

# TABLE: A summary of LSD and therapies

Disease	Clinical Phenotype	Enzyme Deficiency	Chromosome Location	Bone Marrow Transplantation	Animal Model	Human Enzyme Replacement Therapy	Australian Prevalence
Aspartylglucosaminuria		Aspartylglucosaminidase	4q32 - 33		mouse		1 in 2,111,000
Cholesterol ester storage disease	Wolman disease	Acid lipase	10q24-25				1 in 528,000
Cystinosis		Cystine transporter	17				1 in 192,000
Fabry disease	Fabry disease	$\alpha$ -Galactosidase A	Xq22		mouse	Trials in progress	1 in 117,000
Farber Lipogranulomatosis	Farber disease	Acid ceramidase	8p21.3-p22	Not helpful in severe forms			
Fucosidosis		$\alpha$ -L-Fucosidase	1p34	Benefit in dog model	dog		> 1 in 2,000,000
Galactosialidosis types I / II		Protective protein	20q13.1		sheep		
Gaucher disease types I / II / III	Gaucher disease	Glucocerebrosidase ( $\beta$ -glucosidase)	1q21	May benefit in type I; uncertain other types	mouse	Type I in clinical practice; Trial in Type III in progress	1 in 57,000
Globoid cell leucodystrophy	Krabbe disease	Galactocerebrosidase	14q31	May benefit presymptomatic patients	mouse, sheep, dog, monkey		1 in 201,000
Glycogen storage disease II	Pompe disease	$\alpha$ -Glucosidase	17q25.2-25.3	Not helpful	dog, cattle, quail,	Trials in progress	1 in 146,000
GM1-Gangliosidosis types I/II/III		$\beta$ -Galactosidase	3p21-3pter	No benefit in dog	cat, dog, sheep, cattle		1 in 384,000
GM2-Gangliosidosis type I	Tay Sachs disease	$\beta$ -Hexosaminidase A	15q23-24	No benefit	mouse		1 in 201,000
GM2-Gangliosidosis type II	Sandhoff disease	$\beta$ -Hexosaminidase A & B	5q13	No benefit	mouse		1 in 384,000
GM2-Gangliosidosis		GM2-activator deficiency	5q32-33		dog		
$\alpha$ -Mannosidosis types I / II		$\alpha$ -D-Mannosidase	19p13.2-q12		mouse, cat, cattle, guinea pig		1 in 1,056,000
$\beta$ -Mannosidosis		$\beta$ -D-Mannosidase	4q22-q25		goats, cattle		
Metachromatic leucodystrophy		Arylsulphatase A	22q13.3-qter	May benefit presymptomatic patients	mouse		1 in 92,000
Metachromatic leucodystrophy		Saposin B	10q2	May benefit presymptomatic patients			
Mucopolipidosis type I	Sialidosis types I / II	Neuraminidase	6p21.3				
Mucopolipidosis types II / III	I-cell disease; pseudo-Hurler polydystrophy	Phosphotransferase	4q.21-23	Benefit reported for one MLII patient	cat		1 in 325,000
Mucopolipidosis type IIIC	pseudo-Hurler polydystrophy	Phosphotransferase $\gamma$ -subunit	16p				
Mucopolipidosis type IV		Unknown	19p13.2-p13.3				
Mucopolysaccharidosis type I	Hurler syndrome Scheie syndrome	$\alpha$ -L-Iduronidase	4p16.3	May benefit presymptomatic patients	cat, dog, mouse	Trials in progress	1 in 88,000
Mucopolysaccharidosis type II	Hunter syndrome	Iduronate-2-sulphatase	Xq27-28	May benefit presymptomatic patients	mouse, dog	Trials planned	1 in 136,000
Mucopolysaccharidosis type IIIA	Sanfilippo syndrome	Heparan-N-sulphatase	17q25.3	Not helpful in symptomatic patients	mouse, dog		1 in 114,000
Mucopolysaccharidosis type IIIB	Sanfilippo syndrome	$\alpha$ -N-Acetylglucosaminidase	17q21	Not helpful in symptomatic patients	mouse, emu		1 in 211,000
Mucopolysaccharidosis type IIIC	Sanfilippo syndrome	AcetylCoA:N-acetyltransferase	unknown	Not helpful in symptomatic patients			1 in 1,407,000
Mucopolysaccharidosis type IIID	Sanfilippo syndrome	N-Acetylglucosamine 6-sulphatase	12q14	Not helpful in symptomatic patients	goat		1 in 1,056,000
Mucopolysaccharidosis type IVA	Morquio syndrome	Galactose 6-sulphatase	16q24.3	Not helpful			1 in 169,000
Mucopolysaccharidosis type IVB	Morquio syndrome	$\beta$ -Galactosidase	3p21-3pter	Not helpful			
Mucopolysaccharidosis type VI	Maroteaux-Lamy syndrome	N-Acetylglucosamine 4-sulphatase	5q11-13	May benefit	cat, rat, dog, mouse	Trials planned	1 in 235,000
Mucopolysaccharidosis type VII	Sly syndrome	$\beta$ -Glucuronidase	7q21.1.11		dog, mouse, cat		1 in 2,111,000
Multiple sulphatase deficiency		Multiple sulphatases	unknown				1 in 1,407,000
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-Spielmeier disease	Protein function not known	16p12.1		mouse, dog, sheep		
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	Not helpful for type A		Trials planned	1 in 248,000
Niemann-Pick disease type C1	Niemann-Pick disease	Cholesterol trafficking	18q11-12		cat, mouse		1 in 211,000
Niemann-Pick disease type C2	Niemann-Pick disease	Cholesterol trafficking	unknown				
Pycnodysostosis		Cathepsin K	1q21				
Schindler disease types I / II	Schindler disease	$\alpha$ -Galactosidase B	22q13.1-13.2				
Sialic acid storage disease	Sialuria, Salla disease	Sialic acid transporter	6q14-15				1 in 528,000

Prevalence figures quoted from Melkie et al., JAMA 281:249-254 (1999). Prevalence and ratio of lysosomal storage disorders may vary from country to country.