

Lysosomal Storage Diseases

S.N	Disease Name	Substrate	Enzyme/Protein	Gene
1	Tay-sachs disease	Ganglioside-gm2	Beta-hexosaminidase A	HEXA
2	Gaucher disease	Glucocerebroside	Beta-glucocerebrosidase	GBA
3	Hunter syndrome	Sulfated alpha-l-iduronic acid	Iduronate 2-sulfatase	IDS
4	Pompe disease	Glycogen	Acid maltase	GAA
5	Niemann pick disease	Sphingomyelin	Acid sphingomyelinase	SMPD1
6	Fabry disease	Globotriaosylceramide	Alpha-galactosidase A	GLA
7	Metachromatic leukodystrophy	Sulfatides	Arylsulfatase A	ARSA
8	Krabbe	Galactosylceramide	Galactosylceramidase	GALC
9	Salla disease	Sialic acid	Sialin	SLC17A5
10	Obalamin C disease	Cobalamin	Cobalamin reductases	MMACHC
11	Mucopolidosis II	Functional components	Glcnae-1-phosphotransferase	GNPTAB
12	Sandhoff disease	GM2 ganglioside	Beta-hexosaminidase	HEXB
13	Cystinosis	Cystine	Cystinosis	CTNS

Lysosomes And Lysosomal Storage Diseases

Michael Seilmaier



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