

COMBINED LYSOSOMAL AND PEROXISOMAL DISORDERS

GENE LIST (~82)

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

	Gene Name	OMIM #	Associated Disorder(s)	Inheritance Pattern
Lysosomal Disorders	<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 or 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	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	Autosomal recessive
	<i>CTSF</i>	615362	Neuronal ceroid lipofuscinosis 13	Autosomal recessive
	<i>DNAJC5</i>	162350	Neuronal ceroid lipofuscinosis 4B	Autosomal recessive
	<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	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 or Morquio syndrome; GM1-gangliosidosis	Autosomal recessive
	<i>GNE</i>	603824	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 or Sanfilippo type D	Autosomal recessive
	<i>GPC3</i>	312870	Simpson-Golabi-Behmel syndrome type 1	X-linked
	<i>GUSB</i>	253220	Mucopolysaccharidosis VII or Sly syndrome	Autosomal recessive
	<i>HEXA</i>	606869	GM2-gangliosidosis or Tay-Sachs disease	Autosomal recessive
	<i>HEXB</i>	268800	Sandhoff disease	Autosomal recessive
	<i>HGSNAT</i>	252930	Mucopolysaccharidosis IIIC or Sanfilippo type C	Autosomal recessive
<i>HYAL1</i>	601492	Mucopolysaccharidosis IX	Autosomal recessive	
<i>IDS</i>	309900	Mucopolysaccharidosis II or Hunter syndrome	X-linked	
<i>IDUA</i>	607014	Mucopolysaccharidosis I or Hurler/Scheie syndrome	Autosomal recessive	
<i>LAMP2</i>	300257	Danon disease	X-linked	

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Lysosomal Disorders	<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	Mucopolipidosis IV	Autosomal recessive
	<i>MFSD8</i>	610951	Neuronal ceroid lipofuscinosis 7	Autosomal recessive
	<i>NAGA</i>	609241	Schindler disease type I; Schindler disease type II or Kanzaki disease; Schindler disease type III	Autosomal recessive
	<i>NAGLU</i>	252920	Mucopolysaccharidosis IIIB or 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	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
	<i>SCARB2</i>	254900	Progressive myoclonic epilepsy 4; Action myoclonus-renal failure syndrome	Autosomal recessive
	<i>SGSH</i>	252900	Mucopolysaccharidosis IIIA or Sanfilippo type A	Autosomal recessive
	<i>SLC17A5</i>	604322	Free sialic acid storage disease; Salla disease	Autosomal recessive
	<i>SMPD1</i>	607608	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; Autosomal recessive spinocerebellar ataxia type 7	Autosomal recessive
<i>VPS33A</i>	617303	Mucopolysaccharidosis-plus syndrome	Autosomal recessive	
Peroxisomal Disorders	<i>ABCD1</i>	300100	X-linked Adrenoleukodystrophy	X-linked
	<i>ACOX1</i>	264470	Peroxisomal acyl-CoA oxidase deficiency	Autosomal recessive
	<i>AGPS</i>	600121	Rhizomelic chondrodysplasia punctata type 3	Autosomal recessive
	<i>AMACR</i>	614307	Alpha-methylacyl-CoA racemase deficiency	Autosomal recessive
	<i>DNM1L</i>	614388	DNM1L-associated leukoencephalopathy	Autosomal dominant and Autosomal recessive
	<i>FAR1</i>	616154	Peroxisomal fatty acyl-CoA reductase 1 disorder	Autosomal recessive
	<i>GNPAT</i>	222765	Rhizomelic chondrodysplasia punctata type 2	Autosomal recessive
	<i>HSD17B4</i>	601860	D-bifunctional protein deficiency; Perrault syndrome 1	Autosomal recessive
	<i>PEX1</i>	602136	Zellweger spectrum disorder; Heimler syndrome 1	Autosomal recessive
	<i>PEX2</i>	614866	Zellweger spectrum disorder	Autosomal recessive
	<i>PEX3</i>	617370	Zellweger spectrum disorder	Autosomal recessive
	<i>PEX5</i>	600414	Zellweger spectrum disorder; Rhizomelic chondrodysplasia punctata type 5	Autosomal recessive
	<i>PEX6</i>	601498	Zellweger spectrum disorder; Heimler syndrome 2	Autosomal recessive
	<i>PEX7</i>	614879	Rhizomelic chondrodysplasia punctata; Refsum syndrome	Autosomal recessive

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Peroxisomal Disorders	<i>PEX10</i>	614870	Zellweger spectrum disorder	Autosomal recessive
	<i>PEX11B</i>	614920	Zellweger spectrum disorder	Autosomal recessive
	<i>PEX12</i>	614859	Zellweger spectrum disorder	Autosomal recessive
	<i>PEX13</i>	614883	Zellweger spectrum disorder	Autosomal recessive
	<i>PEX14</i>	614887	Zellweger spectrum disorder	Autosomal recessive
	<i>PEX16</i>	614876	Zellweger spectrum disorder	Autosomal recessive
	<i>PEX19</i>	614886	Zellweger spectrum disorder	Autosomal recessive
	<i>PEX26</i>	614872	Zellweger spectrum disorder	Autosomal recessive
	<i>PHYH</i>	266500	Adult Refsum syndrome	Autosomal recessive
	<i>SCP2</i>	604105	Sterol carrier protein deficiency	Autosomal recessive
	<i>TRIM37</i>	253250	Mulibrey nanism	Autosomal recessive