Glycogenosis/ Classification and basic characteristics of glycogenoses

Classification and basic characteristics various types of glycogenosis				
Type	**Name (*synonyms*)**	**Enzyme deficit**	**Affected organs**	**Clinical signs**
0		glycogenosyntase	liver	hypoglycemia, ketosis, early exitus (death)
Ia	Hepatorenal glycogenosis (*von Gierke*)	glukose–6–phosphatese	liver, kidneys, bowel	hepatomegalia, enlargment of kidneys, hypoglycemia on empty stomach, acidosis, hyperlipidemia, hyperuricemia, dysfunction of thrombocytes, later also creation of hepatic adenoma and glomerulosclerosis
Ib		microsomal membrane glucose–6–phosphate translokase	liver	like type Ia; plus recurrentneutropenia, bacterial infection
Ic		microsomal P–transporter	liver	like type Ia
II	Generalized glycogenosis (*Pompe disease*, *cardiac glycogenosis*)	lysosomal acid α–1,4–glucosidase	all organs	cardiomegalia, hepatomegalia (without hypoglycemia)
III	Limit dextrinosis (*Cori disease, Forbes disease*)	debranching enzyme (amylo–1→6–glukosidase and oligo–1→4–glucantransferase)	liver (muscles, heart), abnormal glycogen	hepatomegalia, normal lipids and ECG, mild hypoglycemia, elevated AST a ALT (cirhosis, elevated CK)
IV	Amylopektinosis (*Andersen disease, glykogenosis of defect branching*)	amylo–1→4–1→6–transglucosidase ("branching enzyme")	abnormal glycogen (amylopectin in organs)	hepatosplenomegalia, ascites, cirhosis, liver failure
V	McArdle disease	muscle phosphorylase (myophosphorylase)	skeletal muscles (only)	weakness and muscle spasms during exercise, without hyperlactatemia, but elevated CK
VI	Deficit of liver phosphorylase (*Hers syndrome*)	liver glycogen phosphorylase (50 % of normal)	liver	hepatomegalia, normal spleen, hypoglycemia, without hyperlipemia or ascites
VII		phosphofructokinase	muscles, erytrocytes	muscle weakness, spasms during exercise, mild hemolytic anemia
previously VIII, IX, VIb		phosphorylase b kinase	liver, leukocytes, muscles (?)	hepatomegalia, hypoglycemia (like type VI), X-linked inheritance
X		cAMP dependent proteinkinase	liver, muscles	hepatomegalia, not a disease of skeletal muscle or myocard
Nonclassified		activity of all enzymes is normal	liver or liver and kidneys	hepatomegalia with Fanconi syndrome; hypophosphatemic rickets (previously called vitamin D-resistant rickets); hereditary fructose intolerance