Families’ Experiences After Diagnosis:
An assessment of access and availability of care, resources, and support

Elise Holmes, MPP | Policy & Planning Analyst
Disclosures

This work is supported by APHL through HRSA/HHS under grant number #G9MC30369 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.
Overview of Project

• MDH began screening for the following in 2017:
  • Mucopolysaccharidosis type I (MPS I)
  • Pompe disease
  • X-linked adrenoleukodystrophy (X-ALD)

• **Project Purpose:** Better understanding the needs and experiences of families of children/youth to inform public health follow-up and related programming.
Caregiver Empowerment Framework

- Knowledge and understanding
- Decision-making
- Instrumentality (Navigating systems)
- Future-orientation
### Participant Characteristics

<table>
<thead>
<tr>
<th>Total Caregivers Interviewed (n)</th>
<th>24</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Child diagnosis</strong></td>
<td></td>
</tr>
<tr>
<td>Pompe disease</td>
<td>11</td>
</tr>
<tr>
<td>MPS I</td>
<td>7</td>
</tr>
<tr>
<td>X-ALD</td>
<td>6</td>
</tr>
<tr>
<td><strong>Identification method</strong></td>
<td></td>
</tr>
<tr>
<td>Clinical presentation</td>
<td>15</td>
</tr>
<tr>
<td>Newborn screening</td>
<td>9</td>
</tr>
<tr>
<td><strong>Child age</strong></td>
<td></td>
</tr>
<tr>
<td>Ranges 3 months – 20 years</td>
<td></td>
</tr>
<tr>
<td><strong>Child race/ethnicity</strong></td>
<td></td>
</tr>
<tr>
<td>White/Caucasian</td>
<td>24</td>
</tr>
<tr>
<td><strong>Location</strong></td>
<td></td>
</tr>
<tr>
<td>14 states (all NCC Regional Genetics Networks)</td>
<td></td>
</tr>
<tr>
<td><strong>Caregiver education</strong></td>
<td></td>
</tr>
<tr>
<td>Post-graduate</td>
<td>9</td>
</tr>
<tr>
<td>4-year college</td>
<td>7</td>
</tr>
<tr>
<td>2-year degree</td>
<td>3</td>
</tr>
<tr>
<td>Some college</td>
<td>2</td>
</tr>
<tr>
<td>High school or GED</td>
<td>2</td>
</tr>
<tr>
<td>Unknown</td>
<td>1</td>
</tr>
</tbody>
</table>
“The first thing our provider said was, don’t google it. But obviously no one is not going to google that. Yes, that was horrifying.”
Information Needs: Key Points

• Timely
• Consistent
• Action-oriented
• Realistic
• Hopeful
• Sensitive

“We just wanted answers, answers on his diagnosis, from someone, doesn’t matter who. We were in limbo for about a month, not knowing what was going on or what the next steps were.”
Challenges & Supports

“You’re trying to make sure that your family isn’t just defined by this condition.”
Challenges: Key Themes

- Care coordination
- Family relationships
- Work impact
- Traveling for care & extended stays
- Financial hardships

“Sometimes I feel like I’m on the phone more with doctors or therapists than I am playing with [my child].”
Challenges: Insurance

- Home services & transportation
- High deductibles and copays
- Appeals
- Understanding of coverage
- Specialty referrals
- Out-of-state care
- Jargon
Support: Key Themes

• Connecting with other families
• Primary & specialty care providers
• Effective care coordination
• School & home-based services
• Mental health supports

“It’s nice see when you see kids doing real well. And you don’t feel so alone when you’re going through something horrible, and you have other people who you can reach out to.”
Advocacy & Hopes

“I think it’s important for me to not focus on the bad, to make a difference.”
Advocacy: Key Themes

• Self-advocacy
• Consistency in care
• Raising funds
• Accommodations at school/child care
• Newborn Screening

“She also plays a role in advocating for herself. She is very good at telling people about what she has. She is not shy about educating people.”
Hopes: Key Themes

• Research (Better treatment options)

• Late-onset, remain symptom-free

• Healthy childhood/adulthood

“I want to do whatever we can do to be a data point. We’re not going to be the miracle but... if there are 100,000 data points, then maybe they can piece it together to figure it out.”
Recommendations
Recommendations

• Develop, compile, & disseminate verified and up-to-date, condition-specific information

• Facilitate family-to-family support

• Support families & systems in care coordination

• Convene and collaborate with stakeholders to address system gaps

• Further evaluation of the NBS system
Our Team

Elise Holmes
Project Manager

Kristi Bentler
LTFU Public Health Nurse

Nicole Brown
LTFU Supervisor

Tony Steyermark
Previous LTFU Supervisor

Amy Gavaglio
STFU Supervisor

Mariyam Naadhya
Consultant

Ashley Johnson
Consultant
Thank you!

elise.holmes@state.mn.us