Glycogen Storage Diseases

Type	Enzyme Defect	Affected Tissues (s)	Main Clinical features
Ia (Von Gierke Disease)	Glucose-6-phosphate	Liver, kidney	Classic form of GSD; hypoglycemia & metabolic acidosis 3-4 hours post metal; hepatomegaly with protuberant abdomen and lordosis; elevated triglycerides; doll facies and xanthomas; impaired platelet function/bleeding; elevated uric acid
Ib-c	Glucose-6-phosphate related transport	Liver	Ib- neutropenia and recurrent infections association with IBD Ic- Impaired insulin secretion
II (Pompe Disease)	Acid-α-glucosidase	Heart, muscle	Cardiorespiratory failure cardiomyopathy
III (Forbes Disease)	Debranching enzymes	Liver, muscle	Hepatomelagaly, fasting hypoglycemia, hyperlipidemia, progressive myopathy
IV (Andersen's Disease)	Branching enzymes(α-1,4-glucan 6 glucosyltransferase)	Liver	Hypoglycemia is rare; liver disease and cardiomyopathy are common