Condition: Glycogen storage disease (Type I)

Inheritance:

Autosomal recessive

Genetic etiology:

Glycogen storage disease is a group of disorders associated with deficiency of enzymes involved in breakdown of glycogen to glucose. Type I is due to mutation either in *G6PC* or *SLC37A4*, which encode glucose-6-phosphatase and glucose-6-phosphate translocase, respectively.

Frequency:

Type I 1/100,000, with increased frequency in Ashkenazi Jewish population.

Clinical features:

Type I GSD is due to failure to break down glycogen, leading to hepatomegaly and fasting hypoglycemia. If untreated, there is growth retardation, neutropenia and impaired platelet function, and formation of xanthomas.

Management:

Frequent feeding with complex carbohydrates to maintain blood glucose, including continuous overnight infusion in infants; supportive care.

Genetic counseling:

Based on autosomal recessive inheritance; molecular genetic testing available.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.