

Rare Glycogen Storage Diseases Natural History Study

NCT06795152

Status RECRUITING
Sponsor Duke University
Enrollment 200 participants

Key Eligibility Criteria

Inclusion (9)

- Diagnosis of a rare GSD, including 0a, 0b, VII, X, XII, XIII, XV, PRKAG2 syndrome or Danon disease
 - Two variants in the gene associated with the specific GSD type (for autosomal recessive diseases)
 - One variant in the gene associated with the specific GSD type (for autosomal dominant or X-linked diseases)
 - Deficient enzyme activity in liver, muscle, skin fibroblast or other tissue
 - One variant in causative gene with evidence of disease, per a clinician
- ... and 4 more (see full listing online)

Exclusion (1)

- Unable to provide informed consent for participation for one's self or by legally authorized representative/legal guardian/parent

Locations (1 total)

Duke University, Durham, North Carolina, United States