Transitioning from pediatric to adult health care in lysosomal diseases: The MPS experience

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Background: Rare Diseases

- With improvements in diagnostic techniques and medical care, people with rare diseases are increasingly surviving into adulthood.1
- The need for a smooth transition from pediatric to adult health care is crucial to maintain the overall well-being of people with rare diseases.
- Health care transition is the purposeful, planned movement of people with chronic conditions from child-centered to adult-oriented health care systems.”2
- Transitioning care can be difficult because adult providers often have little experience caring for people with childhood-onset rare diseases.3
- Thus, people with rare diseases may lose access to important medical services and experience worse health outcomes as they age.3

MPS: A Lysosomal Disease with Notable Transition Challenges

- The mucopolysaccharidoses (MPS) are a group of 8 rare, lysosomal storage disorders characterized by progressive, multisystem disease.4
- With medical advances, people with MPS are living longer and need support transitioning from pediatric to adult health care.3
- People with MPS and their caregivers in France reported the process of transitioning to adult care was abrupt with inadequate support.5
- Parents of children with MPS IIIB in Turkey were reluctant to transition due to limited knowledge and pediatric-sized equipment among adult providers.6
- No research has been conducted on transitioning care in the US.

Research Study: Objectives

- With a team of physicians, researchers, and patient and family advocates, we are conducting a mixed methods study to:
  - Describe the experiences and needs of people with MPS and their caregivers during the transition to adult health care in the US.
  - Identify strategies to improve health care transitions for people with MPS and their caregivers.

Methods

- Online quantitative and qualitative survey
- Survey designed from literature review and 2 focus groups with members of MPS community
- Survey will be available mid-2024
- People with any MPS condition and their caregivers (parents, siblings, guardians, etc.) who are located in the US may participate.
- Recruiting participants through patient advocacy organizations, clinics, and conferences.

Focus Groups

- Conducted 2 focus groups to understand experiences and develop surveys.
  - 2 people with MPS and 3 caregivers participated.
  - Average age of people with MPS: 19 years old
  - Average age of caregivers: 46 years old
  - All participants were female.
- Range of MPS diagnoses:

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>MPS1</th>
<th>MPSII</th>
<th>MPSIII</th>
<th>MPSIVA</th>
<th>MPSIVB</th>
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<tbody>
<tr>
<td>Pediatric or Adult Care</td>
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<td>Location: OK, CA, DE, NC, NY</td>
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<td>3/5 transitioned to adult health care</td>
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Quotes from People with MPS

- “My transition timeline hasn’t been consistent among my different specialists. For instance, most of my care has been transitioned to the adult hospital but my pediatric cardiologist hasn’t given me a timeline to move out of his care.”

Quotes from Caregivers

- “I’m having surgery in two weeks and they have still not decided if I will have the procedure at the adult hospital or children’s hospital due to the equipment available for my body size. Yes, I am an adult but my body isn’t adult size.”

- “A few years ago, I was experiencing heart palpitations and went to my local emergency room. They didn’t know anything about MPS and it was really scary. I had to give them my pediatric doctor’s number to provide guidance. It prompted the conversation with my doctor on how we can manage emergency situations since adult physicians aren’t familiar with these disorders.”

Conclusion

- People with MPS and their caregivers experience fear and uncertainty around the transition process.
- We are currently in the process of distributing the survey, which can be found here:

  - Families need preparation, care coordination, emergency care planning, appropriate-sized medical equipment, and trained providers.
  - Results can guide providers, hospitals, and advocacy organizations to develop resources to support those with MPS and other rare conditions.
JM0 Suggest to use (MPS) not (MPSs).
There are currently 8 MPS disorders. I have attached the reference for the latest MPS disorder - MPS X (Verheyen S et al. J Med Genet (2022); 59:957-964) and have added the new # 4 ref
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