

Types of Lysosomal Storage Disorders

Disease	Clinical Phenotype	Enzyme Deficiency	Chromosome Location
Aspartylglucosaminuria		Aspartylglucosaminidase	4q32-33
Cholesterol ester storage disease	Wolman disease	Acid lipase	10q24-25
Cystinosis		Cystine transporter	17
Fabry disease	Fabry disease	α -Galactosidase A	Xq22
Farbar Lipogranulomatosis	Farber disease	Acid ceramidase	8p21.3-p22
Fucosidosis		α -L-Fucosidase	1p34
Galactosialidosis types I / II		Protective protein	20q13.1
Gaucher disease types I / II / III	Gaucher disease	Glucocerebrosidase (β -glucosidase)	1q21
Globoid cell leucodystrophy	Krabbe disease	Galactocerebrosidase	14q31
Glycogen storage disease II	Pompe disease	α -Glucosidase	17q25.2-25.3
GM1-Gangliosidosis types I/II/III		β -Galactosidase	3p21-3pter
GM2-Gangliosidosis type I	Tay Sachs disease	β -Hexosaminidase A	15q23-24
GM2-Gangliosidosis type II	Sandhoff disease	β -Hexosaminidase A & B	5q13
GM2-Gangliosidosis		GM2-activator deficiency	5q32-33
α -Mannosidosis types I / II		α -D-Mannosidase	19p13.2-q12
β -Mannosidosis		β -D-Mannosidase	4q22-q25
Metachromatic leucodystrophy		Arylsulphatase A	22q13.3-qter
Metachromatic leucodystrophy		Saposin B	10q2
Mucopolipidosis type I	Sialidosis types I / II	Neuraminidase	6p21.3
Mucopolipidosis types II / III	I-cell disease: pseudo-Hurler Polydystrophy	Phosphotransferase	4q.21-23
Mucopolipidosis type IIIC	pseudo-Hurler Polydystrophy	Phosphotransferase γ -subunit	16p
Mucopolipidosis type IV		Unknown	19p13.2-p12.3
Mucopolysaccharidosis type I	Hurler syndrome Schele syndrome	α -L-Iduronidase	4p16.3
Mucopolysaccharidosis type II	Hunter syndrome	Iduronate-2-sulphatase	Xq27-28
Mucopolysaccharidosis type IIIA	Sanfilippo syndrome	Heparan-N-sulphatase	17q25.3
Mucopolysaccharidosis type IIIB	Sanfilippo syndrome	α -N-Acetylglucosaminidase	17q21
Mucopolysaccharidosis type IIIC	Sanfilippo syndrome	AcetylCoA:N-acetyltransferase	Unknown
Mucopolysaccharidosis type IIID	Sanfilippo syndrome	N-Acetylglucosamine 6-sulphatase	12q14
Mucopolysaccharidosis type IVA	Morquio syndrome	Galactose 6-sulphatase	16q24.3
Mucopolysaccharidosis type IVB	Morquio syndrome	β -Galactosidase	3p21-3pter
Mucopolysaccharidosis type VI	Maroteaux-Lamy syndrome	N-Acetylgalactosamine 4-sulphatase	5q11-13
Mucopolysaccharidosis type VII	Sly syndrome	β -Glucuronidase	7q21.1.11
Multiple sulphatase deficiency		Multiple sulphates	Unknown
Neuronal Ceroid Lipofuscinosis, CLN1	Batten disease	Palmitoyl protein thioesterase	1p34
Neuronal Ceroid Lipofuscinosis, CLN2	Batten disease	Tripeptidyl peptidase I	11p15.5
Neuronal Ceroid Lipofuscinosis, CLN3	Vogt-Spielmeyer disease	Protein function not known	16p12.1
Neuronal Ceroid Lipofuscinosis, CLN5	Batten disease	Protein function not known	13q22
Neuronal Ceroid Lipofuscinosis, CLN8	Northern Epilepsy	Protein function not known	8pter-p23
Niemann-Pick disease Types A / B	Niemann-Pick disease	Acid sphingomyelinase	11p15.1-p15.4
Niemann-Pick disease type C1	Niemann-Pick disease	Cholesterol trafficking	18q11-12
Niemann-Pick disease type C2	Niemann-Pick disease	Cholesterol trafficking	Unknown
Pycnodysostosis		Cathepsin K	1q21
Schindler disease types I / II	Schindler disease	α -Galactosidase B	22q13.1-13.2
Sialic acid storage disease	Sialuria, Sall disease	Sialic acid transporter	6q14-15

Source: Information adapted from LDA Newsletter April 2000, Lysosomal Diseases Australia, www.LDA.org.au