Lysosomal Storage Diseases/Sphingolipidoses

Disease	***Enzyme Deficiency***	***Accumulating Product***	***Results/Characteristics*** **autosomal recessive unless otherwise noted)**
Fabry's	alpha-galactocerebrosidase A Alpha-galctosidase A	Ceramide Trihexoside	**X-linked** recessive; renal failure ***Enzyme Replacement Therapy***
Krabbe's	galactosylceramide B-galacto-sdase, Galactosyl ceramidase	galactocerebroside (brain)	optic atrophy, spasticity, early death
Gaucher's	B-glucocerebrosidase Glucocosylceramidase	glucocerebroside (brain, liver, spleen, bone marrow)	"crinkled paper" enlarged cytoplasm ***Enzyme Replacement Therapy*** liver and spleen enlargement **mental retardation in infantile form only**
Niemann-Pick	sphingomyelinase	sphingomyelin & cholesterol (reticuloendothelial & parenchymal cells)	Death by age 3, enlarged liver & spleen **mental retardation**
Tay-Sachs	Hexosaminidase A	GM2 ganglioside	Death by age 3, cherry-red spot on macula Carrier rate: 1/30 Jews of European descent 1/300 for others), **mental retardation**, blindness
Metachromatic Leukodystrophy	arylsulfatase A sulfatidase	sulfatide (brain, kidney, liver, peripheral nerves**)**	white matter signs, peripheral neuropathy **mental retardation**, demyelination, Nerves stain yellowish brown with cresyl violet
Farber's	Acid ceramidase	ceramide	Painful and progressively deformed joints Subcutaneous nodules, ganulomas, fatal early

Mucopolysaccharidoses – Most Common Forms

Hurler's	a-L-iduronidase	corneal clouding, **mental retardation**
Hunter's	iduronate sulfatase	Mild form of Hurler's with no corneal clouding **X-linked recessive**