

Glucose-6-Phosphatase System Deficiency

Glycogen Storage Disease (von Gierke's Disease)

Deficiency in cytoplasmic glucose homeostasis of liver and kidney cells

Glycogen Storage Disease-1a

Causes 80% of GSD

Glucose-6-phosphatase-alpha subunit deficiency

Mutation of the two luminal loops and transmembrane helices

Glycogen Storage Disease-1b

Causes 20% of GSD

Glucose-6-phosphate transporter deficiency

Mutation of a highly conserved residue

Glucose-6-phosphatase-beta deficiency

Deficiency in glucose homeostasis of neutrophils leading to apoptosis

Glucose-6-phosphatase-beta subunit deficiency

and/or
Glucose-6-phosphate transporter deficiency

Mutation of the subunit and transporter