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# Gene List – lysoSEEK™ Lysosomal Storage Disorder Panel

## By Category & Gene

| Gene | Disease | Category |
|---|---|---|
| ATP7A | Menke Disease | Copper Storage Disorders |
| ATP7B | Wilson Disease | Copper Storage Disorders |
| AGA | Aspartylglucosaminuria | Lysosomal Storage Diseases |
| ARSA | Metachromatic Leukodystrophy | Lysosomal Storage Diseases |
| ASAH1 | Farber Lipogranulomatosis | Lysosomal Storage Diseases |
| CTNS | Cystinosis | Lysosomal Storage Diseases |
| CTSA | Galactosialidosis | Lysosomal Storage Diseases |
| FUCA1 | Fucosidosis | Lysosomal Storage Diseases |
| GAA | Pompe Disease | Lysosomal Storage Diseases |
| GALC | Krabbe Disease | Lysosomal Storage Diseases |
| GBA | Gaucher Disease Types 1, 2, & 3; Cardiovascular & Neonatal Lethal Forms | Lysosomal Storage Diseases |
| GLA | Fabry Disease | Lysosomal Storage Diseases |
| GLB1 | GM1 Gangliosidosis | Lysosomal Storage Diseases |
| GM2A | GM2 Gangliosidosis Type AB | Lysosomal Storage Diseases |
| GNE | Sialuria; Autosomal Recessive Inclusion Body Myopathy; Nonaka Myopathy | Lysosomal Storage Diseases |
| GNPTAB | Mucolipidoses II (I-Cell Disease) & III (Pseudo-Hurler Polydystrophy) | Lysosomal Storage Diseases |
| GNPTG | Mucolipidosis III Gamma | Lysosomal Storage Diseases |
| LIPA | Wolman Disease | Lysosomal Storage Diseases |
| MAN2B1 | α-Mannosidosis | Lysosomal Storage Diseases |
| MANBA | β-Mannosidosis | Lysosomal Storage Diseases |
| NEU1 | Sialidosis | Lysosomal Storage Diseases |
| NPC1 | Niemann-Pick Disease Type C | Lysosomal Storage Diseases |
| NPC2 | Niemann-Pick Disease Type C2 | Lysosomal Storage Diseases |
| PSAP | Metachromatic Leukodystrophy (Prosaposine Deficiency) | Lysosomal Storage Diseases |
| SLC17A5 | Sialic Acid Storage Disease; Salla Disease | Lysosomal Storage Diseases |
| SMPD1 | Niemann-Pick Disease Types A & B | Lysosomal Storage Diseases |
| SUMF1 | Multiple Sulfatase Deficiency | Lysosomal Storage Diseases |
| ARSB | Mucopolysaccharidosis VI (Maroteaux-Lamy Syndrome) | Mucopolysaccharidoses |

| Gene | Disease | Category |
|---|---|---|
| GALNS | Mucopolysaccharidosis IV-A (Morquio Syndrome A) | Mucopolysaccharidoses |
| GNS | Mucopolysaccharidosis III-D (Sanfilippo D) | Mucopolysaccharidoses |
| GUSB | Mucopolysaccharidosis VII | Mucopolysaccharidoses |
| HEXA | Tay-Sachs Disease (GM2 Gangliosidosis Type B) | Mucopolysaccharidoses |
| HEXB | Sandhoff Disease (GM2 Gangliosidosis Type O) | Mucopolysaccharidoses |
| HGSNAT | Mucopolysaccharidosis III-C (Sanfilippo C) | Mucopolysaccharidoses |
| HYAL1 | Mucopolysaccharidosis IX | Mucopolysaccharidoses |
| IDS | Mucopolysaccharidosis II (Hunter Disease) | Mucopolysaccharidoses |
| IDUA | Mucopolysaccharidosis I-H, I-S, & I-HS (Hurler, Scheie, & Hurler-Sheie Syndromes) | Mucopolysaccharidoses |
| NAGA | Schindler Disease; Kanzaki Disease | Mucopolysaccharidoses |
| NAGLU | Mucopolysaccharidosis III-B (Sanfilippo B) | Mucopolysaccharidoses |
| SGSH | Mucopolysaccharidosis III-A (Sanfilippo A) | Mucopolysaccharidoses |
| ATP13A2 | Neuronal Ceroid Lipofuscinosis 12 (CLN 12) - Juvenile NCL | Neuronal Ceroid Lipofuscinoses |
| CLN3 | Neuronal Ceroid Lipofuscinosis 3 (CLN 3) - Classic Juvenile NCL ("Spielmeyer-Sjögren"), Adult NCL (Kufs Disease) | Neuronal Ceroid Lipofuscinoses |
| CLN5 | Neuronal Ceroid Lipofuscinosis 5 (CLN 5) - Late Infantile NCL ("Finnish Variant"), Juvenile NCL, Adult NCL (Kufs Disease) | Neuronal Ceroid Lipofuscinoses |
| CLN6 | Neuronal Ceroid Lipofuscinosis 6 (CLN 6) - Late Infantile NCL ("Lake-Cavanagh or Indian Variant"), Adult NCL (Kufs Disease Type A) | Neuronal Ceroid Lipofuscinoses |
| CLN8 | Neuronal Ceroid Lipofuscinosis 8 (CLN 8) - Late Infantile NCL, Northern Epilepsy (Progressive Epilepsy With Mental Retardation or EPMR) | Neuronal Ceroid Lipofuscinoses |
| CTSD | Neuronal Ceroid Lipofuscinosis 10 (CLN 10) - Congenital NCL, Late-Infantile NCL, or Teenage-/Adult-Onset NCL | Neuronal Ceroid Lipofuscinoses |
| CTSF | Neuronal Ceroid Lipofuscinosis 13 (CLN 13) - Adult NCL (Kufs Disease Type B) | Neuronal Ceroid Lipofuscinoses |
| DNAJC5 | Neuronal Ceroid Lipofuscinosis 4 (CLN 4) - Adult NCL (Parry Disease) | Neuronal Ceroid Lipofuscinoses |
| GRN | Neuronal Ceroid Lipofuscinosis 11 (CLN 11) - Adult NCL (Kufs Disease) | Neuronal Ceroid Lipofuscinoses |
| KCTD7 | Neuronal Ceroid Lipofuscinosis 14 (CLN 14) - Late Infantile NCL; Progressive Myoclonic Epilepsy 3 | Neuronal Ceroid Lipofuscinoses |
| MFSD8 | Neuronal Ceroid Lipofuscinosis 7 (CLN7) - Late Infantile NCL ("Turkish Variant") | Neuronal Ceroid Lipofuscinoses |
| PPT1 | Neuronal Ceroid Lipofuscinosis 1 (CLN 1) - Infantile Classic NCL ("Haltia-Santavuori" Type), Late Infantile NCL, Juvenile NCL, Adult NCL (Kufs Disease) | Neuronal Ceroid Lipofuscinoses |
| TPP1 | Neuronal Ceroid Lipofuscinosis 2 (CLN 2) - Late Infantile Classic NCL ("Janský-Bielschowsky" Type), Juvenile NCL, Possibly Later-Onset Type NCL | Neuronal Ceroid Lipofuscinoses |
| DBH | Dopamine Beta-Hydroxylase Deficiency | Neurotransmitter Disorders |
| DDC | Aromatic L-Amino Acid Decarboxylase Deficiency | Neurotransmitter Disorders |
| GCH1 | Dopa-Responsive Dystonia | Neurotransmitter Disorders |
| MAOA | Brunner Syndrome | Neurotransmitter Disorders |
| PTS | Tetrahydrobiopterin Deficiency | Neurotransmitter Disorders |
| SPR | Dopa-Responsive Dystonia | Neurotransmitter Disorders |
| TH | Dopa-Responsive Dystonia | Neurotransmitter Disorders |
| AMT | Non-Ketotic Hyperglycinemia | Non-Ketotic Hyperglycinemia |

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|---|---|---|
| GCSH | Non-Ketotic Hyperglycinemia | Non-Ketotic Hyperglycinemia |
| GLDC | Non-Ketotic Hyperglycinemia | Non-Ketotic Hyperglycinemia |
| PEX1 | Zellweger; Peroxisome Biogenesis Disorder 1A | Peroxisome Biogenesis Disorders |
| PEX10 | Zellweger; Peroxisome Biogenesis Disorder 6A | Peroxisome Biogenesis Disorders |
| PEX12 | Peroxisome Biogenesis Disorder 3B | Peroxisome Biogenesis Disorders |
| PEX13 | Zellweger; Peroxisome Biogenesis Disorder 11A | Peroxisome Biogenesis Disorders |
| PEX14 | Zellweger; Peroxisome Biogenesis Disorder 13A | Peroxisome Biogenesis Disorders |
| PEX16 | Peroxisome Biogenesis Disorder 8B | Peroxisome Biogenesis Disorders |
| PEX19 | Zellweger; Peroxisome Biogenesis Disorder 12A | Peroxisome Biogenesis Disorders |
| PEX26 | Peroxisome Biogenesis Disorder 7B | Peroxisome Biogenesis Disorders |
| PEX3 | Zellweger; Peroxisome Biogenesis Disorder 10A | Peroxisome Biogenesis Disorders |
| PEX5 | Peroxisome Biogenesis Disorder 2B | Peroxisome Biogenesis Disorders |
| PEX6 | Zellweger; Peroxisome Biogenesis Disorder 4A, 4B | Peroxisome Biogenesis Disorders |
| PHYH | Refsum Disease | Peroxisome Biogenesis Disorders |
| FKRP | Walker-Warburg Syndrome | Walker-Warburg Syndrome |
| FKTN | Walker-Warburg Syndrome | Walker-Warburg Syndrome |
| ISPD | Walker-Warburg Syndrome | Walker-Warburg Syndrome |
| LARGE | Walker-Warburg Syndrome | Walker-Warburg Syndrome |
| POMT1 | Walker-Warburg Syndrome | Walker-Warburg Syndrome |
| POMT2 | Walker-Warburg Syndrome | Walker-Warburg Syndrome |
| ALDH7A1 | Pyridoxin(Pyridoxal ) Dependant Seizures | Genes in Differential Dx |
| ASPA | Canavan Disease | Genes in Differential Dx |
| CYP27A1 | Cerebrotendinous Xanthomatosis | Genes in Differential Dx |
| DHCR7 | Smith-Lemli-Opitz Syndrome | Genes in Differential Dx |
| GFAP | Alexander Disease | Genes in Differential Dx |
| HPRT1 | Lesch-Nyhan Syndrome | Genes in Differential Dx |
| L1CAM | L1 Syndrome | Genes in Differential Dx |
| MECP2 | Rett Syndrome | Genes in Differential Dx |
| PANK2 | Pantothenat Kinase-Associated Neurodegeneration | Genes in Differential Dx |
| PLP1 | Pelizaeus-Merzbacher Disease | Genes in Differential Dx |
| PNPO | Pyridoxamine 5'-phosphate oxidase deficiency | Genes in Differential Dx |
| POLG | Alper's Disease | Genes in Differential Dx |
| POMGNT1 | Limb-Girdle Muscular Dystrophy | Genes in Differential Dx |
| RPS6KA3 | Coffin-Lawry Syndrome | Genes in Differential Dx |

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| ARSB | Mucopolysaccharidosis VI (Maroteaux-Lamy Syndrome) | Mucopolysaccharidoses |
| ASAH1 | Farber Lipogranulomatosis | Lysosomal Storage Diseases |
| ASPA | Canavan Disease | Genes in Differential Dx |
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| DBH | Dopamine Beta-Hydroxylase Deficiency | Neurotransmitter Disorders |
| DDC | Aromatic L-Amino Acid Decarboxylase Deficiency | Neurotransmitter Disorders |
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| ISPD | Walker-Warburg Syndrome | Walker-Warburg Syndrome |
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| PNPO | Pyridoxamine 5'-phosphate oxidase deficiency | Genes in Differential Dx |
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| POMT1 | Walker-Warburg Syndrome | Walker-Warburg Syndrome |
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