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=15) 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 daily activities of daily living and fear the complete loss of an independent life.

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

**IN THEIR OWN WORDS**
- **Pediatricians** tell them they’re normal
- Patients believe them and cope by avoiding activities and working harder
- **Pediatricians** tell them they’re normal

**SYMPTOMS**
- **Childhood Symptoms**
  - Age range: 9-13
  - Medically recognized symptoms
  - Memory loss
  - Slurred, slowed speech
  - Weakness

**ADULT SYMPTOMS**
- Age range: 33 years
- Medically recognized symptoms
- Memory loss
- Slurred, slowed speech

**EATING**
- Medically recognized symptoms
- Memory loss
- Slurred, slowed speech

**CYCLE OF NO DIAGNOSIS/MISDIAGNOSIS**
- 66% of HCPs show little compassion
- 22% of HCPs feel diagnosing HCPs are more committed to finding a diagnosis and compassionate

**CYCLE OF NO DIAGNOSIS/MISDIAGNOSIS**
- Neurologists: 66%
- Geneticists: 22%

**MEDICAL VISITS**
- Neurologists: 66%
- Geneticists: 22%

**TRIGGERS FOR HCP VISIT**
- Persistent / worsening symptoms
- Coping with school, jobs, marriage, and family is harder
- Friends / family tell them their issues are not normal and they should see a doctor

**HOW LONG TO DIAGNOSIS**
- Median time to diagnosis: 13.5 years from symptom onset (range: 3-33 years)

**THE GM2 DIAGNOSIS**
- GM2 diagnosis made by:
  - Neurologists: 66%
  - Geneticists: 22%

**LATER SYMPTOMS**
- Can’t climb stairs; move to single floor home; install ramps / rails

**EARLIER SYMPTOMS**
- Can’t climb stairs; move to single floor home; install ramps / rails

**PERSPECTIVE**
- Neurologists in the community aren’t specialists in the disease
- “Tay Sachs is a terrible disease.”
- “Late-onset GM2 takes everything from you. It’s a terrible disease.”

**FUTURE OUTLOOK**
- Patients feel diagnosing HCPs are more committed to finding a diagnosis and compassionate
- Patients feel diagnosing HCPs are more committed to finding a diagnosis and compassionate
- “The specialist said I had atypical ALS. It’s a death sentence.”

**AUTHORS**
- Mariah C. LORSHIRE, MD
- Selena FRANGINE, MD
- Miro PETROVIC, MD
- John BURNS, PhD
- Rebecca GOULD
- Kathy NGUYEN
- Sanofi Genzyme, Cambridge, MA, USA
- Fulcrum Research Group, Waltham, MA, USA

**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