

# Natural History of Glycosphingolipid Storage Disorders and Glycoprotein Disorders

NCT00029965

---

Status	RECRUITING
Sponsor	National Human Genome Research Institute (NHGRI)
Enrollment	200 participants

## Key Eligibility Criteria

---

### Inclusion (1)

- Any individual with GM1 or GM2 gangliosidosis, sialidosis or galactosialidosis documented by enzyme deficiency or mutation analysis in a CLIA-approved laboratory will be eligible for the study.

### Exclusion (1)

- There will be no exclusion based on race, gender, or ethnicity; however particular ethnic groups may be overrepresented due to the frequency of the diseases in a specific population (e.g., Ashkenazi Jews in infantile and adult GM2 and Roma "travelers" in juvenile GM1). The majority of juvenile subjects will have severely impaired decision-making and even informed assent in older children may not be possible. Children with Morquio B disease are not expected to be cognitively impaired. The children with Morquio B ages 7-11 years will be asked to give verbal assent and ages 12-17 years will be asked to give written assent to the protocol. Some subjects who have reached the age of 18 may need to have legally authorized representative (usually their parents) sign consent on their behalf.

## Locations (1 total)

---

National Institutes of Health Clinical Center, Bethesda, Maryland, United States