Background Paper on Glycogen Storage Disease

Glycogen storage disease (**GSD**) is the result of defects in the processing of glycogen synthesis or breakdown within muscles, liver, and other cell types. GSD in humans it is genetic caused by any inborn error of metabolism (genetically defective enzymes) involved in these processes. In livestock, acquired GSD can be caused by ingestion of toxic alkaloids.

There are eleven (11) distinct diseases that are commonly considered to be glycogen storage diseases (some previously thought to be distinct have been reclassified). (Although glycogen synthase deficiency does not result in storage of extra glycogen in the liver, it is often classified with the GSDs as type 0 because it is another defect of glycogen storage and can cause similar problems).

GSD type VIII: In the past, considered a distinct condition, now classified with VI. Has been described as X-linked recessive.

GSD type X: In the past, considered a distinct condition, now classified with VI.

Number	Enzyme deficiency	Eponym	Incidence	Hypo-glycemia?	Hepato-megaly?	Hyperlip-idemia?	Muscle symptoms	Development/ prognosis	Other symptoms
GSD type I	glucose-6-phosphatase	von Gierke's disease	1 in 50,000-100,000 births	Yes	Yes	Yes	None	Growth failure	Lactic acidosis, hyperuricemia
GSD type II	acid alpha-glucosidase	Pompe's disease	1 in 40,000 births	No	Yes	No	Muscle weakness	*Death by age ~2 years (infantile variant)	Heart failure
GSD type III	glycogen debranching enzyme	Cori's disease or Forbes' disease	1 in 100,000 births	Yes	Yes	Yes	Myopathy		
GSD type IV	glycogen branching enzyme	Andersen disease		No	Yes, also cirrhosis	No	None	Failure to thrive, death at age ~5 years	
GSD type V	muscle glycogen phosphorylase	McArdle disease	1 in 100,000[12]	No	No	No	Exercise-induced cramps, Rhabdomyolysis		Renal failure by myoglobinuria, second wind phenomenon
GSD type VI	liver	Hers'	1 in 65,000-	Yes	Yes	Yes [14]	None		

	glycogen phosphorylase	disease	85,000 births[13]						
GSD type VII	muscle phosphofructokinase	Tarui's disease		No	No	No	Exercise-induced muscle cramps and weakness	Growth retardation	Haemolytic anaemia
GSD type IX	phosphorylase kinase, PHKA2	-		Yes	Yes	Yes	None	Delayed motor development, Growth retardation	
GSD type XI	glucose transporter, GLUT2	Fanconi-Bickel syndrome		Yes	Yes	No	None		
GSD type XII	Aldolase A	Red cell aldolase deficiency		?	?	?	Exercise intolerance, cramps		
GSD type XIII	β-enolase	-		?	?	?	Exercise intolerance, cramps	Increasing intensity of myalgias over decades [15]	Serum CK: Episodic elevations; Reduced with rest [15]
GSD type 0	glycogen synthase	-		Yes	No	No	Occasional muscle cramping	Growth failure in some cases	

Glycogen Storage Disease (new Topic WB- DSG only)

Obligatory	**Must not donate if:** Suffers from a Glycogen Storage Disease.
Discretionary	If the potential donor suffers from type 0 (glycogen synthase deficiency), type V (McArdle disease), type XI (Fanconi-Bickel syndrome), type XII (Red cell aldolase deficiency), or type XIII Glycogen Storage Disease, accept.
Additional Information	Glycogen storage disease (GSD) is the result of defects in the processing of glycogen synthesis or breakdown within muscles, liver, and other cell types. GSD in humans is genetic caused by any inborn error of metabolism (genetically defective enzymes) involved in these processes.