



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

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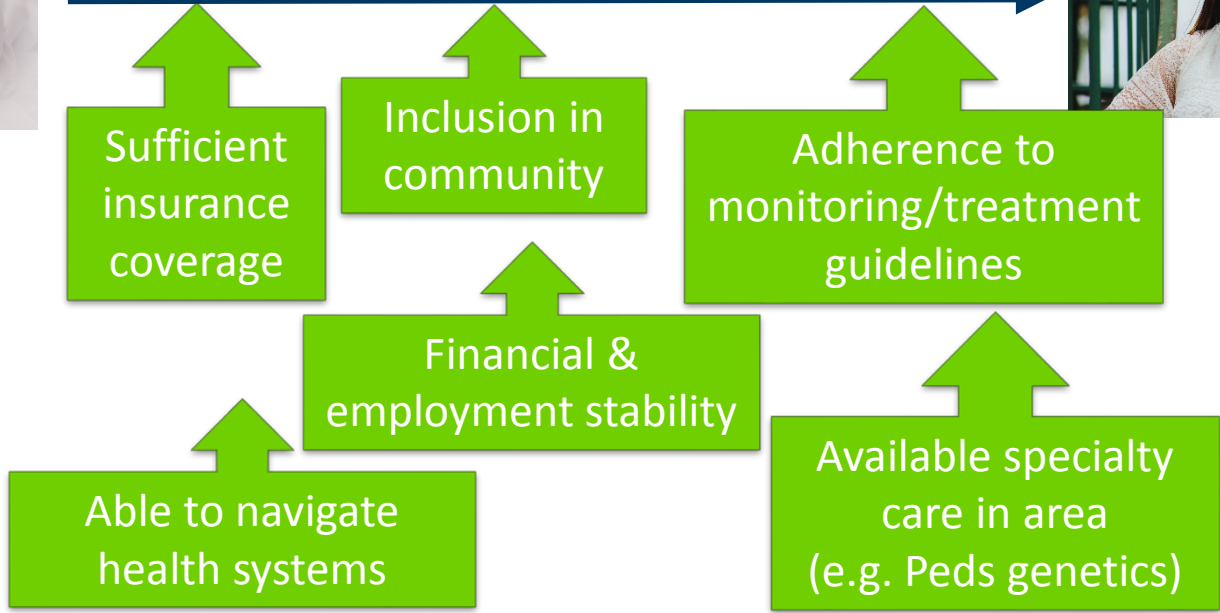
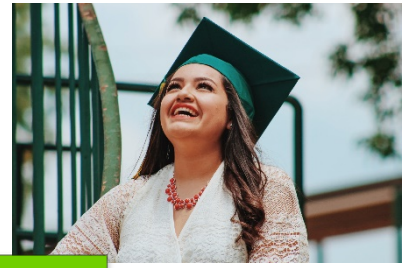
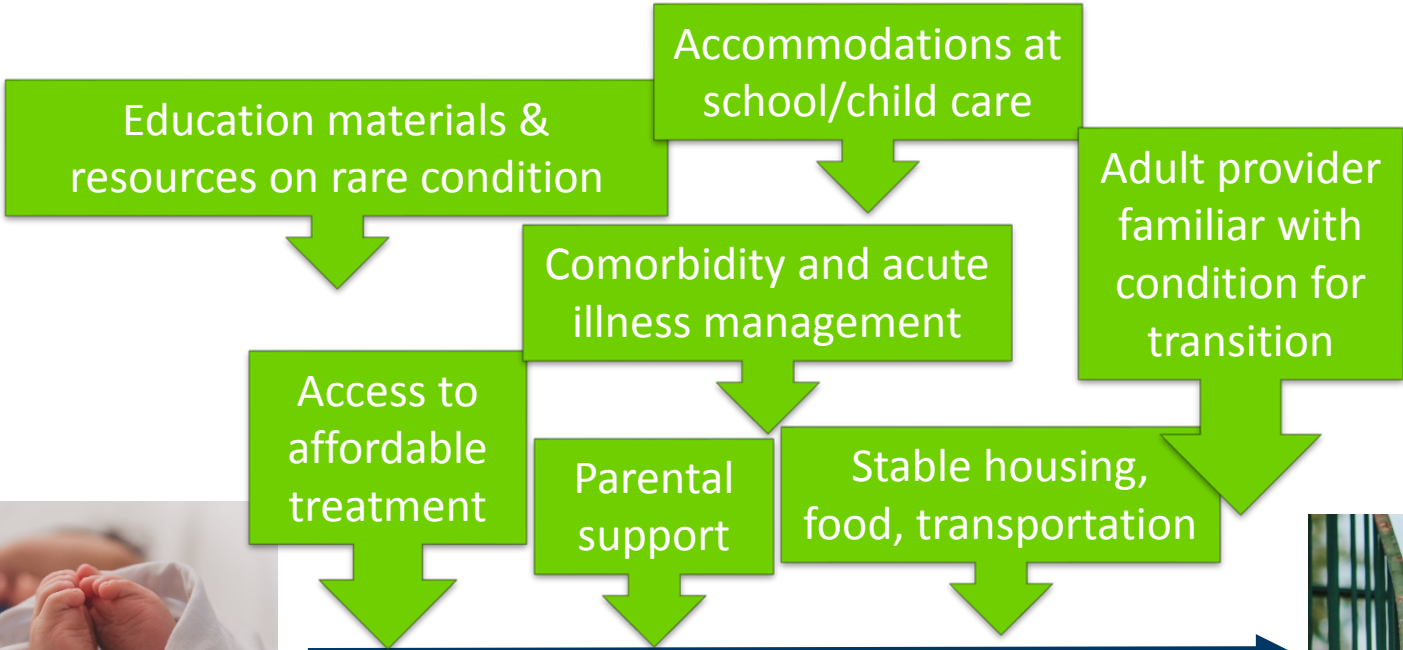


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***Why do we need public
health follow-up for NBS?***





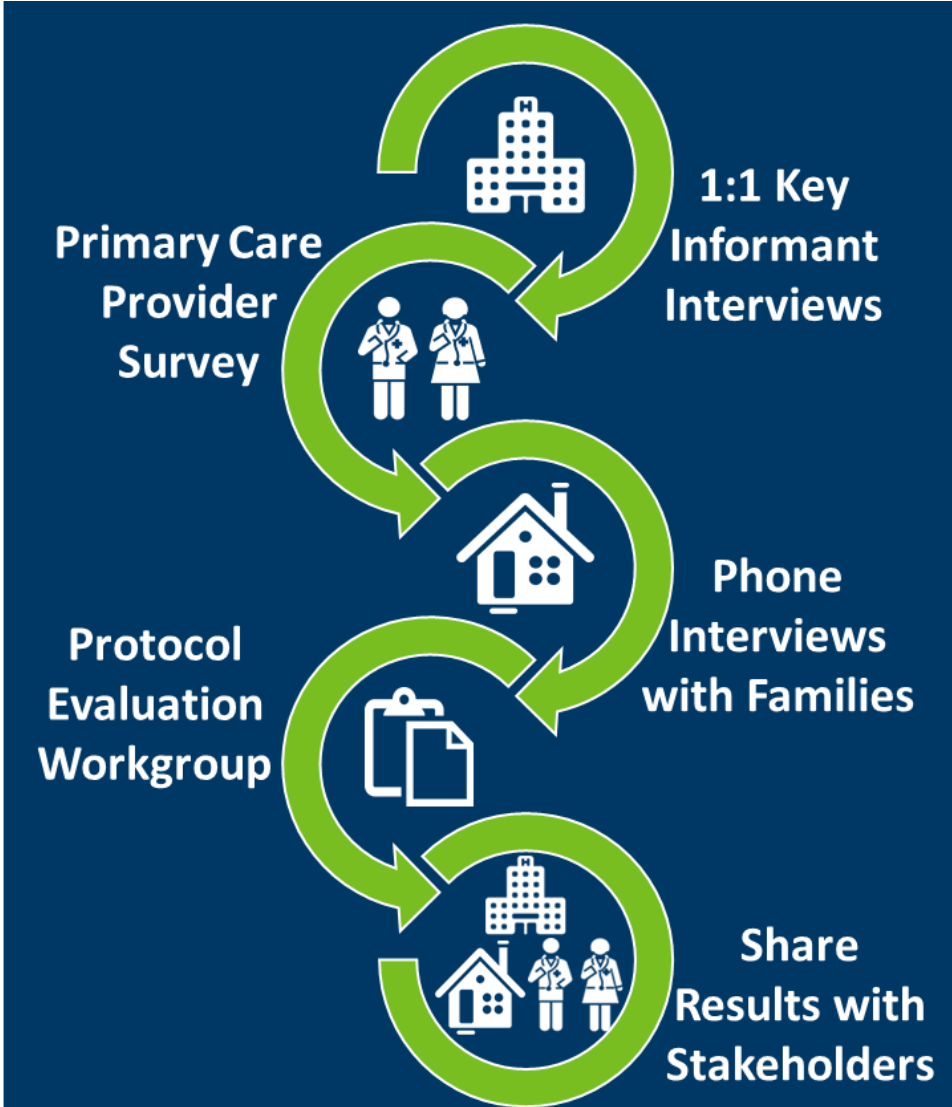
Public Health Role in NBS Follow-up*

- Understanding the impact of NBS
- Assistance for families in finding & accessing support for lifelong needs
- Developing, curating, & sharing information
- Convening people & groups affected by NBS
- Contributing to new and improved understanding of rare NBS conditions
- Identifying & addressing system gaps & disparities

New NBS Condition Follow-up

- 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
 - One of the first states to implement all 3 conditions

Stakeholder Engagement Strategy



1:1 Key Informant Interviews

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- 15 clinicians & ancillary providers
 - Genetics, Neuro, Endo, BMT, CGCs, Nurse coordinators, Social work, Child & family life etc.
- Better understand the clinical landscape
 - Diagnosis, monitoring, and treatment options
 - Barriers/challenges accessing & coordinating care

Primary Care Survey

PCP Survey: Results Summary (n=45)

- **Most challenging:**
 - Providing condition-specific education to families
- **Most 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

Participant Characteristics

Total Caregivers Interviewed (n)		24
Child diagnosis	Pompe disease	11
	MPS I	7
	X-ALD	6
Diagnosis method	Clinical presentation	15
	Newborn screening	9
Child age	Ranges 3 months – 20 years	
Child race/ethnicity	White/Caucasian	24
Location	14 states (all NCC Regional Genetics Networks)	
Caregiver education	Post-graduate	9
	4-year college	7
	2-year degree	3
	Some college	2
	High school or GED	2
	Unknown	1

Family Interviews: Diagnosis & Information Seeking

- Result notification & diagnostic workup
 - Provider knowledge
 - Insensitive messaging
- Condition Information
 - “Don’t Google it!”
 - Desire a roadmap for monitoring, treatment, prognosis



Family Interviews: Ongoing Supports

- Experienced & proactive providers
- Similarly-matched families
 - Timing varies
- In-home services
 - Early intervention
- Mental health services
 - All family members (i.e. siblings, fathers etc.)



Family Interviews: Challenges

- Insurance
- Debt or bankruptcy
- Quit job or reduce/add hours
- Care coordination
- Immune concerns
- Reproductive decisions
- Impact on siblings and marriage



Family Interviews: Advocacy & Hopes

- Empowered as an advocate
 - Consistency in care
 - Inclusion in school/social environments
 - Raising awareness
- Must become expert on condition
- Hope for:
 - Quality of life for child
 - Improved care/treatment options in future



Protocol Workgroup

Protocol Evaluation Workgroup

- Interdisciplinary group
 - Families
 - Clinicians (PCPs and specialty)
 - Health information & family support
 - Etc.
- Facilitated by Management, Analysis, & Development (MAD)
 - Accompanied by MDH staff for ?'s
- Evaluate draft protocols





Protocols for Public Health Follow-up of NBS

- Four primary themes
 - Supportive NBS result notification
 - Timely connection with accurate information & resources
 - Relevant data collection & analysis for discovery & continuous improvement
 - Building capacity & improving systems

- Processes & outcomes under each theme



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

Sharing Results

Thank you!

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