

Inherited glycogen storage diseases can affect tissue glycogen levels, fasting blood glucose levels, lipid metabolism and other pathways.

Glycogen Storage Diseases

Type	Defective Enzyme	Affected Organ	Glycogen	Clinical Features
I Von Gierke's Disease	Glucose 6-Phosphatase (deficient enzyme or translocase)	Liver & Kidney	Increased Normal Structure	Enlarged liver. Failure to thrive. Severe hypoglycemia, Ketosis, Hyperuricemia, Hyperlipidemia, Mental Retardation
II Pompe's Disease	1,4-Glucosidase (lysosomal)	All organs	Massive increase Normal structure	Cardiorespiratory failure. Death, usually before age 2
III Cori's Disease	Amylo-1,6-debranching	Muscle & Liver	Increased short outer	Like type I, but milder
IV Anderson's Disease	Branching Enzyme	Liver & Spleen	Normal amount; Long Branches	Progressive cirrhosis of liver. Liver failure causes death before age 2.
V McArdle's Disease	Phosphorylase	Muscle	Moderate amount; Normal exercise; Structure	Limited ability to perform strenuous muscle exercise; painful muscle cramps.
VI Hers' Disease	Phosphorylase	Liver	Increased amount	Like 1, but milder
VII	PFK-1	Muscle	Increased amount	Like V
VIII	Phosphorylase Kinase	Liver	Increased amount; Normal Structure	Mild liver enlargement; mild hypoglycemia