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*)

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GSD7/Tarui (232800)	Muscle phosphofructokinase	PFKM (610681)	12q13.11	AR	Unknown, but more prevalent among Ashkenazi Jewish population	Hemolytic anemia; muscle weakness; exercise-induced muscle cramping; exertional myopathy; gout/hyperuricemia
GSD9A1/XLG1/formerly GSD8 (306000)	Alpha-2 subunit of liver phosphorylase kinase	PHKA2 (300798)	Xp22.13	XLR	Unknown (~50 cases reported)	Hepatomegaly; growth retardation; motor developmental delay; hypercholesterolemia; hypertriglyceridemia; elevated liver enzymes; fasting hyperketosis
GSD9B/GSD IXb (261750)	Beta subunit of liver and muscle phosphorylase kinase	PHKB (172490)	16q12.1	AR	1 in 100,000	Short stature; hepatomegaly; diarrhea; muscle weakness; hypotonia
GSD9C/GSD IXc (613027)	Hepatic and testis isoform—gamma subunit of phosphorylase kinase	PHKG2 (172471)	16p11.2	AR	1 in 100,000	Growth retardation; hepatomegaly; hypotonia; cognitive delay
GSD9D/GSD IXd (300559)	Alpha subunit of muscle phosphorylase kinase	PHKA1 (311870)	Xq13.1	XLR	Unknown (only 7 cases reported)	Muscle weakness; exercise-induced muscle pain & stiffness; muscle atrophy; variable age onset, adults
GSD10/GSD X/PGAMM deficiency (261670)	Muscle phosphoglycerate mutase	PGAM2 (612931)	7p13	AR	Unknown (only 15 cases reported)	Exercise-induced muscle cramps & pain; exercise intolerance; rhabdomyolysis; myoglobinuria; hyperuricemia/gout; coronary arteriosclerosis; childhood or adolescence onset
GSD11/GSD XI/LDHA deficiency (612933)	Lactate dehydrogenase A	LDHA (150000)	11p15.1	AR	Unknown (only 12 cases reported)	Exercise-induced muscle cramps & pain; rhabdomyolysis; myoglobinuria; uterine muscle stiffness during pregnancy; psoriatic skin lesions; onset in childhood
GSD12/GSD XII/Aldolase deficiency (611881)	Fructose-1,6-bisphosphate aldolase A in red cell	ALDOA (103850)	16p11.2	AR	Unknown (<10 cases reported)	Short stature; myopathy; mental retardation; delayed puberty; hemolytic anemia; dysmorphic facies; hepatosplenomegaly; rhabdomyolysis with febrile illness
GSD13/GSD XIII/Enolase 3 deficiency (612932)	Beta-enolase	ENO3 (131370)	17p13.2	AR	Unknown (only 3 cases reported)	Exercise intolerance; myalgia; rhabdomyolysis

Table 1 (*continued*)

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GSD14/GSD XIV/CDG1t/ PGM1 deficiency (614921)	Phosphoglucomutase-1	PGM1 (171900)	1p31.3	AR	Unknown (only 22 cases reported)	Short stature; cleft palate; bifid uvula; Pierre Robin sequence; hepatopathy/chronic hepatitis; intermittent hypoglycemia; dilated cardiomyopathy; exercise intolerance; muscle weakness; rhabdomyolysis; hypogonadotropic hypogonadism; susceptibility to malignant hypothermia; hepatopathy/chronic hepatitis; intermittent hypoglycemia; variable phenotype
GSD15/GSD XV/GYG1 deficiency (613507)	Glycogenin-1	GYG1 (603942)	3q24	AR	Unknown (less than 20 cases reported)	Cardiac arrhythmias; muscle weakness
Fanconi-Bickel syndrome/ previously GSD XI (227810)	None (glucose transport defect)	GLUT2/SLC2A2 (138160)	3q26.2	AR	Unknown (200 cases reported)	Tubular nephropathy; hepatorenal glycogen storage; failure to thrive; polyuria; rickets; hyperuricemia; hyperaminoaciduria; hyperlipidemia; Ketotic hypoglycemia; hepatosplenomegaly
GSD Heart, lethal congenital (261740)	Gamma-2 subunit of AMP-activated protein kinase/cardiac muscle phosphorylase kinase	PRKAG2 (602743)	7q36.1	AD	Unknown (193 cases reported)	Hypoglycemia; heart failure; failure to thrive; cardiomegaly; cardiomyopathy; renomegaly; WPW syndrome; fatal in early infancy
Danon disease/lysosomal-associated membrane protein-2 deficiency/ formerly GSD2b or GSD IIb (300257)	[lysosomal-associated membrane protein-2 def-cy]	LAMP2 (309060)	Xq24	XLD	Unknown (171 cases reported)	Cardiomyopathy; skeletal myopathy; WPW syndrome; intellectual disability; hepatomegaly; retinopathy; arrhythmia
Brain GSD/Laforin deficiency (254780)	Laforin; E3 ligase	EPM2A (607566); NHLRC1/EPM2B (608072)	6q24.3; 6p22.3	AR; AR	Unknown; unknown	Epilepsy; hallucinations; dementia

AD, autosomal dominant; AR, autosomal recessive; XLD-X-linked dominant, XLR-X-linked recessive.