. Stakeholders in the workgroup mentioned the importance of PH NBS LTFU continuing through the transition to adulthood (See Appendices A & B). While a standardized timeline for all NBS conditions would provide consistency, it was also mentioned that the milestones (e.g., the median age of symptom onset) of a child with one condition may fall at a different time frame than a child with another condition and could be missed if data collection points were too infrequent. The potential for loss to follow-up, particularly given the complex nature of late-onset disease, was mentioned by stakeholders as another concern if points of data collection are widely spaced.

METHODS OF DATA CAPTURE

A variety of methods can be used to capture either individual or aggregate data on children/youth identified with X-ALD, MPS I, and Pompe disease through NBS. Abilities and methods of data collection for NBS follow-up often rely on state resources and legal parameters. For instance, a collection of child's clinical health status, both in terms of overall health, preventative care, and condition-specific factors might be accomplished through medical record abstraction, specialty center partnerships/reporting, payers' claims data, or otherwise.

State agencies outside of the NBS program also collect data that is relevant to families of a child/youth identified with a condition through NBS. For example, the MN Early Childhood Longitudinal Data System (ECLDS) (http://eclds.mn.gov/) provides cross-sectional information on child test scores, school absences, and use of educational accommodations. The MN Early Hearing Detection & Intervention (EHDI) (http://www.improveehdi.org/mn/) program leverages this data to assess child status and inform QI efforts. In addition, data linkages such as with vital records or immunizations can provide context to mortality and preventative care.

In addition to outcome indicators collected longitudinally, there are also benefits to special projects and data collection through other means such as surveys or focus groups. An important stakeholder perspective often underrepresented in NBS systems is that of the child/youth with the condition. It would be advantageous to find mechanisms to engage this population directly and appropriately.

4. Building Capacity and Improving Systems

The NBS system is broad and requires effective and regular communication and connection between stakeholders. PH can effectively use these partnerships to build capacity and provide technical assistance to partners serving children/youth with a condition identified by NBS and their families.

What MDH heard from stakeholders:

- PH has a unique role as convener
- Community organizations are supporting many needs expressed by stakeholders
  - Some families are unaware of these organizations/opportunities
- Need for ongoing, collaborative communication
  - Between HCP, state PH, schools, families, community organizations etc.

PH ROLE AS CONVENER

PH alone cannot accomplish all of the desired actions and fulfill the unmet needs of the systems serving children/youth identified with a condition through NBS and their families. Stakeholders across multiple communities (e.g., families, health care, education, community services) have important strengths to contribute. PH can play a role in bringing together stakeholders to facilitate conversations and pursue collaboration. Some proposed collaboration opportunities for PH NBS mentioned by stakeholders in this project included:

- Work alongside the Department of Education to inform the educational environment for children/youth with X-ALD, MPS I, and Pompe disease and lessen the communication burden on families and providers.
- Partner with the Department of Human Services to address challenges of families with X-ALD, MPS I, and Pompe disease such as housing, child care, financial needs etc.
- Share MDH experiences with NBS for X-ALD, MPS I, and Pompe disease nationally to inform or improve other NBS systems.

IDENTIFYING & ADDRESSING GAPS IN SYSTEMS

PH is able to identify gaps and inequities in systems through ongoing community engagement and data collection and analysis. Once gaps and/or disparities are identified, PH may be able to address needs directly through QI efforts, or build the capacity of other partners (e.g., community-based organizations) already working on or with expertise in the identified gap/disparity.

One significant unmet need mentioned in this project, as well as in published NBS-related literature, was care coordination. Although it is not likely within the capacity of most state PH NBS programs to provide direct care coordination and transition support to all individuals identified with a condition through NBS, PH can play a role in providing grants and technical assistance to clinics and organizations already

pursuing these activities. As an example, MDH currently facilitates a community of practice focused on care coordination for the pediatric population for care coordinators, care navigators, case managers, social workers, PH nurses and others who wish to network with and learn from their peers. Through these avenues, MDH can make known the specific coordination and transition challenges as expressed by stakeholders in the NBS system, and equip those within the system to close gaps.

Next Steps

Translation to PH NBS Follow-up State Protocol

The aforementioned themes and considerations for PH NBS follow-up for X-ALD, MPS I, and Pompe disease will require differing implementation strategies based on state resources, legislation, and other factors. Appendix G provides a basic outline for state-specific program implementation based on the Donabedian model of health care quality improvement using structures, processes, and outcomes as the framework (Donabedian, 1988).

Further Work Needed

This project provided a wealth of information for potential PH NBS follow-up improvement, some of which will require additional exploration. The following set of follow-up projects would complement and advance the work already accomplished.

QI PROJECT ON HCP NOTIFICATION & DISSEMINATION OF INFORMATION TO FAMILIES

State PH NBS programs and others have spent significant time creating, revising, and disseminating print and online information on NBS conditions. However, stakeholders reported that accurate information often does not always reach the intended audiences at the desired times and in the manner expected. It would be beneficial to conduct further work on effective dissemination of these materials and conveying messaging tactfully.

CREATION OF GENERAL ANTICIPATORY GUIDELINES, "ROADMAP" FOR CONDITIONS

While stakeholders expressed a desire for standardized clinical guidelines, a basic "roadmap" is also a widely-expressed need for families. As an example, MDH currently provides a similar resource to families through the MDH EHDI program (Appendix F). Families and educational staff also desired updated information pertaining to the child's life stage as they approached adulthood.

PH could co-develop basic "roadmaps" for X-ALD, MPS I, and Pompe disease in collaboration with stakeholders. While stakeholder input in this project was specific to these conditions, it is reasonable to explore the potential benefits of similar resources for other conditions on the Recommended Uniform Screening Panel. Such resources may not need to be state or program specific. Therefore, it would be appropriate to initiate longer-term projects and partnerships through regional or national stakeholders.

CREATION OF RESOURCES FOR CHILDREN/YOUTH WITH X-ALD, MPS I, AND POMPE DISEASE AND THEIR SIBLINGS

Another expressed need of stakeholders was developmentally-appropriate information for children/youth with X-ALD, MPS I, and Pompe disease and their siblings. As an example, MDH, in collaboration with stakeholders, previously created a children's book for congenital adrenal hyperplasia (http://www.health.state.mn.us/divs/cfh/topic/diseasesconds/content/document/pdf/cahgrow.pdf) that continues to be well-received. Similar to the life stage information mentioned above, a longer-term partnership will be necessary to create and update these materials.

STRATEGY TO ENGAGE & INCREASE MEDICAL WORKFORCE FOR X-ALD MPS I, AND POMPE DISEASE

Although not central to the protocol discussion, multiple stakeholders mentioned the need for an expanded workforce that serves children/youth with X-ALD, MPS I, and Pompe disease into the future. Positions such as medical geneticists, neurologists, CGCs, and internal medicine-pediatrics physicians committed to care coordination and successful transition were all specifically mentioned. PH often has relationships with universities and students that could be more effectively utilized to build capacity. Family stories are also an important source of inspiration to professionals considering a specialty career.

ASSESS TIMELINESS & COVERAGE OF CONFIRMATORY TESTING

Timely health insurance coverage for confirmatory testing, particularly molecular testing, was mentioned as a barrier for some families. HCPs reported that while insurance sometimes approved testing after appeal, some conditions, such as infantile-onset Pompe disease, require expedited results to ensure the most appropriate treatment is started in a timely manner. Further exploration of the scope of this issue and the potential solutions will require collaboration among a variety of stakeholders.

EXPLORE OPTIONS FOR CONDITION-SPECIFIC FAMILY-TO-FAMILY SUPPORT

Given the expressed need for condition-specific family-to-family support from families involved in this project, PH should consider their role in ensuring families of children/youth identified X-ALD, MPS I, or Pompe disease by NBS have an opportunity for connections with desired parent-to-parent and other family supports. PH can also consider direct and indirect opportunities to facilitate family attendance at conferences or other networking opportunities. It is imperative that PH acknowledge traditionally underrepresented groups in the family engagement process to enable effective, equitable support for all families.

CONNECT WORK FROM THIS PROJECT TO OTHER NBS CONDITIONS

While this project focused on the needs of stakeholders affected by X-ALD, MPS I, and Pompe disease, many of the findings likely apply to other NBS conditions. Further engagement and collaboration is needed with families, HCPs, and others to assess commonalities and differences between populations,

share information between “siloes” of PH work, and translate findings into effective solutions for other PH system gaps and meet the needs of other NBS conditions.

CONDITION-SPECIFIC DATA ELEMENTS

The MDH NBS LTFU program has spent significant efforts to develop a set of overarching core indicators and related outcome measures to be used over all NBS conditions. However, to understand the unique experiences and outcomes of children/youth with different NBS conditions, there is further work needed to develop small sets of measures for specific conditions and condition groups. Examples of measures would include access to and timeliness of receipt of best practice care and treatment, as well as monitoring more specific health outcomes. Development of condition-specific measures will take additional engagement from stakeholders including specialty providers and families, as well as further examining existing measures in sources such as the LPDR.

Sharing Results

This report and other companion documents are available through direct email with MDH. MDH will also report the findings of this project and the considerations for protocol development on their website. MDH, in partnership with collaborators, is also drafting a manuscript to be submitted to a peer-reviewed journal, and applying for opportunities to share results at national conferences. Findings will also be shared with advisors to the NBS system in MN as well as made available to other NBS state programs through APHL NewSTEPs.

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Appendix A: WG Morning Meeting Notes

Newborn screening (NBS) follow-up protocols workgroup

Meeting 1, July 13, 2018, 7 – 10 a.m.

Objectives

- Develop a clear description of a robust PH follow-up program in order to inform a plan for the PH role in NBS follow-up for X-linked adrenoleukodystrophy (X-ALD), Pompe disease, and mucopolysaccharidosis type I (MPS I)
- Build relationships with stakeholders for ongoing communication

In attendance

- 2 Parents of a child with a peroxisomal condition
- 2 Pharmacists
- 1 Primary care provider
- 1 School nurse consultant
- 1 Genetic counselor
- 1 BMT nurse coordinator
- 3 MDH NBS follow-up employees*
- 2 MAD consultants*

**Not active participants in idea generation/consensus*

Introductions and background

The group began with introductions, a review of the meeting agenda, and agreement on ground rules. Minnesota Department of Health (MDH) staff along with the Management Analysis and Development (MAD) consultant presented a brief refresher about the project to date on newborn screening follow-up for X-linked adrenoleukodystrophy (X-ALD), Pompe disease, and mucopolysaccharidosis type I (MPS I).

Defining features of a "robust follow-up program"

In teams of three, participants discussed the following question:

> Referring to the information provided in advance, as well as your own experience and knowledge, "What would you expect to see as the features of a robust follow-up program?"
> *Consider: data, data infrastructure, resources and costs (family, program, health systems),*

information needs, school and childcare, length of time/frequency of follow-up, care coordination, SDOH/inequities, evaluation/QI, initial follow-up and post-diagnosis follow-up

- What features would you see/experience as a family/caregiver?
- What features would you see/experience as a provider?

As a full group, they reviewed the small groups' responses and developed the following categories. Boldface titles represent the group's consensus about the category heading, and the bulleted items represent ideas contributed by individual small groups.

A. Respect for diversity (overarching all categories)

B. Evidence-based hope

- Portray hope at the time of notification; Realistic
- Clear, hopeful, anticipatory guidance

C. Care coordination plan clearly outlined

- Clear process: 1) connecting to care, 2) within care system, 3) community providers
- Comprehensive and cohesive care coordination
- Ongoing, collaborative communication (between HCP, specialists, school)
- Need standing referrals
- Coordination between agencies that serve families

D. Consistent, clear medical management recommendations

E. Equitable financial coverage

- A "path through the maze"
- PH provides genetic testing or guarantee of payment (payer of last resort) – in a timely way
- Minimize financial barriers – includes prior authorizations, other hoops

F. Support for all family members; Diverse, family-specific care

- Family-based (rather than individual-based) care
- Equitable, multicultural care

G. Help identifying resources

- Help identifying/securing resources (financial, occupational therapy (OT)/physical therapy (PT)/speech therapy (ST), transportation)
- Immediate grief counseling and crisis management
- Similar scenario family pairings
- Early access to up-to-date, reliable online resources
- Effective, updated resources
- Immediate financial counseling and support

- Life span financial counseling and support

H. Timely resources/information right away

- Minimize initial contacts
- Experts upfront
- Accurate online resources
- Helpful, knowledgeable primary care (talking points and resources for them)
- "Education for dummies"

Detailed description of a robust system

The group reviewed each of the categories above and collectively restated and added detail to each one. Along with this, they responded to the question, "What is the public health role in this area?"

A. Respect for diversity (overarching all categories)

B. Evidence-based hope

Groups discussed how, due to the rarity of these conditions, the information available does not accurately depict reality, and that hope in treatment is not often conveyed during the initial contact or call to parents. Groups discussed the need to convey that the condition is on the NBS panel because there is treatment (i.e., hope) available. Group also discussed whether PH can play a role in supporting HCPs to provide this information to patients and families. Example given of a family receiving notification of positive NBS result and only finding information online about how child would die by age four. Family continued to believe this for one month until the initial subspecialty consult when given more hopeful and accurate information. Group agreed this was unacceptable and needed prompt improvement.

C. Care coordination plan clearly outlined

Group discussed the need for customized and flexible care coordination support for families. They discussed effective and comprehensive care coordination support including child care, school, etc. Some examples discussed include the Sanford medical home model (www.sanfordhealth.org/medical-services/pediatric-medical-home), which includes medical, financial and education support to families as part of the model. Group discussed that PH can play a role in this including developing or refining a tool for families to reduce the burden of coordination.

D. Consistent, clear medical management recommendations

Group discussed the need for clear pathways for families in understanding the medical and treatment options available and recommended. This was especially relevant as it pertains to financial planning and employment decisions (e.g., Approximately how many subspecialty visits, procedures, hospital stays will we have in the first year? First 5 years?)

E. Equitable financial coverage – a path through the maze

Group recommended that families should be provided some anticipatory guidance on lab tests/imaging, appointments, and treatment that is condition-specific. These should include information about supplementary insurance. Group discussed the experience of hospital social workers in this area, but unfortunately, due to billing limitations and other factors, institutions lacked staffing capacity to provide this support routinely to all families. Participants discussed role of PH in guiding families to identify resources such as a list of experienced subspecialists, including setting up criteria to determine which specialty providers receive initial referrals, vetting credentials of resources, and helping make connections. They also discussed support for providing genetic testing or coverage of genetic testing as a "payer of last resort."

F. Support for all family members; Diverse, family specific care

Groups discussed the need to have condition and phenotype-specific information. They also discussed the importance of family-centered care that is specific to each family and their needs. This issue was discussed in-depth as it related to the unique needs of the mother of a newborn identified with X-ALD, as well as the need for extended family testing and support.

G. Help identifying resources

Group discussed PH role in identifying and relaying accurate information to families. Special attention should be given to ensure that families are connected to resources such as CGCs right away, and that information is given in a way that is not overwhelming. Other resources identified included financial, OT/PT/ST, transportation, grief counselling and crisis management, pairing families who have similar experiences, etc. Group elaborated on the importance of being proactive rather than reactive regarding family needs, and anticipating needs of new families based on previous experiences. Group discussed need for PH to coordinate with other agencies such as Department of Education and Department of Human Services, and Women, Infants, and Children Program (WIC) to identify gaps, fill needs, and ensure comprehensive identification of resources.

H. Timely resources/information right away

Group discussed the uncertainty and ambiguity experienced by families following the notification of a positive NBS. They expressed the need for connecting families with experts upfront and providing helpful talking points for HCPs when calling the family with the NBS result. Group did not have a strong opinion as to who the initial notification came from as long as they have the correct, basic information and some counseling ability/sensitivity (possibly a CGC). Group mentioned the importance of informing families upfront that information online will often be outdated for a child identified with NBS, and most importantly, to redirect families to accurate sources of information. Group discussed the role of PH in providing accurate information, such as a video, "education for dummies," and other materials that can be understood by families who do not have a medical background.

Appendix B: WG Afternoon Meeting Notes

Newborn screening (NBS) follow-up protocols workgroup

Meeting 2, July 13, 2018, 3 – 6 p.m.

Objectives

- Develop a clear description of a robust PH follow-up program in order to inform a plan for the PH role in NBS follow-up for X-linked adrenoleukodystrophy (X-ALD), Pompe disease, and mucopolysaccharidosis type I (MPS I)
- Build relationships with stakeholders for ongoing communication

In attendance

- 2 Parents of a child with a lysosomal storage condition
- 2 Pediatric medical geneticists
- 1 Pediatric neuropsychologist
- 1 Family support organization representative
- 1 Genetic counselor
- 3 MDH NBS follow-up employees*
- 2 MAD consultants*

**Not active participants in idea generation/consensus*

Introductions and background

The group began with introductions, a review of the meeting agenda, and agreement on ground rules. Minnesota Department of Health (MDH) staff along with the Management Analysis and Development (MAD) consultant presented a brief refresher about the project to date on newborn screening follow-up for X-linked adrenoleukodystrophy (X-ALD), Pompe disease, and mucopolysaccharidosis type I (MPS I).

Defining features of a "robust follow-up program"

In teams of three, participants discussed the following question:

> Referring to the information provided in advance, as well as your own experience and knowledge, "What would you expect to see as the features of a robust follow-up program?" *Consider: data, data infrastructure, resources and costs (family, program, health systems), information needs, school and childcare, length of time/frequency of follow-up, care coordination, SDOH/inequities, evaluation/QI, initial follow-up and post-diagnosis follow-up*

- What features would you see/experience as a family/caregiver?
- What features would you see/experience as a provider?

As a full group, they reviewed the small groups' responses and developed the following categories. Boldface titles represent the group's consensus about the category heading, and the bulleted items represent ideas contributed by individual small groups.

A. Education and training

- From the Department of Health to the Department of Education
- Better educational materials distributed right after notification; Across all stages of monitoring and treatment
  - Primary providers
  - Specialists
  - Families
  - Caregivers
- Class/training on how the medical system works for parents
- Central list of all subspecialties that you may need in your treatment along with subspecialist provider experience with the condition
- More parent control or info on where they are referred for their initial subspecialty consult (with information on insurance coverage/what is in-network)
- Good, reliable, and cutting-edge educational materials

B. Connections

- Social media to help make connections
- Formal and informal peer-to-peer
  - Providers
  - Parent-to-parent: access/information about organizations like Family Voices that provide a broad array of supports; families control access to genetic information
- Better peer-to-peer and condition-specific connections for families and providers, across state lines possibly (due to rare nature of conditions)
  - Website/family organization
  - Patient advocate groups

C. New knowledge discovery; Improvements in care

- Better natural history, LTFU tracking patients; Reporting genotype-phenotype correlation
- Making knowledge of current clinical trials (clinicaltrials.gov) more accessible to families
- Acknowledgement of what the situation is – reality check
  - "This is a rare disease."
  - What you know, what you don't know
- Diet
  - Acknowledge possibilities of complementary/alternative/exploratory therapies
  - If saying "no proof it works," also say, "no proof it doesn't work"
- Longitudinal pediatric data resource (LPDR) possible resource for NBS longitudinal data
- Curated (verified) information and resources shared

D. Empowerment and coordination

- Cross-facility care coordination (PCP role?)
- Point person to offer insurance/financial education and guidance
- Central/uniform accessible medical records providers and HCPs

E. Adequate long-term living supports

- Longer-term living support after an extensive procedure
- List of grants for families
- Early prep for transition

Detailed description of a robust system

The group reviewed each of the categories above and collectively restated and added detail to each one. Along with this they responded to the question, "What is the public health role in this area?"

A. Education and training

Group discussed the need for interagency coordination at a state level. Relying on individual providers to provide educational guidance to each school, for each child, was deemed inefficient. Group discussed the possibility of creating a summary sheet (good example for tuberous sclerosis (https://tsalliance.org/individuals-families/school-issues/)) for child care/school staff about the condition and possible educational supports. Group also recommended PH provide information about the conditions and care instructions as well as offer classes to help families navigate financial and medical processes. Educational materials provided to parents by medical providers or found online were deemed outdated and/or too medically complex. Group mentioned the educational materials provided by pharmaceutical companies as easy to understand, but were wary of information from a private company and saw a potential role for PH to provide "unbiased" information. Group also discussed the importance of not "sugar-coating" medical information for families that provides a false sense of hope – tactfully convey what is and isn't known. It is important to acknowledge it is a rare disease and we are *all* still learning.

B. Connections

Group discussed the need for improved peer-to-peer and condition-specific connection across state lines. This could include use of social media or family message boards facilitated by PH. Social media to help make connections including formal and informal peer-to-peer connections. An important point raised was that families need to understand the subspecialists they need to go to and they suggested PH can play a role in providing information about experienced subspecialists without endorsing specific institutions.

C. New knowledge discovery; Improvements in care

Group discussed the importance of documenting the natural/"unnatural" history of rare conditions through research and registries. Group discussed more in-depth study of genotype-phenotype correlation as primarily an academic or private research activity, but with possible implications or room for collaboration with PH, while protecting data privacy. Group also acknowledged family interest in participating in research and the possible role of PH or clinicians in making families aware of research opportunities.

D. Empowerment and coordination

Group discussed the need for families to have updated information about existing financial education services, and applying for Medicaid, TEFRA, SSI. Group mentioned it would be best if there was a single source or "point-person," either through PH, community organizations, or their medical institution, that could go through this information with families.

E. Adequate long-term living supports

Group discussed that now that more children are surviving longer, there is an aspect of long-term living support that needs to be provided to families. Group discussed the need for PH to play a role in providing information such as a list of grants or programs like Help Me Grow (http://helpmegrowmn.org/HMG/index.htm) and how parents can apply or be referred. Group discussed future needs for guardianship documentation or other preparation for transition as a priority for future PH work.

A helpful summary by one group member was regarding the key role of PH as facilitator: facilitating dissemination of information to families/providers/schools/etc; facilitating conversations between groups; and facilitating the collection of data/discovery.

Appendix C: Key Informant Interviews

Summary of Key Informant Interviews

DEVELOPMENT OF X-ALD, MPS I, AND POMPE DISEASE PROTOCOLS FOR PUBLIC HEALTH NBS FOLLOW-UP

Background

MDH met with 19 clinicians and other stakeholders with expertise in X-ALD, MPS I, and Pompe disease. The purpose of the interviews was to better understand clinical features of the conditions and approaches to care management, existing resources that support children and families with the conditions, and unmet condition-related needs. Expertise of interviewees ranged from neurology, genetics, and endocrinology, to child and family life, care coordination, dentistry, and more.

Primary Themes

Meeting notes were organized with a qualitative coding structure and analyzed for themes. The following section summarizes the ideas and experiences of interviewees, not the official recommendation or opinion of the Minnesota Department of Health.

Diagnosis, Monitoring, and Treatment

- There are significant benefits to receiving monitoring/care with subspecialty providers who have experience with these conditions. There are intricacies of a rare condition that other medical providers may not address that could be detrimental to the family (e.g., suggesting BMT/HSCT prematurely/when not indicated).
- Families of a child without signs or symptoms may experience medical fatigue and not adhere to an appointment schedule for monitoring if it is too frequent or burdensome.
- There are possible health disparities regarding access to diagnosis, treatment, and monitoring. It is important to capture that information and find solutions.
- Female individuals with X-ALD do not need the same extent of specialty clinical monitoring as boys, but heterozygosity for an *ABCD1* mutation is not inconsequential.
- All individuals (including females with X-ALD) will need genetic counseling around or prior to the onset of puberty to discuss reproductive options and other relevant topics.
- Children with MPS I may have complex dental needs.
    - The two most commonly asked dental questions for families of children with MPS I are: 1) Can my child receive orthodontic treatment? 2) Why have their teeth not erupted?
    - These are addressed on a case-by-case basis due to the wide variety of presentation in combination with other hereditary and treatment factors (e.g., age of BMT/HSCT, parents' dental record).

Data Collection and Use

- There is a need for quality, longitudinal data on individuals identified with X-ALD, MPS I, and Pompe disease through NBS that is also tied to their phenotype and therapies. It is unclear who is responsible for collecting, analyzing and reporting this data and with whom it should be shared.
- The quality of data is highly dependent on the person inputting the data and the standardization/ease of protocols. Funding and designated staff will be necessary to input information consistently and correctly.
- Many providers do not have a standardized system to determine if a patient is "lost" to follow-up (e.g., electronic workflow reminder).
- Data infrastructure exists (e.g., LPDR) as well as possible models for data systems (e.g., Congenital Adrenal Hyperplasia database, Children's Oncology Group, Cystic Fibrosis Foundation registry etc.) to collect longitudinal information.
- Medical professionals, PH, and private agencies lack communication and coordination regarding what data on NBS-identified children and youth they are collecting, why, and with whom it should be shared.
- It would be optimal to have data on extended family members identified with these conditions, not just the affected newborn, to measure the true impact of NBS screening on the entire population.
- The creation of a comprehensive PH database of NBS-identified individuals with X-ALD, MPS I, and Pompe disease would require legal/regulatory counsel to ensure proper data privacy and protection standards (i.e., need for consent or IRB approval).

Public health follow-up data elements identified as important:

Monitoring: age of symptom presentation; method of identification; cognitive, motor & other developmental functions; loss/regression of skills

Treatment: type of treatment; when treatment was initiated; access barriers (e.g., scheduling, distance, in-home care etc.)

Medical encounters: ED visits and unscheduled hospitalizations (what reason); well-child visits

X-ALD specific: presence/severity of adrenal insufficiency; cerebral symptoms; outcome data to at least 12yrs age for boys to understand access to BMT/HSCT; # family members identified (especially boys <12 years of age)

Pompe disease specific: echocardiogram & electrocardiogram results; liver function; antibody response to ERT; CRIM status; use/need for mobile or other supports

MPS I specific: echocardiogram & electrocardiogram results; vision/hearing symptoms; skeletal manifestations; surgeries indicated/underwent; changes in IQ; use/need for mobility or other supports

Health Care Providers' Relationship with Public Health

- Most HCPs desire increased communication from the NBS program. A specific topic, format, or frequency of communication was not identified.
- The divisions of roles and responsibilities between PH and HCPs (especially in follow-up data collection and patient resource support) was not clearly defined.

Co-management of Care with other Providers

- The roles and responsibilities of PCPs are incredibly variable regarding their capability and willingness to participate in diagnosis, monitoring, and treatment of these individuals.
- PCPs can manage acute illness and well-child care for most families.
- For X-ALD, multiple specialists stated that PCPs should be capable of ordering diagnostic VLCFA testing for the proband and their siblings. This allows a quicker turn-around time for the specialist to discuss results with the family upon initial consult.
- Most specialty providers stated that direct communication via phone is the most effective way to answer specific questions or discuss a treatment plan with other providers.
- Most providers share appointment notes with other providers as an attached letter in a patient's electronic health record.
- It is often unclear, to parents and other providers, which HCPs or other staff are responsible for providing care coordination.
- It can be helpful to encourage families to receive primary care from a family practice or internal medicine/pediatrics (Med-Peds) provider. This can allow for a more seamless transition into adulthood.

Insurance & Finances

- Insurance does not always cover monitoring at the desired frequency of the subspecialist. This is especially apparent for "asymptomatic" individuals regarding scheduled MRIs and neuropsychological/neurocognitive assessments.
- Although there are significant benefits to receiving care from a pediatric subspecialist who has experience with the child's condition, insurance does not always cover out-of-network care to see these providers (e.g., the family can get an MRI at a local hospital instead).
- There is not comprehensive data on the out-of-pocket direct and indirect costs (e.g., copays, meals, transportation, lodging, etc.) for families and how that affects their quality of life.
- Timeliness of insurance approval for enzyme replacement therapy (ERT) is a concern for lysosomal conditions such as Infantile Onset Pompe Disease (IOPD).
- Insurance coverage for BMT/HSCT (prepping for transplant, during hospitalization, and post-transplant) is often difficult to obtain.
- A national consensus on standards of care and routine monitoring would likely streamline insurance approval processes.

Observed Family Experiences

- It has been difficult to connect all possibly-affected family members with a CGC for testing and follow-up. Possible reasons for this include insurance barriers, overwhelmed family of affected newborn, a lack of understanding of heredity, a lack of communication within family, not knowing where to seek testing/counseling, and denial due to a lack of symptoms.
- It is important to educate families and PCPs regarding the severity of signs and symptoms and what actionable steps should be taken (e.g., go to urgent care, go to the ER, call the specialist etc.).

- Parents typically find meeting other families with a child with the same condition very helpful. However, it is important to make sure you match families strategically (e.g., similar phenotype).
- Parents have to do a great deal of explaining and re-explaining (to family, educational staff, PCP, etc.) the details of the child's condition. This is especially relevant when the child's condition is asymptomatic/pre-symptomatic/attenuated and may not be as outwardly apparent.
- Families, as well as child care providers/teachers, are integral in detecting symptom progression or loss/regression of skills (e.g., changes in handwriting).
- One of the most difficult things for families, as perceived by interviewees, is how often they hear, "We don't know what your child's condition will look like in 5 or 10 or 20 years." The unknown brings a great deal of anxiety.
- Cost, time off work, child care, and transportation are seen as most likely barriers to families making regular specialty and well-child medical appointments.
- There will likely be barriers to access & adherence in the time of transition of affected individuals into adolescence and adulthood. This was mentioned primarily in relation to adrenal insufficiency as well as ERT.
- Many female individuals with X-ALD desire to know their "carrier" status early in life (age 13 or before).
- Mothers of newborns identified with X-ALD are within the time period of exhibiting symptoms of adrenomyeloneuropathy (AMN). It is important to address the needs of the entire family, even when the initial focus is the newborn.

Education & Resources

- There is a need for condition-specific, neutral/un-branded educational materials in multiple languages and at an appropriate reading level.
- There is a need for condition-specific educational materials that can be understood by children (individuals with these conditions and their siblings). Specifically, something to help address the questions, "Why do I have to go to the doctor so much? What is happening in my body?"
- Parents are often overwhelmed by the quantity of information around the time of diagnosis.
- Paper materials are helpful for parents to look back on as a resource, even if they are not read immediately after receiving them.
- Fact sheets from MDH around the time of diagnosis are very valuable, especially with tangible next steps. It would be helpful to have a separate fact sheet regarding male siblings and how PCP should discuss that aspect with family and seek additional testing/monitoring.
- There are very few resources for women with X-ALD. Part of this is due to the lack of research on the condition presentation in women.

Appendix D: PCP Survey

Summary of PCP Survey

DEVELOPMENT OF X-ALD, MPS I, AND POMPE DISEASE PROTOCOLS FOR PUBLIC HEALTH NBS FOLLOW-UP

Background

MDH developed and launched a survey for PCPs in the State of Minnesota who have experience caring for children with Inborn Errors of Metabolism (IBEM). The purpose of the survey was to understand PCPs' real experiences seeking information, attending to acute and chronic needs, co-managing care with subspecialists, and interacting with PH. This document summarizes the ideas and experiences of survey respondents, not the official recommendation or opinion of MDH.

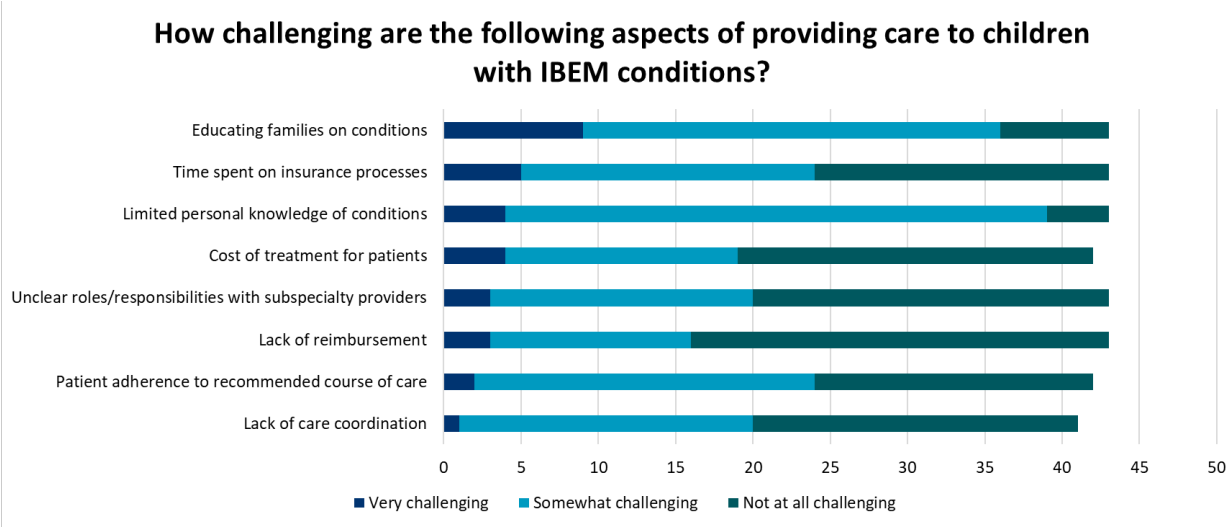
Characteristics	% Respondents (n=44)
Years of Practice	
Less than 10	30%
11 to 20	27%
Greater than 20	43%
Medical Specialty	
General Pediatrics	66%
Family Medicine	18%
Internal Medicine-Pediatrics	14%
Other	2%
Provider Type	
Physician	86%
Advanced Practice Registered Nurse	12%
Physician Assistant	2%
Location	
Metro	49%
Central	23%
Southeast	14%
Southwest	3%
Northwest	6%
Northeast	6%
West Central	0%
Condition Experience in Primary Care	
Amino acid disorders	25%
Fatty acid oxidation disorders	23%
Organic acid conditions	17%
Peroxisomal disorders	10%
Lysosomal storage disorders	9%
Other	16%

Survey Responses

Challenges Providing Care

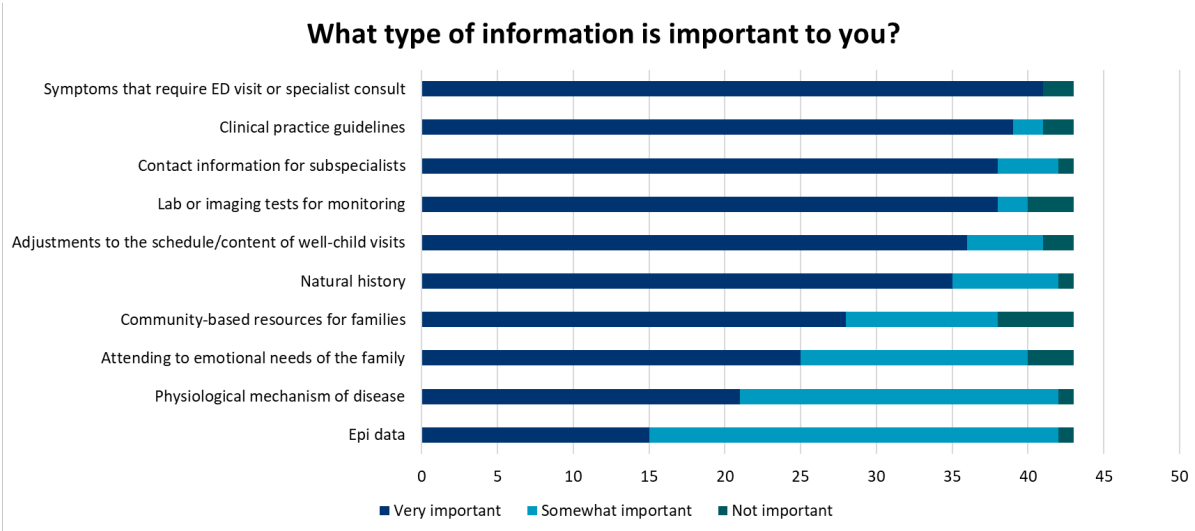
When asked about their level of confidence and the challenges providing care to children with IBEM conditions, most respondents listed condition-specific education and knowledge as a barrier. Additionally, the time spent on insurance processes and cost of treatment for patients also provided challenges.

"To make sure I don't mislead patients, I rely on specialists to provide patients with condition-specific information"



Information Needs

Related to the previously-stated challenges regarding condition-specific knowledge, respondents desired information on symptoms requiring an ED visit or subspecialist consult, clinical practice guidelines, and lab or imaging tests for monitoring. Most of this information was desired both around the time of diagnosis, as well as during ongoing condition management.

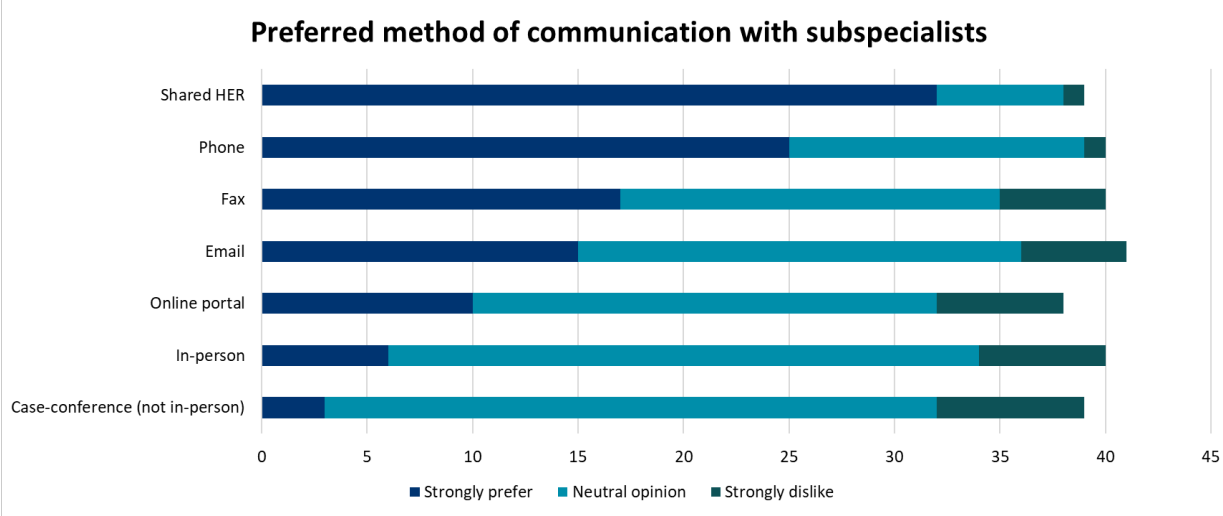


Respondents found direct communication with subspecialists the most useful. Some specifically noted centralized sources like UpToDate (https://www.uptodate.com/) as their primary source of reference.

"I mostly use UpToDate and then rely on specialists to fill in the gaps."

Co-management and Communication

Almost all (98%) of respondents in this survey "Strongly Agreed" or "Agreed" to having positive experiences co-managing care with subspecialists. The preferred methods of contact were shared electronic health records or phone. Case conferencing was listed as somewhat helpful, but nearly impossible to schedule with necessary parties. Half (50%) of respondents stated they "always" have a written management/treatment plan for their patients with an IBEM condition. However, 98% of respondents listed having a written management/treatment plan as "useful".



Role of Public Health

The last question on the survey was an open-ended question, "How can we at the Minnesota Department of Health help you better care for children/youth with IBEM conditions?" Respondents were appreciative of the fact sheets and just-in-time information provided by MDH at the time of positive newborn screen. The most common response was directing them to the "best sources of information." Some also listed needs for more county-specific resources.

"Keep families and affected person well-informed so they are best prepared to advocate for best practices themselves."

Appendix E: Family Interviews

Executive Summary of Family Interviews

PREPARED BY MANAGEMENT ANALYSIS & DEVELOPMENT

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 PH 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 disease, 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 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 the 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 their 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, CGC, or pharmaceutical company had also connected them.
- Other common support systems included societies or professional organizations, medical teams, friends, and family.
- Families discussed a variety of additional supports that would be helpful, including mental health support for siblings, postpartum mothers, and spouses or partners.
- A few families discussed condition-specific information and guidelines for care as a 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.

Appendix F: MN EHDI Roadmap Example

Hearing Loss

Identification

Audiologist: _____

_____

DATE OF DIAGNOSIS

After measuring how much your child can hear, we have determined the hearing loss in each ear to be the following:

Left Ear

- ☐ mild _____
- ☐ moderate _____
- ☐ severe _____
- ☐ profound _____

Right Ear

- ☐ mild _____
- ☐ moderate _____
- ☐ severe _____
- ☐ profound _____

Your audiologist will:

> Review the ***different choices in hearing technology*** and ways to communicate.

> ***Inform your child's primary care provider*** and the Minnesota Department of Health about your child's hearing loss.

> ***Connect your family to resources.***

The next step...

Your child needs to return to the audiologist/physician for further care.

Appointment Date: ___ /___ /___ Time: _____

Location: _____

_____

Your child has been identified as having a hearing loss.

While you may feel overwhelmed, with the right care, your child will develop good language and communication skills. Development can be delayed, however, without immediate action. So, use this brochure as your road map on the course you need to follow.

In this brochure you will learn the...

> ***resources and programs*** available to you

> ***doctors and professionals*** that you need to have involved in your child's care, and

> ***each step you need to take over the next year*** to ensure your child's development

Two symbols are used to help guide you:

Indicates an **action you need to perform**

Indicates a **resource available to help** your child

The first and most important step you can take is to ***start now!*** Children change so rapidly in their first months and years you just cannot wait.

To begin, follow the steps and utilize the programs and support available. Each program will help you to make appropriate decisions for your child and for your family, ensuring your child's good health and development.

Since every minute matters,

let's get started...

The FIRST step is to connect with

Programs to help your child

Your child's primary care provider and audiologist are your resources to connect you to the programs available. Programs are voluntary and, as parents, you have the option to decline. Both local and statewide programs work together to ensure the needs of your child are met. Programs include the following:

Minnesota Department of Health (MDH)

Minnesota Children & Youth with Special Health Needs

1-800-728-5420

www.health.state.mn.us/cyshn

Here's how MDH can help...

- A binder of information will be sent from MDH's Early Hearing Detection and Intervention (EHDI) Program that will tell you about resources available to you.

Your Local Public Health (LPH) Agency offers newborn and child follow-up services to help navigate and guide you through the process. MDH will link you with the agency in your area that provides the services you may need. When LPH calls you, they will be able to answer your questions about and connect you to:

- Financial Resources
- Insurance Resources
- Identifying Health Care Specialists
- Family-to-Family Networking
- Early Childhood Intervention
- Medical Condition Information
- Community Resources

Minnesota Hands & Voices

1-866-DHOHKID Voice Toll-free

Email: MNHV@lifetrack-mn.org

mnhandsandvoices.org

Contact: _____

Staffed by parents with children that are deaf and hard of hearing.

- A parent guide in your region will contact your family to provide information and support.

Early Childhood Special Education

1-866-693-GROW (4769)

www.helpmegrowmn.org

Coordinator: _____

Deaf & Hard of Hearing Teacher: _____

Most children with permanent hearing loss are eligible to receive early intervention and educational support services. Children ages 0-5 years are referred through Help Me Grow (HMG); children who are older can be connected through your elementary school. Services are provided through your local public school district at no cost to you, regardless of income or immigration status.

Minnesota Department of Human Services

Deaf and Hard of Hearing Services

651-431-5940

1-800-456-7589 (Toll-free)

https://mn.gov/dhs/people-we-serve/people-with-disabilities/services/deaf-hard-of-hearing/

Contact: _____

Promote equal access and opportunities for Minnesotans with hearing loss through effective communication, collaboration and direct services.

NEXT step is to coordinate with your child's

Doctors

Your audiologist will inform your child's primary doctor of the test results.

Doctor: _____

In the next few weeks, you need to discuss your child's hearing loss with your child's primary doctor.

Your doctor may connect you with specialists who work with young children in each of the following care areas:

ENT Doctor

(Ear, Nose and Throat Specialist)

Specializes in ear problems and will provide the medical clearance for hearing aids *(if chosen)*.

Doctor: _____

Location: _____

Phone: _____

Ophthalmologist

(Eye Specialist)

Cares for children's eyesight, which is very important for a child with hearing loss.

Doctor: _____

Location: _____

Phone: _____

Geneticist

(Heredity and Gene Specialist)

A geneticist can advise if your child's hearing loss is related to other health issues.

Doctor: _____

Location: _____

Phone: _____

Appendix G: Implementation Outline

NBS Follow-up Protocol Implementation

A STATE-SPECIFIC OUTLINE BASED ON THE DONABEDIAN MODEL

I. Purpose

This document outlines the protocol for [health department name] public health follow-up of individuals whose newborn screening (NBS) results shows borderline or presumptive positive results for [condition name] to:

[Examples of purpose including, but not limited to:

- Facilitate rapid and reliable diagnoses;
- Facilitate connection with evidence-based monitoring, treatment and care;
- Detect disparities in access to care (including enabling diagnosis) and therapy; and
- Promote best possible clinical outcomes and quality of life for identified individuals through service and resource connection.]

II. Structures

A. Legal Authority

NBS and follow-up for blood spot conditions in [state] is mandated by [list and link to state statutes].

[Explanation of statutes including, but not limited to:

- Activities covered by statute(s)
- Stakeholders to which statute(s) apply
- Authorization to collect medical information
- Data privacy, protection, and retention
- Legislation that applies to specific conditions]

B. Program Resources

1. Budget & Funding Stream(s)

[Description of financial management and funding sources for PH NBS follow-up]

2. Staffing

[Description of staff numbers (FTEs) and roles in PH NBS follow-up]

C. Data Infrastructure

1. Short-term Follow-up Case Management

[Description of use and capabilities of IT system for STFU]

2. Long-term Public Health Surveillance

[Description of use and capabilities of IT system for LTFU]

3. Other, Related Data Infrastructure

[Description of use and capabilities of IT adjacent to, supporting PH NBS follow-up]

III. Processes

A. MDH Short-term Follow-up Protocol

[Start-to-finish description/visual representation of actions taken by STFU staff including, but not limited to:

- Method, timing, and point of contact notified of positive NBS result (family, provider, other)
- Guidance to HCP to ensure family receives hopeful notification, accurate information on the condition, and relevant/timely resources
- Description of data collection, management, analysis, use, and reporting
- Resources and educational materials, dissemination strategy, and how materials are sustained/updated
- References to national standards for timeliness or other QI measures]

B. MDH Long-term Follow-up Protocol:

[Start-to-finish description/visual representation of actions taken by LTFU staff including, but not limited to:

- Timing and method of hand-off from STFU to LTFU (if PH programming is separate)
- Method, timing, and point of contact following up on child with NBS condition (family, provider, other)
- Any procedural changes based on sex, condition severity etc.
- Description of data collection, management, analysis, use, and reporting
- Resources and educational materials, dissemination strategy, and how materials are sustained/updated]

C. Stakeholder Engagement

[Description of activities taken by NBS STFU/LTFU programs to engage relevant stakeholder such as:

- Identification of stakeholders
- Methods of stakeholder engagement and communication
- Ongoing/periodic assessments of stakeholder-identified needs and resulting PH actions]

D. Program Evaluation & Quality Improvement

[Description of activities to evaluate and improve programming.]

1. NBS Program Logic Model

[A logic model is a useful tool in planning and evaluating the primary components and goals of a program. Identification of desired outputs, outcomes, and impact will be necessary to establish measures for ongoing evaluation. Assistance in the development of a logic model can be found below:

- CDC Guide to Logic Models: (https://cdc.gov/eval/tools/logic_models/index.html)
- MDH Quality Improvement Toolbox:
- (http://health.state.mn.us/divs/opi/qi/toolbox/logic.html)
- W.K. Kellogg Logic Model Development Guide: (https://naccho.org/uploads/downloadable-resources/Programs/Public-Health-Infrastructure/KelloggLogicModelGuide_161122_162808.pdf)]

2. NBS Program Evaluation Plan

[The evaluation plan is an important exercise as well as working document to guide continuous quality improvement. Evaluation plans can vary, but will often include components related to stakeholder engagement, data collection, analysis, and reporting, results reporting and more. Guides for creating an evaluation plan can be found below:

- CDC Evaluation Workbook: (https://cdc.gov/obesity/downloads/CDC-Evaluation-Workbook-508.pdf)
- MDH Evaluation Plan Template: (http://health.state.mn.us/divs/opi/pm/ran/docs/1307ran_evalplan.pdf)]

IV. Outcomes

TABLE A: INDICATORS FOR STFU

Evaluation question	Measure(s)	Source	Application	Frequency of collection

TABLE B: INDICATORS FOR LTFU

Evaluation question	Measure(s)	Source	Application	Frequency of collection

TABLE C: STATE NBS PROGRAM STFU/LTFU EVALUATION MEASURES

Evaluation Question	Measure(s)	Source	Application	Frequency of collection