Chart of Allied Diseases

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A. LYSOSOMAL STORAGE DISORDERS

1) Disorders of lipid and sphingolipid degradation

Disease	Enzyme Defect	OMIM#	Inheritance Pattern	Age of Onset	Cognitive Impairment	Links
GM1 Gangliosidosis	b-Galactosidase-1	230500	AR	variable	progressive psychomotor deterioration	
Tay-Sachs Disease	b-Hexosaminidase A	272800	AR	variable	progressive psychomotor deterioration	
Sandhoff Disease	b-Hexosaminidases A and B	268800	AR	variable	progressive psychomotor deterioration	
GM2 Gangliodisosis, AB variant	GM2 Activator Protein	272750	AR	infancy	progressive psychomotor deterioration	
Fabry Disease	8-Galactosidase A	301500	X-linked	adolesence - adulthood	normal intelligence	www.fabry.org
Gaucher Disease, Type 1	Glucocerebrosidase	230800	AR	variable	normal intelligence	www.gaucherdisease.org, www.gaucherdisease.org.uk
Gaucher Disease, Type II	Glucocerebrosidase	230900	AR	infancy	severe	www.gaucherdisease.org, www.gaucherdisease.org.uk
Gaucher Disease, Type III	Glucocerebrosidase	231000	AR	childhood	mild	www.gaucherdisease.org, www.gaucherdisease.org.uk
Metachromatic Leukodystrophy	Arylsulfatase A	250100	AR	infancy to adulthood	progressive psychomotor deterioration	www.ulf.org, www.MLDFoundation.org
Krabbe Disease	Galactosylceramidase	245200	AR	infancy to adulthood	progressive psychomotor deterioration	www.huntershope.org
Niemann-Pick, Type A	Sphingomyelinase	257200	AR	infancy	progressive psychomotor deterioration	www.nnpdf.org
Niemann-Pick, Type B	Sphingomyelinase	607616	AR	infancy - childhood	none to mild	www.nnpdf.org
Niemann-Pick, Type C1, Type C2	NPC1, HE1 protein (Cholesterol Trafficking Defect)	257220	AR	variable	progressive psychomotor deterioration	www.parseghian.org
Farber Disease	Acid Ceramidase	228000	AR	infancy	variable	
Wolman Disease (Chol.Esther Storage disease)	Lysosomal Acid Lipase	278000	AR	neonatal	progressive psychomotor deterioration	

2. Disorders of mucopolysaccharide degradation

Disease	Enzyme Defect	OMIM#	Inheritance Pattern	Age of Onset	Cognitive Impairment	Links
Hurler Syndrome (MPSI)	L-Iduronidase	252800	AR	infancy	severe mental retardation	www.mpssociety.org
Scheie Syndrome (MPS IS)	L-Iduronidase	252800	AR	childhood	normal intelligence	www.mpssociety.org
Hurler-Scheie (MPS IH/S)	L-Iduronidase	252800	AR	childhood	normal intelligence	www.mpssociety.org
Hunter Syndrome (MPS II)	Iduronate Sulfatase	309900	X-linked	infancy - childhood	variable	www.mpssociety.org
Sanfilippo A (MPS IIIA)	Heparan N--Sulfatase	252900	AR	infancy - childhood	progressive psychomotor deterioration	www.mpssociety.org
Sanfillippo B (MPS IIIB)	N-Acetylglucosaminidase	252920	AR	infancy - childhood	progressive psychomotor deterioration	www.mpssociety.org
Sanfillippo C (MPS IIIC)	Acetyl-CoA-Glucosaminidase	252930	AR	infancy - childhood	progressive psychomotor deterioration	www.mpssociety.org
Sanfillipo D (MPS IIID)	Acetyltransferase	252940	AR	infancy - childhood	progressive psychomotor deterioration	www.mpssociety.org
Morquio A (MPS IVA)	Acetylglucosamine-6-Sulfatase	253000	AR	infancy - childhood	normal intelligence	www.mpssociety.org, www.Morquio.com
Morquio B (MPS IVB)	Galactosamine-6--Sulfatase	253010	AR	variable	normal intelligence	www.mpssociety.org, www.Morquio.com
Maroteaux-Lamy (MPS VI)	Arylsulfatase B	253200	AR	infancy - childhood	normal intelligence	www.mpssociety.org
Sly Syndrome (MPS VII)	Glucuronidase	253220	AR	variable	variable	www.mpssociety.org

3. Disorders of glycoprotein degradation

Disease	Enzyme Defect	OMIM#	Inheritance Pattern	Age of Onset	Cognitive Impairment	Links
Alpha Mannosidosis	mannosidase	248500	AR	infancy - adolesence	mild to severe mental retardation	www.mannosidosis.org
Beta Mannosidosis	mannosidase	248510	AR	childhood - adulthood	mental retardation	www.mannosidosis.org
Fucosidosis	l-fucosidase	230000	AR	infancy - adolesence	mental retardation	www.mannosidosis.org
Asparylglucosaminuria	Aspartylglycosaminidase	208400	AR	childhood	mental retardation	www.mannosidosis.org
Mucolipidosis I (Sialidosis)	Neuraminidase	256550	AR	adolesence	none (type I) mental retardation (type II)	www.mannosidosis.org
Galactosialidosis	Lysosomal protective protein	256540	AR	infancy - adulthood	variable	www.mannosidosis.org
Schindler Disease	Lysosomal 8-N-acetylgalactosaminidase	104170	AR	infancy	progressive psychomotor deterioration	www.mannosidosis.org
Schindler Disease Type II/Kanzaki Disease	Lysosomal 8-N-acetylgalactosaminidase	104170	AR	adulthood	mild intellectual impairment	www.mannosidosis.org

4. Other lysosomal storage disorders

Disease	Enzyme Defect	OMIM#	Inheritance Pattern	Age of Onset	Cognitive Impairment	Links
Santavuori-Haltia Disease (Infantile Neuronal Ceroid Lipofuscinosis Type 1)	Palmitoyl-protein thioesterase	256730	AR	infancy	progressive psychomotor deterioration	www.bdsra.org
Jansky-Bielschowsky Disease (Late Infantile Neuronal Ceroid Lipofuscinosis Type 2)	at least 4 subtypes	204500	AR	late infancy	progressive psychomotor deterioration	www.bdsra.org
Batten Disease (Juvenile Neuronal Ceroid Lipofuscinosis Type 3)	Lysosomal membrane protein	204200	AR	childhood	slow intellect loss/psychosis/variable	www.bdsra.org
Kufs Disease (Neuronal Ceroid Lipofuscinosis Type 4)	Unknown	204300	AR	adulthood	dementia/psychosis	www.bdsra.org
Von Gierke Disease (Glycogen storage disease type Ia)	Glucose-6-phosphatase	232200	AR	infancy	normal intelligence	www.agsdus.org
Glycogen storage disease type Ib	Glucose-6-phosphate translocase	232220	AR	infancy	normal intelligence	www.agsdus.org
Pompe Disease (Glycogen Storage Disease Type II)	Acid maltase	232300	AR	infancy - adulthood	normal intelligence	www.pompe.com, www.amda-pompe.org
Forbes or Cori Disease (Glycogen storage disease type III)	Debrancher enzyme amylo-1,6 glucosidase	232400	AR	early childhood	normal intelligence	www.agsdus.org

Mucolipidosis II (I-Cell Disease)	N-acetylglucosamine-1- phosphotransferase	252500	AR	infancy	severe psychomotor retardation/developmental delay/mental retardation	www.mpssociety.org
Mucolipidosis III (Pseudo-Hurler Polydystrophy)	N-acetylglucosamine-1- phosphotransferase	252600	AR	childhood	mild to moderate mental retardation/learning disabilities/variable	www.mpssociety.org
Mucolipdosis IV (Sialolipidosis)	Ganglioside sialidase (neuraminidase)	252650	AR	infancy	psychomotor retardation	www.ml4.org
Cystinosis (adult nonnephropathic type)	Lysosomal cystine transport protein	219750	AR	adulthood	normal intelligence	www.cystinosisfoundation.org
Cystinosis (infantile nephropathic type)	Lysosomal cystine transport protein	219800	AR	infancy	normal intelligence	www.cystinosisfoundation.org
Cystinosis (juvenile or adolescent nephropathic)	Lysosomal cystine transport protein	219900	AR	adolescence	normal intelligence	www.cystinosisfoundation.org
Salla Disease/Infantile Sialic acid storage disorder	Sialic acid transport protein	269920	AR	infancy - adulthood	psychomotor retardation/mental retardation	
Saposin Deficiencies	Saposins A, B, C or D	176801	AR	infancy - adulthood	neurological deterioration, variable	

B. LEUKODYSTROPHY

Disease	Enzyme Defect	OMIM#	Inheritance Pattern	Age of Onset	Cognitive Impairment	Links
Abetalipoproteinemia	Microsomal triglyceride transfer protein/apolipoprotein B	200100	AR	infancy - adulthood	normal intelligence	www.ntsad.org
Adrenoleukodystrophy	Peroxisomal membrane transfer protein	300100	X-Linked	childhood and adolescence	progressive psychomotor deterioration	www.ulf.org, www.myelin.org
Neonatal Adrenoleukodystrophy	Peroxins	202370	AR	neonatal	severe mental retardation	www.ulf.org
Canavan Disease	Aspartoacylase	271900	AR	infancy	progressive psychomotor deterioration	www.canavanfoundation.org, www.ntsad.org
Cerebrotendinous Xanthromatosis	Sterol-27-hydroxlase	213700	AR	childhood	some have mental retardation while others retain normal intelligence	www.ulf.org
Pelizaeus Merzbacher Disease	Proteolipid protein	312080	X-linked	infancy	perhaps progressive cognitive impairment/dementia but need more neurologic findings	www.pmdfoundation.org
Tangier Disease	ABC1 transporter	205400	AR	variable	normal intelligence	www.ntsad.org
Refum Disease, infantile	Peroxisome membrane protein 3 or Peroxisome biogenesis factor 1	266510	AR	infancy	mental retardation and developmental delay	www.ntsad.org
Refum Disease, classic	Phytanic acid oxidase	266500	AR	childhood - adulthood	normal intelligence	www.ntsad.org

Yes* = if affected family member is found. Note: Reccurance risk after one affected child for all listed diseases = 25% for each pregnancy.

This table reflects information current at the time of printing. Reproductive and/or therapeutic decisions should not be made on the information provided without first consulting a geneticist or genetic counselor for any updated information.