

LYSOSOMAL DISORDERS GENE TABLE

Note: This panel can be ordered from the Mitochondrial/Metabolic test requisition form.

Gene Name	OMIM #	Associated Disorder(s)	Inheritance Pattern
<i>ABHD5</i>	275630	Chanarin-Dorfman syndrome	Autosomal recessive
<i>ADAMTSL2</i>	231050	Geleophysic dysplasia 1	Autosomal recessive
<i>AGA</i>	208400	Aspartylglucosaminuria	Autosomal recessive
<i>ARSA</i>	250100	Metachromatic leukodystrophy	Autosomal recessive
<i>ARSB</i>	253200	Mucopolysaccharidosis type VI; Maroteaux-Lamy syndrome	Autosomal recessive
<i>ASAH1</i>	613468	Farber lipogranulomatosis; Spinal muscular atrophy with progressive myoclonic epilepsy	Autosomal recessive
<i>ATP6AP1</i>	300972	Immunodeficiency 47	X-linked
<i>CLN3</i>	204200	Neuronal ceroid lipofuscinosis 3; Batten disease	Autosomal recessive
<i>CLN5</i>	256731	Neuronal ceroid lipofuscinosis 5	Autosomal recessive
<i>CLN6</i>	606725	Neuronal ceroid lipofuscinosis 6; Kufs disease	Autosomal recessive
<i>CLN8</i>	600143	Neuronal ceroid lipofuscinosis 8	Autosomal recessive
<i>CTNS</i>	606272	Cystinosis	Autosomal recessive
<i>CTSA</i>	256540	Galactosialidosis	Autosomal recessive
<i>CTSD</i>	610127	Neuronal ceroid lipofuscinosis 10; Cathepsin D deficiency	Autosomal recessive
<i>CTSF</i>	615362	Neuronal ceroid lipofuscinosis 13; Kufs disease type B	Autosomal recessive
<i>DNAJC5</i>	162350	Neuronal ceroid lipofuscinosis 4B	Autosomal dominant
<i>FUCA1</i>	230000	Fucosidosis	Autosomal recessive
<i>GAA</i>	232300	Glycogen storage disease II; Pompe disease	Autosomal recessive
<i>GALC</i>	245200	Krabbe disease	Autosomal recessive
<i>GALNS</i>	253000	Mucopolysaccharidosis IVA; Morquio syndrome A	Autosomal recessive
<i>GBA</i>	606463	Gaucher disease	Autosomal recessive
<i>GLA</i>	301500	Fabry disease	X-linked
<i>GLB1</i>	611458	Mucopolysaccharidosis type IVB; Morquio syndrome B; GM1-gangliosidosis	Autosomal recessive
<i>GNE</i>	603824	GNE myopathy; Nonaka myopathy; Sialuria	Autosomal recessive and Autosomal dominant
<i>GNPTAB</i>	607840	Mucopolipidosis II; Mucopolipidosis III	Autosomal recessive
<i>GNPTG</i>	252605	Mucopolipidosis III gamma	Autosomal recessive
<i>GNS</i>	252940	Mucopolysaccharidosis IIID; Sanfilippo type D	Autosomal recessive
<i>GPC3</i>	312870	Simpson-Golabi-Behmel syndrome type 1	X-linked
<i>GRN</i>	138945	Neuronal ceroid lipofuscinosis 11; Frontotemporal lobar degeneration, ubiquitin-positive	Autosomal dominant and Autosomal recessive
<i>GUSB</i>	253220	Mucopolysaccharidosis VII; Sly syndrome	Autosomal recessive
<i>HEXA</i>	606869	GM2-gangliosidosis; Tay-Sachs disease	Autosomal recessive
<i>HEXB</i>	268800	Sandhoff disease	Autosomal recessive
<i>HGSNAT</i>	252930	Mucopolysaccharidosis IIIC; Sanfilippo type C	Autosomal recessive
<i>HYAL1</i>	601492	Mucopolysaccharidosis IX	Autosomal recessive
<i>IDS</i>	309900	Mucopolysaccharidosis II; Hunter syndrome	X-linked

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<i>IDUA</i>	607014	Mucopolysaccharidosis I; Hurler/Scheie syndrome	Autosomal recessive
<i>LAMP2</i>	300257	Danon disease	X-linked
<i>LIPA</i>	278000	Wolman disease; Cholesteryl ester storage disease	Autosomal recessive
<i>LYST</i>	214500	Chediak-Higashi syndrome	Autosomal recessive
<i>MAN2B1</i>	248500	Alpha-mannosidosis	Autosomal recessive
<i>MANBA</i>	248510	Beta-mannosidosis	Autosomal recessive
<i>MCOLN1</i>	252650	Mucopolidosis IV	Autosomal recessive
<i>MFSD8</i>	610951	Neuronal ceroid lipofuscinosis 7	Autosomal recessive
<i>NAGA</i>	609241	Schindler disease; Infantile neuroaxonal dystrophy	Autosomal recessive
<i>NAGLU</i>	252920	Mucopolysaccharidosis IIIB; Sanfilippo type B	Autosomal recessive
<i>NEU1</i>	608272	Sialidosis type I; Sialidosis type II	Autosomal recessive
<i>NPC1</i>	257220	Niemann-Pick disease type C	Autosomal recessive
<i>NPC2</i>	607625	Niemann-Pick disease type C	Autosomal recessive
<i>PNPLA2</i>	610717	Neutral lipid storage disease with myopathy	Autosomal recessive
<i>PPT1</i>	256730	Neuronal ceroid lipofuscinosis 1	Autosomal recessive
<i>PSAP</i>	176801	Prosaposin deficiency	Autosomal recessive
<i>SCARB2</i>	254900	Progressive myoclonic epilepsy 4; Action myoclonus-renal failure syndrome	Autosomal recessive
<i>SGSH</i>	252900	Mucopolysaccharidosis IIIA; Sanfilippo type A	Autosomal recessive
<i>SLC17A5</i>	604322	Free sialic acid storage disease; Salla disease	Autosomal recessive
<i>SMPD1</i>	607608	Acid sphingomyelinase deficiency; Niemann-Pick disease type A; Niemann-Pick disease type B	Autosomal recessive
<i>SUMF1</i>	272200	Multiple sulfatase deficiency	Autosomal recessive
<i>TPP1</i>	607998	Neuronal ceroid lipofuscinosis 2; Spinocerebellar ataxia type 7	Autosomal recessive
<i>VPS33A</i>	617303	Mucopolysaccharidosis-plus syndrome	Autosomal recessive



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