Public Health Follow-up for X-ALD, MPS I, and Pompe Disease:
Results from Stakeholder Engagement

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Disclosures

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Why do we need public health follow-up for NBS?
Access to affordable treatment

Sufficient insurance coverage

Financial & employment stability

Able to navigate health systems

Education materials & resources on rare condition

Comorbidity and acute illness management

Parental support

Stable housing, food, transportation

Accommodations at school/child care

Adult provider familiar with condition for transition

Inclusion in community

Adherence to monitoring/treatment guidelines

Available specialty care in area (e.g. Peds genetics)

Able to navigate health systems
MN started screening for 3 new conditions in 2017

- X-linked Adrenoleukodystrophy (X-ALD), Mucopolysaccharidosis Type I (MPS I), & Pompe disease

Unique challenges

- Potential for later-onset features
- Limited ability to predict phenotype/severity of disease
- New pediatric specialists to engage
- X-linked inheritance
Stakeholder Engagement Strategy

1. Primary Care Provider Survey
2. Protocol Evaluation Workgroup
3. 1:1 Key Informant Interviews
4. Phone Interviews with Families
5. Share Results with Stakeholders
1:1 Key Informant Interviews

- Data capture and analysis
- Communication barriers
  - e.g., providers, public health, families
- Care coordination
- Educational materials & resources
PCP Survey: Results Summary

• Need Support:
  • Providing condition-specific education to families

• Confident:
  • Providing routine care and care coordination

• Most important information:
  • Signs/symptoms requiring ED visit or subspecialty consult
  • Clinical guidelines
  • Additional screening/labs/imaging needed
Family Interviews: Diagnosis & Information Seeking

• Diagnosis & Information Seeking
• Ongoing Supports
• Challenges
• Advocacy & Hopes

Learn more at 1:30pm today!
Protocol Evaluation Workgroup

- Evidence-based hope
  - Information, education, and training
- Coordination & planning of care
- Guidance through (and simplification of) systems
- Supports for all family members’ needs
- Meaningful connections
- New knowledge discovery
Considerations for Public Health Newborn Screening Follow-up Protocols for X-ALD, MPS I, and Pompe Disease:

Recommendations Based on Stakeholder Collaboration

**OUR PROJECT:**

In 2017, the Minnesota Department of Health (MDH) began **KIDDIE CARE** (Kinder, Ignite, Diagnose, Defend, Educate, and Engage) for X-linked Adrenoleukodystrophy (X-ALD), Metachromatic Leukodystrophy (MPS I), and Pompe disease. Also in 2017, we received funding from the **Association of Public Health Laboratories (APHL)**. With this grant, we wanted to know, "What can MDH do to improve the health and well-being of newborns that screen positive for X-ALD, MPS I, or Pompe disease and their families?" The activities of our project focus on listening to people who are directly and indirectly impacted by newborn screening for X-ALD, MPS I, and Pompe disease, families, medical professionals, public health, and more. After hearing about their experiences, we assembled a group to create a plan on how public health can best support newborns identified with these conditions and their families. The project has four main parts:

1. **Key Informant Interviews**: We began by interviewing experts on X-ALD, MPS I, and Pompe disease. These experts included pediatric subspecialists, genetic counselors, and other health and community professionals. The interviews helped us better understand the conditions, existing resources, and what could help inform our project.

2. **Primary Care Provider Survey**: Next, we created an online survey for primary care providers to learn about their experiences and needs, working with children who have similar conditions. These physicians, physician assistants, and advanced practice registered nurses are very important in managing the everyday health needs of children and families.

3. **Family Phone Interviews**: We also included the valuable voices of parents or guardians of children and youth across the United States with these conditions through telephone interviews. Our goal was to better understand the types and timing of resources families need, how and with whom they would like to communicate, and what lessons learned could be incorporated into public health follow-up.

4. **Protocol Evaluation Workshop**: After all of this information was collected, we brought together a group of parents, medical community, and public health professionals. The group made recommendations based on their expertise and then evaluated a proposed set of considerations for public health follow-up of children and youth with X-ALD, MPS I, and Pompe disease identified by newborn screening.

MDH is grateful for all the collaboration on this project. If you have specific questions about the project, or wish to provide comments, contact the Project Planner, Elise Holmes, at elise.holmes@state.mn.us.


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Further Work Needed

1. QI Project on PCP notification & dissemination of information to parents
2. Develop general anticipatory guidelines, “road map” for conditions
3. Develop resources for children/youth with X-ALD, MPS I, and Pompe disease and their siblings
4. Purposefully engage NBS families from underrepresented groups
5. Strategy to engage & increase medical workforce for X-ALD, MPS I, and Pompe
6. Assess timeliness & coverage of confirmatory testing
7. Explore options for condition-specific family-to-family support
8. Connect work from this project to other NBS conditions
Ongoing Work in Minnesota

• Exploring methods and outcomes for data capture

• Expand stakeholder engagement to other conditions

• Developing roadmaps
Our Team

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Thank you!

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