COMBINED LYSOSOMAL AND PEROXISOMAL DISORDERS
GENE TABLE

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

	Gene Name	OMIM #	Associated Disorder(s)	Inheritance Pattern
Lysosomal Disorders	*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 or 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	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	Autosomal recessive
	CTSF	615362	Neuronal ceroid lipofuscinosis 13	Autosomal recessive
	DNAJC5	162350	Neuronal ceroid lipofuscinosis 4B	Autosomal recessive
	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	Autosomal recessive
	GBA	606463	Gaucher disease	Autosomal recessive
	GLA	301500	Fabry disease	X-linked
	GLB1	611458	Mucopolysaccharidosis type IVB or Morquio syndrome; GM1-gangliosidosis	Autosomal recessive
	GNE	603824	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 or Sanfilippo type D	Autosomal recessive
	GPC3	312870	Simpson-Golabi-Behmel syndrome type 1	X-linked
	GRN	138945	Neuronal ceroid lipofuscinosis 11; Frontotemporal lobar degeneration with ubiquitin-positive inclusions	Autosomal dominant and Autosomal recessive
	GUSB	253220	Mucopolysaccharidosis VII or Sly syndrome	Autosomal recessive
	HEXA	606869	GM2-gangliosidosis or Tay-Sachs disease	Autosomal recessive
	HEXB	268800	Sandhoff disease	Autosomal recessive
	HGSNAT	252930	Mucopolysaccharidosis IIIC or Sanfilippo type C	Autosomal recessive
	HYAL1	601492	Mucopolysaccharidosis IX	Autosomal recessive
	IDS	309900	Mucopolysaccharidosis II or Hunter syndrome	X-linked

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Lysosomal Disorders	*IDUA*	607014	Mucopolysaccharidosis I or Hurler/Scheie syndrome	Autosomal recessive
	LAMP2	300257	Danon disease	X-linked
	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 type I; Schindler disease type II or Kanzaki disease; Schindler disease type III	Autosomal recessive
	NAGLU	252920	Mucopolysaccharidosis IIIB or 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	Combined prosaposin deficiency; Krabbe disease due to saposin A deficiency; Metachromatic leukodystrophy due to saposin B deficiency; Gaucher disease due to saposin C deficiency	Autosomal recessive
	SCARB2	254900	Progressive myoclonic epilepsy 4; Action myoclonus-renal failure syndrome	Autosomal recessive
	SGSH	252900	Mucopolysaccharidosis IIIA or Sanfilippo type A	Autosomal recessive
	SLC17A5	604322	Free sialic acid storage disease; Salla disease	Autosomal recessive
	SMPD1	607608	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; Autosomal recessive spinocerebellar ataxia type 7	Autosomal recessive
	VPS33A	617303	Mucopolysaccharidosis-plus syndrome	Autosomal recessive
Peroxisomal Disorders	*ABCD1*	300100	X-linked Adrenoleukodystrophy	X-linked
	ACOX1	264470	Peroxisomal acyl-CoA oxidase deficiency	Autosomal recessive
	AGPS	600121	Rhizomelic chondrodysplasia punctata type 3	Autosomal recessive
	AGXT	259900	Primary hyperoxaluria	Autosomal recessive
	AMACR	614307	Alpha-methylacyl-CoA racemase deficiency	Autosomal recessive
	DNM1L	614388	DNM1L-associated leukoencephalopathy	Autosomal dominant and Autosomal recessive
	GNPAT	222765	Rhizomelic chondrodysplasia punctata type 2	Autosomal recessive
	HSD17B4	601860	D-bifunctional protein deficiency; Perrault syndrome 1	Autosomal recessive
	PEX1	602136	Zellweger spectrum disorder; Heimler syndrome 1	Autosomal recessive
	PEX2	614866	Zellweger spectrum disorder	Autosomal recessive
	PEX3	617370	Zellweger spectrum disorder	Autosomal recessive
	PEX5	600414	Zellweger spectrum disorder; Rhizomelic chondrodysplasia punctata type 5	Autosomal recessive

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	Gene Name	OMIM #	Associated Disorder(s)	Inheritance Pattern
Peroxisomal Disorders	*PEX6*	601498	Zellweger spectrum disorder; Heimler syndrome 2	Autosomal recessive
	PEX7	614879	Rhizomelic chondrodysplasia punctata; Refusm syndrome	Autosomal recessive
	PEX10	614870	Zellweger spectrum disorder	Autosomal recessive
	PEX11B	614920	Zellweger spectrum disorder	Autosomal recessive
	PEX12	614859	Zellweger spectrum disorder	Autosomal recessive
	PEX13	614883	Zellweger spectrum disorder	Autosomal recessive
	PEX14	614887	Zellweger spectrum disorder	Autosomal recessive
	PEX16	614876	Zellweger spectrum disorder	Autosomal recessive
	PEX19	614886	Zellweger spectrum disorder	Autosomal recessive
	PEX26	614872	Zellweger spectrum disorder	Autosomal recessive
	PHYH	266500	Adult Refsum syndrome	Autosomal recessive
	SCP2	604105	Sterol carrier protein deficiency	Autosomal recessive
	TRIM37	253250	Mulibrey nanism	Autosomal recessive

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