

Clinical Pediatric Neurology

GLYCOGEN STORAGE DISEASE:

Disease	Enzyme deficiency	Hypo-glycemia	Hepato-megaly	Hyperlip- idemia	Muscle symptoms	mnemonics
von Gierke's	glucose-6-phosphatase	Yes	Yes	Yes	None	
Pompe's	acid maltase (Lys 1-4 glucosidase)	No	Yes	No	Muscle weakness	P for Pump (develop HF)
Cori's	glycogen debrancher	Yes	Yes	Yes	Myopathy	
Andersen	glycogen branching enzyme	No	Yes	No	None	
McArdle'	muscle glycogen phosphorylase	No	No	No	Exercise-induced cramps	M for muscles (glycogen phos)
Hers'	liver glycogen phosph.	Yes	Yes	No	None	H for Hepatic (glycogen phos)

LYSOSOMAL STORAGE DISEASE:

Disease	Finding	Deficient Enzyme	Accumulated substrate	Inheritance
Gaucher's	Hepatosplenomegaly, aseptic necrosis of femur, bone crises, Gaucher's cells (macrophages look like crumpled paper)	Glucocerebrosidase	Glucocerebroside	AR
Niemann-pick	Progressive neurodegeneration, hepatosplenomegaly, cherry-red spot (on macula), foam cells	Sphingomyelinase	Sphingomyelin	AR
Tay-Sachs	Progressive neurodegeneration, developmental delay, Cherry red spot, lysosomes with onion skin	Hexosaminidase A	GM2 ganglioside	AR
Metachromatic leukodystrophy	Central and peripheral demyelination with ataxia, dementia	Arylsulfatase A	Cerebroside sulfate	AR
Fabry's	Peripheral neuropathy of hands/feet, angiokeratomas, cardiovascular disease	Galactosidase A	Ceramide trihexoside	XR
Krabbe's	Peripheral neuropathy, developmental delay, optic atrophy, <u>globoid</u> cells	Galacto-cerebrosidase	Galactocerebroside	AR
Hurler's	Developmental delay, Gargoylism, airway obstruction, corneal clouding, hepatosplenomegaly	Iduronidase	Heparan sulfate, dermatan sulfate	AR
Hunter's	Mild Hurler + aggressive behavior, no corneal clouding	Iduronate sulfatase	Heparan sulfate, dermatan sulfate	XR

- Gaucher's: no neurological symptoms
- Niemann-Pick and Tay Sachs: both associated with cherry red spot, Niemann-Pick also has HSM
- Fabry's and Krabbe's: both have peripheral neuropathy. Fabry's has normal development and angiokeratomas, Krabbe's has developmental delay and optic atrophy
- Hurler's and Hunter's: Hunter's is a milder form of Hurler's (remember, hunters need good vision, no corneal clouding)