Table 1 Characteristics of inborn errors of glycogen metabolism

GSD type/name (phenotype MIM number)	Enzyme defect	Gene defect (OMIM number)	Chromosome location	Inheritance	Incidence	Clinical features
GSD0A/Liver GSD 0 (240600)	Liver glycogen synthase	GYS2 (138571)	12p12.1	AR	Unknown (<30 cases reported)	Fasting Ketotic hypoglycemia; hyperketonemia; hypoglycemic seizures; post-prandial hyperglycemia; post-prandial hyperlactatemia
GSD0B/Muscle GSD 0 (611556)	Muscle glycogen synthase	GYS1 (138570)	19q13.33	AR	Unknown (10 cases reported)	Muscle fatigue; seizures (rare); risk of cardiac arrest in childhood
GSD1A/Von Gierke/ Hepatorenal (232200)	Glucose-6-phosphatase	G6PC (613742)	17q21.31	AR	1 in 20,000 (Ashkenazi Jewish population)–1 in 100,000	Fasting hypoglycemia; lactic acidosis; hepatomegaly; growth delay/short stature; doll like facies; elevated liver enzymes; renal dysfunction; hyperuricemia; hypertriglyceridemia; osteoporosis; anemia; hepatic adenoma; hepatocellular carcinoma
GSD1B/G6P transport defect (232220)	Glucose-6-phosphate translocase	SLC37A4 (602671)	11q23.3	AR	Unknown	Recurrent bacterial infections; neutropenia; inflammatory bowel disease; oral/intestinal mucosal ulcers; fasting hypoglycemia; lactic acidosis; hepatomegaly; doll-like facies; anemia; growth delay/short stature; hyperlipidemia; xanthomas
GSD2/Pompe/Cardiac GSD (232300)	Acid maltase [alpha-1,4-glucosidase]	GAA (606800)	17q25.3	AR	1 in 8,684–1 in 40,000	Cardiomyopathy; muscular hypotonia; enlarged tongue; respiratory failure due to muscle weakness; adult onset limb girdle dystrophy
GSD3/Forbes/Cori/IIIa/IIIb	Glycogen debrancher [amylo-1,6 glucosidase]	AGL (610860)	1p21.2	AR	1 in 100,000	Hepatomegaly; hypoglycemia; fasting ketosis; failure to thrive; growth delay/short stature; myopathy; hypertrophic cardiomyopathy; doll like facies; hyperlipidemia; elevated liver enzymes
GSD4/Andersen/ Amylopectinosis/ Neuromuscular/ Polyglucosan (232500)	Glycogen brancher [amylo(1,4 to 1,6) transglucosidase]	GBE1 (607839)	3p12.2	AR	1 in 600,000–1 in 800,000	Failure to thrive; hepatosplenomegaly; progressive liver cirrhosis; fetal akinesia deformation sequence (FADS); hypotonia; muscle wasting/myopathy; cardiomyopathy; neurogenic bladder; peripheral neuropathy; leukodystrophy; cognitive impairment
GSD5/McArdle (232600)	Myophosphorylase	PYGM (608455)	11q13.1	AR	1 in 100,000–1 in 167,000	Skeletal muscle weakness; exercise-induced muscle cramping; rhabdomyolysis; myoglobinuria; “second wind” phenomenon
GSD6/Hers (232700)	Liver glycogen phosphorylase	PYGL (613741)	14q22.1	AR	1 in 1,000 (Mennonite population)–1 in 100,000	Hepatomegaly; growth retardation; mild hypoglycemia; fasting Ketotic hypoglycemia; fatigue; muscle hypotonia; motor developmental delay; osteoporosis

Table 1 (*continued*)