

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

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.

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."

"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."

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.⁵
- People with MPS and their caregivers in **France** reported the process of transitioning to adult care was abrupt with inadequate support.⁶
- Parents of children with MPS IIIB in **Turkey** were reluctant to transition due to limited knowledge and pediatric-sized equipment among adult providers.⁷
- No research has been conducted** on transitioning care in the US.

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.

Quotes from Caregivers

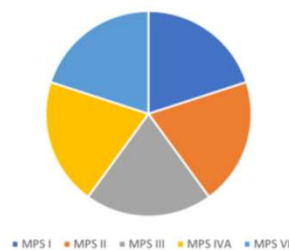
"All of my [child's] doctors are at the same hospital, but they have all told me a different thing about how long they will treat them. It would be nice to have some **uniformity** instead of wondering if this is the day we are going to walk in and they'll say it's the last day they will see us."

"As a mom, I want to get to know my child's future providers far in advance. I think our pediatric physicians should go through my child's caseload with their adult care physicians so that there's a **formal transfer** happening."

Focus Groups

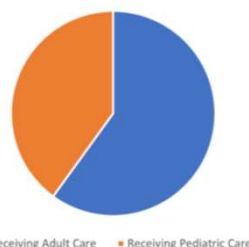
- Conducted **2** focus groups to understand experiences and develop surveys.
- 2** people with MPS and **3** caregivers participated.
- Average age of people with MPS: **19 years old**
- Average age of caregivers: **46 years old**
- All participants were **female**.
- Range of MPS diagnoses:**

Diagnosis



- Location:** OK, CA, DE, NC, NY
- 3/5** transitioned to adult health care

Pediatric or Adult Care



Need for Research

- Through personal experience, we have recognized that people with MPS in the US struggle with the transition to adult health care.
- Research is needed** to specifically identify the experiences of people with MPS in the US.
- Previous studies on other disorders in the US may not be generalizable to MPS, since:
 - MPS is rare and progressive
 - Spectrum of symptoms, including short-stature, airway obstruction, and behavioral challenges, can make transitioning care challenging, especially for surgery, anesthesia, and emergency care.
- It is also important to identify the experiences of **family caregivers, including siblings**.
- Poor transition-of-care process → caregivers assume more caregiving responsibilities → worse mental, physical health outcome.⁸

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.

JMO 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

Muenzer, Joseph,
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