LYSOSOMAL DISORDERS
GENE LIST (~57)

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

Gene Name	OMIM #	Associated Disorder(s)	Inheritance Pattern
ABHD5	275630	Chanarin-Dorfman syndrome	Autosomal recessive
ADAMTSL2	231050	Geleophysic dysplasia 1	Autosomal recessive
AGA	208400	Aspartylglucosaminuria	Autosomal recessive
ARSA	250100	Metachromatic leukodystrophy	Autosomal recessive
ARSB	253200	Mucopolysaccharidosis type VI; Maroteaux-Lamy syndrome	Autosomal recessive
ASAH1	613468	Farber lipogranulomatosis; Spinal muscular atrophy with progressive myoclonic epilepsy	Autosomal recessive
ATP6AP1	300972	Immunodeficiency 47	X-linked
CLN3	204200	Neuronal ceroid lipofuscinosis 3; Batten disease	Autosomal recessive
CLN5	256731	Neuronal ceroid lipofuscinosis 5	Autosomal recessive
CLN6	606725	Neuronal ceroid lipofuscinosis 6; Kufs disease	Autosomal recessive
CLN8	600143	Neuronal ceroid lipofuscinosis 8	Autosomal recessive
CTNS	606272	Cystinosis	Autosomal recessive
CTSA	256540	Galactosialidosis	Autosomal recessive
CTSD	610127	Neuronal ceroid lipofuscinosis 10; Cathepsin D deficiency	Autosomal recessive
CTSF	615362	Neuronal ceroid lipofuscinosis 13; Kufs disease type B	Autosomal recessive
DNAJC5	162350	Neuronal ceroid lipofuscinosis 4B	Autosomal dominant
FUCA1	230000	Fucosidosis	Autosomal recessive
GAA	232300	Glycogen storage disease II; Pompe disease	Autosomal recessive
GALC	245200	Krabbe disease	Autosomal recessive
GALNS	253000	Mucopolysaccharidosis IVA; Morquio syndrome A	Autosomal recessive
GBA	606463	Gaucher disease	Autosomal recessive
GLA	301500	Fabry disease	X-linked
GLB1	611458	Mucopolysaccharidosis type IVB; Morquio syndrome B; GM1-gangliosidosis	Autosomal recessive
GNE	603824	GNE myopathy; Nonaka myopathy; Sialuria	Autosomal recessive and Autosomal dominant
GNPTAB	607840	Mucolipidosis II; Mucolipidosis III	Autosomal recessive
GNPTG	252605	Mucolipidosis III gamma	Autosomal recessive
GNS	252940	Mucopolysaccharidosis IIID; Sanfilippo type D	Autosomal recessive
GPC3	312870	Simpson-Golabi-Behmel syndrome type 1	X-linked
GUSB	253220	Mucopolysaccharidosis VII; Sly syndrome	Autosomal recessive
HEXA	606869	GM2-gangliosidosis; Tay-Sachs disease	Autosomal recessive
HEXB	268800	Sandhoff disease	Autosomal recessive
HGSNAT	252930	Mucopolysaccharidosis IIIC; Sanfilippo type C	Autosomal recessive
HYAL1	601492	Mucopolysaccharidosis IX	Autosomal recessive
IDS	309900	Mucopolysaccharidosis II; Hunter syndrome	X-linked
IDUA	607014	Mucopolysaccharidosis I; Hurler/Scheie syndrome	Autosomal recessive
LAMP2	300257	Danon disease	X-linked

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

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