

# Late-onset GM2 Patient Journey

## LATE-ONSET TAY SACHS/SANDHOFF (GM2)

- Rare, neurodegenerative lysosomal storage disorders
- Tay Sachs disease is caused by mutations in the HEXA gene which result in excessive accumulation of gangliosides (lipids) in the brain and nerve cells\*
- Sandhoff disease results from mutations in the HEXB gene\*

### OBJECTIVES

Understand the physical and emotional aspects of the patient journey, including symptom onset, diagnosis, and ongoing disease management.

### METHODS

- **18 surveys and interviews** with late-onset Tay Sachs/Sandhoff (GM2) patients (n=13) and caregivers (n=5) in the US
- All patients/caregivers 18+ years of age
- **5 interviews** with GM2 expert physicians in the US, UK, DE, and ES
- Research currently ongoing; poster summarizes interim findings

### CONCLUSIONS

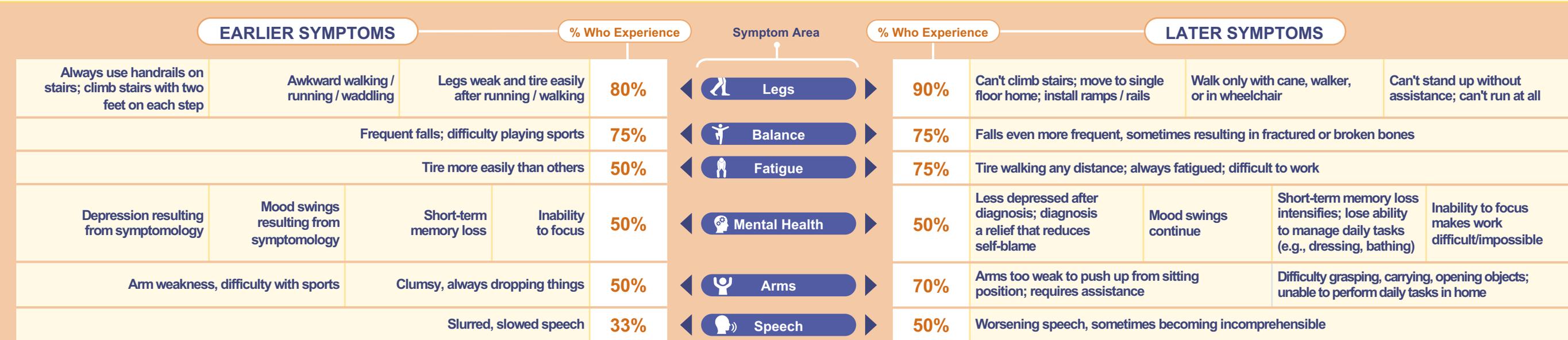
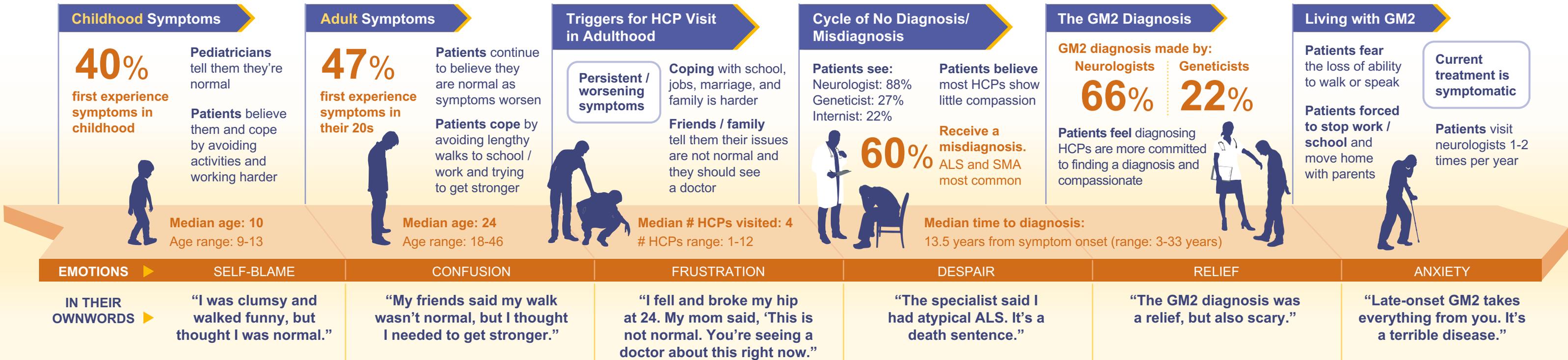
Symptoms typically emerge in childhood or young adulthood. Diagnosis often takes over a decade; common misdiagnoses include Amyotrophic Lateral Sclerosis (ALS) and Spinal Muscular Atrophy (SMA). Treatment is limited to symptomatic management and patients are often frustrated with healthcare providers. Patients lose the ability to perform activities of daily living and fear the complete loss of an independent life.

### AUTHORS

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## THE LATE-ONSET TAY SACHS/SANDHOFF (GM2) PATIENT JOURNEY



### EXPERT PHYSICIAN PERSPECTIVE

"Neurologists in the community would almost never diagnose GM2. It's difficult for me, and I specialize in rare conditions. It's a multi-systemic storage disorder and extremely rare. We need early referrals and genetic testing to diagnose tough patient cases like GM2."

– Physician, UK

\*Adapted from National Organization of Rare Disorders (<https://rarediseases.org/rare-diseases/tay-sachs-disease>)