Oslo universitetssykehus
Avdeling for medisinsk genetikk

Mitokondriesykdommer

Genpanel, versjon v03

* Enkelte genomiske regioner har lav eller ingen sekvensdekning ved eksomsekvensering. Dette skyldes at de har stor likhet med andre områder i genomet, slik at spesifikk gjenkjennelse av disse områdene og påvisning av varianter i disse områdene, blir vanskelig og upålitelig. Disse genetiske regionene har vi identifisert ved å benytte USCS segmental duplication hvor områder større enn 1 kb og ≥90% likhet med andre regioner i genomet , gjenkjennes (https://genome.ucsc.edu).

For noen gener ligger alle ekson i områder med segmentale duplikasjoner, så disse vil ikke bli analysert: **ATAD3A, CA5A, CYCS, MSTO1, SDHA**

Vi gjør oppmerksom på at ved identifiseringav ekson oppstrøms for startkodon kan eksonnummereringen endres uten at transkript ID endres.

Avdelingens websider har en full oversikt over områder som er utelatt pga. segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
AARS	20	NM_001605.2		2-21	Charcot-Marie-Tooth disease, axonal, type 2N OMIM Epileptic encephalopathy, early infantile, 29 OMIM
AARS2	21022	NM_020745.3		1-22	Mitochondrial alanyl-tRNA synthetase deficiency OMIM Combined oxidative phosphorylation deficiency type 8; progressive leukoencephalopathy with ovarian failure OMIM
ABAT	23	NM_020686.5		2-16	GABA transaminase deficiency OMIM
ABCB7	48	NM_004299.6		1-16	ABCB7 deficiency OMIM Sideroblastic anemia and spinocerebellar ataxia OMIM

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ACAD9	21497	NM_014049.4		1-18	ACAD9 deficiency OMIM
ACO2	118	NM_001098.2		1-18	Mitochondrial aconitase deficiency OMIM Infantile cerebellar-retinal degeneration OMIM
ADCK3	16812	NM_020247.4		2-15	COQ8A deficiency OMIM Primary coenzyme Q10 deficiency type 4; ADCK3 deficiency OMIM
ADCK4	19041	NM_024876.3		2-15	COQ8B deficiency OMIM Nephrotic syndrome type 9; ADCK4 deficiency OMIM
AFG3L2	315	NM_006796.2	14	1-17	m-AAA protease AFG3L2 subunit deficiency OMIM Autosomal recessive spastic ataxia type 5; spinocerebellar ataxia type 28 OMIM
AGK	21869	NM_018238.3	16	2-16	Acylglycerol kinase deficiency OMIM Sengers syndrome OMIM
AIFM1	8768	NM_004208.3		1-16	AIFM1 deficiency OMIM Combined oxidative phosphorylation deficiency type 6; Cowchock syndrome OMIM
ANO10	25519	NM_018075.4		2-13	Spinocerebellar ataxia, autosomal recessive 10 OMIM
APOPT1	20492	NM_032374.4		1-5	APOPT1 deficiency OMIM
APTX	15984	NM_175073.2		3-9	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia OMIM
ATAD3A	25567	NM_001170535.2	1-16	1-16	ATAD3A deficiency OMIM Harel-Yoon syndrome OMIM
ATP5A1	823	NM_001001937.1		2-13	Mitochondrial ATP synthase F1 subunit a deficiency OMIM
ATP5D	837	NM_001687.4		1-4	Mitochondrial ATP synthase F1 subunit ? deficiency OMIM

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ATP5E	838	NM_006886.3		1-2	Mitochondrial ATP synthase F1 subunit e deficiency OMIM
ATPAF2	18802	NM_145691.3		1-8	Mitochondrial ATP synthase F1 assembly factor 2 deficiency OMIM
BCS1L	1020	NM_004328.4		3-9	BCS1L deficiency OMIM GRACILE syndrome; Bjornstad syndrome OMIM
BOLA3	24415	NM_212552.2		1-4	BOLA3 deficiency OMIM Multiple mitochondrial dysfunctions syndrome type 2 with hyperglycinemia OMIM
C10orf2	1160	NM_021830.4		1-5	TWINKLE mitochondrial DNA helicase deficiency OMIM Mitochondrial DNA depletion syndrome type 7; Perrault syndrome+type 5; arPEO with mitochondrial DNA deletions type 5 OMIM
C11orf83	34399	NM_001085372.2		1-2	UQCC3 deficiency OMIM
C12orf65	26784	NM_152269.4		2-3	C12orf65 release factor deficiency OMIM Combined oxidative phosphorylation+deficiency+type 7; autosomal recessive spastic paraplegia type 55 OMIM
C19orf12	25443	NM_001031726.3		1-3	?Spastic paraplegia 43, autosomal recessive OMIM Neurodegeneration with brain iron accumulation 4 OMIM
C19orf70	33702	NM_205767.2		1-4	MICOS complex subunit MIC13 deficiency OMIM QIL1 deficiency OMIM
C1QBP	1243	NM_001212.3		1-6	C1q binding protein deficiency OMIM Combined oxidative phosphorylation deficiency+type 33 OMIM
CA5A	1377	NM_001739.1	1-7	1-7	Carbonic anhydrase VA deficiency OMIM

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CARS2	25695	NM_024537.3		1-15	Mitochondrial cysteinyl-tRNA synthetase deficiency OMIM Combined oxidative phosphorylation deficiency type 27 OMIM
CEP89	25907	NM_032816.4		1-19	CEP89 deficiency OMIM
CHCHD10	15559	NM_213720.2		1-4	?Myopathy, isolated mitochondrial, autosomal dominant OMIM Frontotemporal dementia and/or amyotrophic lateral sclerosis 2 OMIM Spinal muscular atrophy, Jokela type OMIM
CHKB	1938	NM_005198.4		1-11	Choline kinase ? deficiency OMIM Congenital muscular dystrophy, megaconial type OMIM
CISD2	24212	NM_001008388.4	3	1-3	Wolfram syndrome 2 OMIM
CLPB	30664	NM_001258394.2		1-18	CLPB deficiency OMIM 3-methylglutaconic aciduria type 7, with cataracts, neurologic involvement and neutropenia OMIM
CLPP	2084	NM_006012.2		1-6	CLPP deficiency OMIM Perrault syndrome type 3 OMIM
CMPK2	27015	NM_207315.3		1-5	Mitochondrial UMP-CMP+kinase+2 deficiency OMIM
COA3	24990	NM_001040431.2		1-2	Cytochrome c oxidase assembly factor 3 deficiency OMIM
COA5	33848	NM_001008215.2		1-3	Cytochrome c oxidase assembly factor 5 deficiency OMIM
COA6	18025	NM_001206641.2		1-3	Cytochrome c oxidase assembly factor 6 deficiency OMIM
COA7	25716	NM_023077.2		1-3	Cytochrome c oxidase assembly factor 7 deficiency OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
COASY	29932	NM_025233.6		1-9	Coenzyme A synthase deficiency OMIM Coenzyme A synthase protein-associated neurodegeneration (CoPAN); neurodegeneration with brain iron accumulation type 6 OMIM
COQ2	25223	NM_015697.8		1-7	COQ2 deficiency OMIM Primary coenzyme Q10 deficiency type 1 OMIM
COQ4	19693	NM_016035.4		1-7	COQ4 deficiency OMIM Primary coenzyme Q10 deficiency type 7 OMIM
COQ5	28722	NM_032314.3		1-7	COQ5 deficiency OMIM
COQ6	20233	NM_182476.2		1-12	COQ6 deficiency OMIM Primary coenzyme Q10 deficiency type 6 OMIM
COQ7	2244	NM_016138.4		1-6	COQ7 deficiency OMIM Primary coenzyme Q10 deficiency type 8 OMIM
COQ9	25302	NM_020312.3		1-9	COQ9 deficiency OMIM Primary coenzyme Q10 deficiency type 5 OMIM
COX10	2260	NM_001303.3	6	1-7	COX10 deficiency OMIM
COX14	28216	NM_032901.3		2	Cytochrome c oxidase assembly factor 14 deficiency OMIM
COX15	2263	NM_004376.6		1-9	COX15 deficiency OMIM
COX20	26970	NM_198076.5		1-4	COX20 deficiency OMIM
COX4I2	16232	NM_032609.2		2-5	Cytochrome c oxidase subunit 4I2 deficiency OMIM Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis OMIM

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COX6A1	2277	NM_004373.3		1-3	Cytochrome c oxidase subunit 6A1 deficiency OMIM Recessive intermediate Charcot-Marie-Tooth disease type D OMIM
COX6B1	2280	NM_001863.4		2-4	Cytochrome c oxidase subunit 6B1 deficiency OMIM
COX7B	2291	NM_001866.2		1-3	Cytochrome c oxidase subunit 7B deficiency OMIM Linear skin defects with multiple congenital anomalies type 2 OMIM
COX8A	2294	NM_004074.2		1-2	Cytochrome c oxidase subunit 8A deficiency OMIM
CYC1	2579	NM_001916.4		1-7	Mitochondrial cytochrome c1 deficiency OMIM
CYCS	19986	NM_018947.5	2-3	2-3	Mitochondrial cytochrome c deficiency OMIM Thrombocytopenia type 4 OMIM
DARS	2678	NM_001349.3		1-16	Hypomyelination with brainstem and spinal cord involvement and leg spasticity OMIM
DARS2	25538	NM_018122.4		1-17	Mitochondrial aspartyl-tRNA synthetase deficiency OMIM Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation OMIM
DCAF17	25784	NM_025000.3		1-14	Woodhouse-Sakati syndrome OMIM
DGUOK	2858	NM_080916.2		1-7	Mitochondrial deoxyguanosine kinase deficiency OMIM Mitochondrial DNA depletion syndrome type 3; arPEO with mitochondrial DNA deletions type 4; noncirrhotic portal hypertension+ OMIM
DHTKD1	23537	NM_018706.6		1-17	2-aminoadipic 2-oxoadipic+aciduria OMIM Charcot-Marie-Tooth disease type 2Q OMIM

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DLAT	2896	NM_001931.4		1-14	Dihydrolipoamide acetyltransferase+deficiency OMIM Pyruvate dehydrogenase E2 deficiency OMIM
DLD	2898	NM_000108.4		1-14	Dihydrolipoamide dehydrogenase deficiency OMIM E3 deficiency OMIM
DNA2	2939	NM_001080449.2		1-21	DNA2 helicase deficiency OMIM adPEO with mitochondrial DNA deletions type 6 OMIM
DNAJC19	30528	NM_145261.3		1-6	DNAJC19 deficiency OMIM Dilated cardiomyopathy with ataxia (DCMA syndrome); 3-methylglutaconic aciduria type 5 OMIM
DNM1L	2973	NM_012062.4		1-20	Dynamin-like protein 1 deficiency OMIM Optic atrophy type 5; encephalopathy due to defective mitochondrial and peroxisomal fission type 1 OMIM
E4F1	3121	NM_004424.4		1-14	?Leigh disease PubMed
EARS2	29419	NM_001083614.1		1-9	Mitochondrial glutamyl-tRNA synthetase deficiency OMIM Combined oxidative phosphorylation deficiency type 12 OMIM
ECHS1	3151	NM_004092.3		1-8	Mitochondrial short-chain enoyl-CoA hydratase 1+deficiency OMIM Crotonase deficiency OMIM
EHHADH	3247	NM_001966.3		1-7	L-bifunctional protein deficiency OMIM Fanconi renotubular syndrome type 3 OMIM
ELAC2	14198	NM_018127.6		1-24	Ribonuclease Z 3' tRNA processing enzyme deficiency OMIM Combined oxidative phosphorylation deficiency 17 OMIM

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EPG5	29331	NM_020964.3		1-44	EPG5 deficiency OMIM Vici syndrome OMIM
ETHE1	23287	NM_014297.4		1-7	Mitochondrial sulfur dioxygenase deficiency OMIM Ethylmalonic encephalopathy OMIM
FARS2	21062	NM_006567.4		2-7	Mitochondrial phenylalanyl-tRNA synthetase deficiency OMIM Combined oxidative phosphorylation deficiency type 14 OMIM
FASTKD2	29160	NM_014929.3		2-12	FASTKD2 deficiency OMIM
FBXL4	13601	NM_012160.4		3-9	FBXL4 deficiency OMIM Mitochondrial DNA depletion syndrome type 13 OMIM
FDX1L	30546	NM_001031734.3		1-5	Ferredoxin 2 deficiency OMIM
FDXR	3642	NM_001258014.3		1-12	Ferredoxin reductase deficiency OMIM Auditory neuropathy and optic atrophy OMIM
FH	3700	NM_000143.3		1-10	Fumarate hydratase deficiency OMIM Fumarase deficiency OMIM Fumarate hydratase deficiency, tumoral phenotype OMIM Reed syndrome OMIM
FOXRED1	26927	NM_017547.3		1-11	FOXRED1 deficiency OMIM
FTL	3999	NM_000146.3		1-4	Ferritin light chain deficiency OMIM Hereditary L-ferritin deficiency OMIM Ferritin light chain superactivity OMIM Neuroferritinopathy; neurodegeneration with brain iron accumulation 3 OMIM Ferritin light chain dysregulation OMIM Hyperferritinemia-cataract syndrome OMIM

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FXN	3951	NM_000144.4	5	1-5	Frataxin deficiency OMIM Friedreich ataxia OMIM
GARS	4162	NM_002047.3		1-17	Mitochondrial and cytoplasmic glycil-tRNA synthetase deficiency OMIM Charcot-Marie-Tooth disease type 2D; distal hereditary motor neuronopathy+type 5A OMIM
GATC	25068	NM_176818.2	4	1-4	Mitochondrial glutamyl-tRNA(Gln) amidotransferase subunit C deficiency OMIM
GDAP1	15968	NM_018972.3		1-6	GDAP1 deficiency OMIM Axonal Charcot-Marie-Tooth type 2K; demyelinating Charcot-Marie-Tooth disease type 4A OMIM
GFER	4236	NM_005262.2		1-3	GFER deficiency OMIM
GFM1	13780	NM_024996.5		1-18	Mitochondrial elongation factor G1 deficiency OMIM Combined oxidative phosphorylation+deficiency+type 1 OMIM
GFM2	29682	NM_032380.4		2-21	Mitochondrial elongation factor G2 deficiency OMIM
GLRX5	20134	NM_016417.2		1-2	Glutaredoxin 5 deficiency OMIM
GOT2	4433	NM_002080.3	10	1-10	Mitochondrial aspartate aminotransferase deficiency OMIM
GPD1	4455	NM_005276.3		1-8	Cytosolic glycerol-3-phosphate dehydrogenase deficiency OMIM Transient infantile hypertriglyceridemia OMIM
GTPBP3	14880	NM_133644.3		1-8	tRNA 5-taurinomethyluridine modifier deficiency OMIM Combined oxidative phosphorylation deficiency type 23 OMIM

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HARS2	4817	NM_012208.3		1-13	Mitochondrial histidyl-tRNA synthetase deficiency OMIM Perrault syndrome type 2 OMIM
HCCS	4837	NM_005333.4		2-7	Holocytochrome c synthase deficiency OMIM Linear skin defects with multiple congenital anomalies type 1 OMIM
HIBCH	4908	NM_014362.3		1-14	3-hydroxyisobutyryl-CoA hydrolase+deficiency OMIM ?-hydroxyisobutyryl-CoA deacylase deficiency OMIM
HSPA9	5244	NM_004134.6		1-17	HSPA9 deficiency OMIM Sideroblastic anemia type 4; epiphyseal, vertebral, ear, nose, plus associated malformations (EVEN-plus) syndrome OMIM
HSPD1	5261	NM_002156.4	9-12	2-12	HSP60 deficiency OMIM Hypomyelinating leukodystrophy type 4 (recessive); autosomal dominant spastic paraplegia type 13 OMIM
HTRA2	14348	NM_013247.4		1-8	HTRA2 deficiency OMIM 3-methylglutaconic aciduria type 8 OMIM
IARS2	29685	NM_018060.3		1-23	Mitochondrial isoleucyl-tRNA synthetase deficiency OMIM Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia (CAGSSS) OMIM
IBA57	27302	NM_001010867.3		1-3	IBA57 deficiency OMIM
IDH2	5383	NM_002168.3		1-11	Mitochondrial NADH-dependent isocitrate dehydrogenase 2 superactivity OMIM D-2-hydroxyglutaric aciduria type 2 OMIM
IDH3B	5385	NM_006899.4		1-12	Mitochondrial NADPH-dependent isocitrate dehydrogenase 3 ? subunit deficiency OMIM

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ISCA1	28660	NM_030940.3	4	1-4	ISCA1 deficiency OMIM Multiple mitochondrial dysfunctions syndrome type 5 OMIM
ISCA2	19857	NM_194279.3		1-4	ISCA2 deficiency OMIM Multiple mitochondrial dysfunctions syndrome type 4 OMIM
ISCU	29882	NM_213595.3		1-5	ISCU deficiency OMIM Hereditary myopathy with lactic acidosis, Swedish type myopathy with exercise intolerance OMIM
KARS	6215	NM_001130089.1		2-15	Mitochondrial and cytoplasmic lysyl-tRNA synthetase deficiency OMIM
LARS2	17095	NM_015340.3		3-22	Mitochondrial leucyl-tRNA synthetase deficiency OMIM Perrault syndrome type 4 OMIM
LIAS	16429	NM_006859.3		1-11	Lipoic acid synthase deficiency OMIM Hyperglycinemia, lactic acidosis, and seizures OMIM
LIPT1	29569	NM_145199.2		2	Lipoyltransferase 1 deficiency OMIM
LIPT2	37216	NM_001144869.2		1-2	Lipoyltransferase 2 deficiency OMIM Neonatal severe encephalopathy with lactic acidosis and brain abnormalities (NELABA) OMIM
LONP1	9479	NM_001276480.1		3-18	LONP1 deficiency OMIM Cerebral, ocular, dental, auricular, and skeletal+(CODAS) syndrome OMIM
LRPPRC	15714	NM_133259.3		1-38	LRPPRC deficiency OMIM
LYRM4	21365	NM_020408.5		1-3	ISD11 deficiency OMIM
LYRM7	28072	NM_181705.3		1-5	LYRM7 deficiency OMIM

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MARS2	25133	NM_138395.3		1	Mitochondrial methionyl-tRNA synthetase deficiency OMIM Autosomal recessive spastic ataxia type 3 OMIM
MDH2	6971	NM_005918.3		1-9	Mitochondrial malate dehydrogenase deficiency OMIM Early infantile epileptic encephalopathy type 51+ OMIM Mitochondrial malate dehydrogenase deficiency, tumoral phenotype OMIM
MECR	19691	NM_001024732.3		2-10	Mitochondrial enoyl-CoA reductase deficiency OMIM Mitochondrial enoyl-CoA reductase protein-associated neurodegeneration (MEPAN) OMIM
MFF	24858	NM_020194.5		3-11	Mitochondrial fission factor deficiency OMIM Encephalopathy due to defective mitochondrial and peroxisomal fission type 2 OMIM
MFN2	16877	NM_014874.3		3-19	Mitofusin 2 deficiency OMIM Axonal Charcot-Marie-Tooth type 2A2 OMIM
MGME1	16205	NM_052865.3		2-5	Mitochondrial genome maintenance exonuclease 1+deficiency OMIM Mitochondrial DNA depletion syndrome type 11 OMIM
MICU1	1530	NM_006077.3		2-12	Mitochondrial calcium uniporter deficiency OMIM Myopathy with extrapyramidal signs OMIM
MIPEP	7104	NM_005932.3	1-3	1-19	Mitochondrial intermediate peptidase deficiency OMIM Combined oxidative phosphorylation deficiency+type 31 OMIM
MPC1	21606	NM_016098.3		1-5	Mitochondrial pyruvate carrier deficiency OMIM

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MPV17	7224	NM_002437.4		2-8	MPV17 deficiency OMIM Mitochondrial DNA depletion syndrome type 6 OMIM
MRPL12	10378	NM_002949.3		1-5	Mitochondrial ribosomal large subunit 12 deficiency OMIM
MRPL3	10379	NM_007208.3	10	1-10	Mitochondrial ribosomal large subunit 3 deficiency OMIM Combined oxidative phosphorylation+deficiency+type 9 OMIM
MRPL44	16650	NM_022915.3		1-4	Mitochondrial ribosomal large subunit 44 deficiency OMIM Combined oxidative phosphorylation+deficiency+type 16 OMIM
MRPS16	14048	NM_016065.3		1-3	Mitochondrial ribosomal small subunit 16 deficiency OMIM Combined oxidative phosphorylation+deficiency+type 2 OMIM
MRPS2	14495	NM_016034.4		1-4	Mitochondrial ribosomal small subunit 2 deficiency OMIM
MRPS22	14508	NM_020191.3		1-8	Mitochondrial ribosomal small subunit 22 deficiency OMIM Combined oxidative phosphorylation+deficiency+type 5 OMIM
MRPS23	14509	NM_016070.3		1-5	Mitochondrial ribosomal small subunit 23 deficiency OMIM
MRPS34	16618	NM_023936.1		1-3	Mitochondrial ribosomal small subunit 34 deficiency OMIM Combined oxidative phosphorylation+deficiency+type 32 OMIM
MRPS7	14499	NM_015971.3		1-5	Mitochondrial ribosomal small subunit 7 deficiency OMIM

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MSTO1	29678	NM_018116.3	1-14	1-14	MSTO1 deficiency OMIM Mitochondrial myopathy and ataxia OMIM
MTFMT	29666	NM_139242.3		1-9	Mitochondrial methionyl-tRNA formyltransferase deficiency OMIM Combined oxidative phosphorylation deficiency type 15 OMIM
MTO1	19261	NM_012123.3		1-12	tRNA 5-carboxymethylaminomethyl transferase deficiency OMIM Combined oxidative phosphorylation deficiency type 10 OMIM
MTPAP	25532	NM_018109.3		1-9	Mitochondrial poly(A) polymerase deficiency OMIM
NADK2	26404	NM_001287340.1		4-12	Mitochondrial NAD kinase 2 deficiency OMIM 2,4-dienoyl-CoA reductase deficiency with hyperlysinemia OMIM
NARS2	26274	NM_024678.5		1-14	Mitochondrial asparaginyl-tRNA synthetase deficiency OMIM Combined oxidative phosphorylation deficiency type 24 OMIM
NDUFA1	7683	NM_004541.3		1-3	NADH dehydrogenase ? subcomplex subunit 1 deficiency OMIM
NDUFA10	7684	NM_004544.3		1-10	NADH dehydrogenase ? subcomplex subunit 10 deficiency OMIM
NDUFA11	20371	NM_175614.4		1-4	NDUFA11 deficiency OMIM
NDUFA12	23987	NM_018838.4		1-4	NADH dehydrogenase ? subcomplex subunit 12 deficiency OMIM
NDUFA13	17194	NM_015965.6		1-5	NDUFA13 deficiency OMIM
NDUFA2	7685	NM_002488.4		1-3	NADH dehydrogenase ? subcomplex subunit 2 deficiency OMIM
NDUFA4	7687	NM_002489.3		1-4	NDUFA4 deficiency OMIM

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NDUFA9	7693	NM_005002.4		1-11	NADH dehydrogenase ? subcomplex subunit 9 deficiency OMIM
NDUFAF1	18828	NM_016013.3		2-5	NADH dehydrogenase ? subcomplex assembly factor 1 deficiency OMIM
NDUFAF2	28086	NM_174889.4		1-4	NADH dehydrogenase ? subcomplex assembly factor 2 deficiency OMIM
NDUFAF3	29918	NM_199069.1		1-5	NADH dehydrogenase ? subcomplex assembly factor 3 deficiency OMIM
NDUFAF4	21034	NM_014165.3	3	1-3	NADH dehydrogenase ? subcomplex assembly factor 4 deficiency OMIM
NDUFAF5	15899	NM_024120.4		1-11	NADH dehydrogenase ? subcomplex assembly factor 5 deficiency OMIM
NDUFAF6	28625	NM_152416.3		1-9	NADH dehydrogenase ? subcomplex assembly factor 6 deficiency OMIM
NDUFB11	20372	NM_001135998.2		1-3	NADH dehydrogenase ? subcomplex subunit 11 deficiency OMIM Linear skin defects with multiple congenital anomalies type 3 OMIM
NDUFB3	7698	NM_002491.2		2-3	NADH dehydrogenase ? subcomplex subunit 3 deficiency OMIM
NDUFB8	7703	NM_001284368.1		2-5	NADH dehydrogenase ? subcomplex subunit 8 deficiency OMIM
NDUFB9	7704	NM_005005.2		1-4	NDUFB9 deficiency OMIM
NDUFS1	7707	NM_005006.6		2-19	NADH dehydrogenase iron-sulfur protein 1 deficiency OMIM
NDUFS2	7708	NM_004550.4		2-15	NADH dehydrogenase iron-sulfur protein 2 deficiency OMIM
NDUFS3	7710	NM_004551.2		1-7	NADH dehydrogenase iron-sulfur protein 3 deficiency OMIM
NDUFS4	7711	NM_002495.3		1-5	NADH dehydrogenase iron-sulfur protein 4 deficiency OMIM
NDUFS6	7713	NM_004553.4		1-4	NADH dehydrogenase iron-sulfur protein 6 deficiency OMIM

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NDUFS7	7714	NM_024407.4		1-8	NADH dehydrogenase iron-sulfur protein 7 deficiency OMIM
NDUFS8	7715	NM_002496.3		2-7	NADH dehydrogenase iron-sulfur protein 8 deficiency OMIM
NDUFV1	7716	NM_007103.3		1-10	NADH dehydrogenase flavoprotein 1 deficiency OMIM
NDUFV2	7717	NM_021074.4		1-8	NADH dehydrogenase flavoprotein 2 deficiency OMIM
NFS1	15910	NM_021100.4		1-13	NFS1 deficiency OMIM
NFU1	16287	NM_001002755.2		1-8	NFU1 deficiency OMIM Multiple mitochondrial dysfunctions syndrome type 1 OMIM
NNT	7863	NM_012343.3		2-22	Nicotinamide nucleotide transhydrogenase+deficiency OMIM Glucocorticoid deficiency type 4 OMIM
NUBPL	20278	NM_025152.2		1-11	NUBPL deficiency OMIM
OAT	8091	NM_000274.3		2-10	Ornithine aminotransferase deficiency OMIM Gyrate atrophy of choroid and retina OMIM
OGDH	8124	NM_002541.3		2-23	?-ketoglutarate dehydrogenase deficiency OMIM
OPA1	11957	NM_015560.2		1-28	OPA1 deficiency OMIM Optic atrophy type 1 (dominant); Behr syndrome (recessive) OMIM
OPA3	8142	NM_025136.3		1-2	OPA3 deficiency OMIM Optic atrophy type 3 (dominant); 3-methylglutaconic aciduria type 3, Costeff syndrome (recessive) OMIM
OTC	8512	NM_000531.5		1-10	Ornithine transcarbamylase deficiency OMIM
OXCT1	8527	NM_000436.3		1-17	Succinyl-CoA:3-oxoacid-CoA transferase+deficiency OMIM

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PAM16	29679	NM_016069.10		1-5	MAGMAS deficiency OMIM Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type OMIM
PANK2	15894	NM_153638.3		1-7	Pantothenate kinase 2 deficiency OMIM Pantothenate kinase-associated neurodegeneration (PKAN); neurodegeneration with brain iron accumulation type 1 OMIM
PARK2	8607	NM_004562.2		1-12	Parkin deficiency OMIM Early-onset Parkinson disease type 2 OMIM
PARS2	30563	NM_152268.3		2	?Alpers syndrome PubMed
PC	8636	NM_000920.3		3-22	Pyruvate carboxylase deficiency OMIM
PCK2	8725	NM_004563.3		1-10	Mitochondrial phosphoenolpyruvate carboxykinase deficiency OMIM
PDE12	25386	NM_177966.6		1-3	Mitochondrial poly(A) exoribonuclease deficiency OMIM
PDHA1	8806	NM_000284.3		1-11	Pyruvate dehydrogenase E1-? deficiency OMIM
PDHB	8808	NM_000925.3		1-10	Pyruvate dehydrogenase E1-? deficiency OMIM
PDHX	21350	NM_003477.2		1-11	Pyruvate dehydrogenase E3-binding protein deficiency OMIM Pyruvate dehydrogenase component X deficiency OMIM
PDK3	8811	NM_001142386.2		1-12	Pyruvate dehydrogenase kinase isoenzyme 3 superactivity OMIM
PDP1	9279	NM_018444.3		2	Pyruvate dehydrogenase phosphatase deficiency OMIM
PDSS1	17759	NM_014317.4		1-12	Prenyl diphosphate synthase subunit+1 deficiency OMIM Primary coenzyme Q10 deficiency type 2 OMIM

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PDSS2	23041	NM_020381.3		1-8	Prenyl diphosphate synthase subunit+2 deficiency OMIM Primary coenzyme Q10 deficiency type 3 OMIM
PET100	40038	NM_001171155.1		1-4	PET100 deficiency OMIM
PET112	8849	NM_004564.2		1-13	Mitochondrial glutamyl-tRNA(Gln) amidotransferase subunit B deficiency OMIM
PINK1	14581	NM_032409.2		1-8	PINK1 deficiency OMIM Early-onset Parkinson disease type 6 OMIM
PITRM1	17663	NM_014889.3		1-27	Pitrilysin metallopeptidase 1+deficiency OMIM
PMPCA	18667	NM_015160.2		1-13	Mitochondrial processing peptidase alpha deficiency OMIM Autosomal recessive spinocerebellar ataxia type 2 OMIM
PMPCB	9119	NM_004279.2		1-13	Mitochondrial processing peptidase ? deficiency OMIM
PNPLA2	30802	NM_020376.3		2-10	Adipose triglyceride lipase deficiency OMIM Neutral lipid storage disease with myopathy OMIM
PNPLA4	24887	NM_004650.2		2-7	PNPLA4 deficiency OMIM
PNPLA6	16268	NM_006702.4		3-35	PNPLA6 deficiency OMIM Autosomal recessive spastic paraplegia type 39; Oliver-McFarlane syndrome; Boucher-Neuhauser syndrome; Laurence-Moon syndrome OMIM
PNPLA8	28900	NM_001256009.2		2-10	PNPLA8 deficiency OMIM Mitochondrial myopathy with lactic acidosis OMIM

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PNPT1	23166	NM_033109.4	28	1-28	Mitochondrial RNA import protein deficiency OMIM Combined oxidative phosphorylation deficiency type 13 OMIM
POLG	9179	NM_002693.2		2-23	Mitochondrial DNA polymerase g catalytic subunit deficiency OMIM Mitochondrial DNA depletion syndrome type 1; Alpers-Huttenlocher syndrome; mitochondrial recessive ataxia syndrome (MIRAS); arPEO type 1; adPEO type 1 OMIM
POLG2	9180	NM_007215.3		1-8	Mitochondrial DNA polymerase g accessory subunit deficiency OMIM adPEO with mitochondrial DNA deletions type 4 OMIM
PPA2	28883	NM_176869.2		1-12	Mitochondrial inorganic pyrophosphatase 2 deficiency OMIM
PTRH2	24265	NM_016077.4		2	Peptidyl-tRNA hydrolase 2 deficiency OMIM Infantile-onset multisystem neurologic, endocrine, and pancreatic disease OMIM
PUS1	15508	NM_025215.5		1-6	Pseudouridine synthase 1 deficiency OMIM Myopathy, lactic acidosis, and sideroblastic anemia type 1 OMIM
QARS	9751	NM_005051.2		1-24	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy OMIM
QRSL1	21020	NM_018292.4		1-11	Mitochondrial glutamyl-tRNA(Gln) amidotransferase subunit A deficiency OMIM
RARS2	21406	NM_020320.4		1-20	Mitochondrial arginine-tRNA synthetase deficiency OMIM Pontocerebellar hypoplasia type 6 OMIM

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RMND1	21176	NM_017909.3	2	2-12	RMND1 deficiency OMIM Combined oxidative phosphorylation+deficiency+type 11 OMIM
RNASEH1	18466	NM_002936.5		1-8	Mitochondrial ribonuclease H1 deficiency OMIM arPEO with mitochondrial DNA deletions type 2 OMIM
RRM2B	17296	NM_015713.4		1-9	Mitochondrial ribonucleotide reductase small subunit deficiency OMIM Mitochondrial DNA depletion syndrome+type 8; adPEO with mitochondrial DNA deletions type 5 OMIM
RTN4IP1	18647	NM_032730.5		1-9	Nogo-interacting mitochondrial protein deficiency OMIM Optic atrophy type 10 OMIM
SACS	10519	NM_014363.5		2-10	Sacsin deficiency OMIM Autosomal recessive spastic ataxia of Charlevoix-Saguenay OMIM
SAMHD1	15925	NM_015474.3		1-16	SAMHD1 deficiency OMIM Aicardi-GoutiEres syndrome type 5; stenosis, aneurysm, moyamoya and stroke (SAMS association) OMIM
SARS2	17697	NM_017827.3		1-16	Mitochondrial seryl-tRNA synthetase deficiency OMIM Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis (HUPRA) OMIM
SCO1	10603	NM_004589.3		1-6	SCO1 deficiency OMIM
SCO2	10604	NM_005138.2		2	SCO2 deficiency OMIM

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SDHA	10680	NM_004168.3	1-15	1-15	Succinate dehydrogenase subunit A deficiency OMIM Succinate dehydrogenase subunit A deficiency, tumoral phenotype OMIM Hereditary paraganglioma syndrome type 5 OMIM
SDHAF1	33867	NM_001042631.2		1	Succinate dehydrogenase complex assembly factor 1 deficiency OMIM
SDHAF2	26034	NM_017841.2		1-4	Succinate dehydrogenase complex assembly factor 2 deficiency, tumoral phenotype OMIM Hereditary paraganglioma syndrome type 2 OMIM
SDHB	10681	NM_003000.2		1-8	Succinate dehydrogenase subunit B deficiency OMIM Succinate dehydrogenase subunit B deficiency, tumoral phenotype OMIM Hereditary paraganglioma syndrome type 4; Cowden syndrome type 2 OMIM
SDHC	10682	NM_003001.3		1-6	Succinate dehydrogenase subunit C deficiency, tumoral phenotype OMIM Hereditary paraganglioma syndrome type 3 OMIM
SDHD	10683	NM_003002.3		1-4	Succinate dehydrogenase subunit D deficiency OMIM Succinate dehydrogenase subunit D deficiency, tumoral phenotype OMIM Hereditary paraganglioma syndrome type 1; Cowden syndrome type 3 OMIM
SERAC1	21061	NM_032861.3		2-17	SERAC1 deficiency OMIM 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome (MEGDEL) OMIM
SFXN4	16088	NM_213649.1		1-14	Sideroflexin 4 deficiency OMIM Combined oxidative phosphorylation deficiency+type 18 OMIM

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SLC19A2	10938	NM_006996.2		1-6	Thiamine transporter 1 deficiency OMIM Thiamine-responsive megaloblastic anemia; Rogers syndrome; thiamine+metabolism+dysfunction syndrome type 1 OMIM
SLC19A3	16266	NM_025243.3		2-6	Thiamine transporter 2 deficiency OMIM Biotin-thiamine-responsive basal ganglia disease; thiamine metabolism dysfunction syndrome type 2+ OMIM
SLC25A1	10979	NM_005984.4		1-9	Mitochondrial citrate carrier deficiency OMIM Combined D-2- and L-2-hydroxyglutaric aciduria OMIM
SLC25A10	10980	NM_001270888.1		1-11	Mitochondrial dicarboxylate transporter deficiency OMIM
SLC25A12	10982	NM_003705.4	9	1-18	Aspartate-glutamate carrier 1 deficiency OMIM Early infantile epileptic encephalopathy type 39 OMIM
SLC25A13	10983	NM_014251.2		1-18	Citrin deficiency OMIM
SLC25A15	10985	NM_014252.3	2, 6-7	2-7	Mitochondrial ornithine transporter deficiency OMIM Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome; ornithine translocase deficiency OMIM
SLC25A19	14409	NM_021734.4		3-8	Mitochondrial thiamine pyrophosphate transporter deficiency OMIM Amish lethal microcephaly, thiamine metabolism dysfunction syndrome type 3 (severe); bilateral striatal necrosis and progressive polyneuropathy, thiamine metabolism dysfunction syndrome type 4 (milder) OMIM

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SLC25A20	1421	NM_000387.5		1-9	Carnitine-acylcarnitine translocase deficiency OMIM
SLC25A22	19954	NM_024698.5		2-10	Mitochondrial glutamate transporter deficiency OMIM Early infantile epileptic encephalopathy type 3 OMIM
SLC25A24	20662	NM_013386.4		1-10	Mitochondrial+ATP-Mg/phosphate+transporter deficiency OMIM Gorlin-Chaudhry-Moss syndrome; Fontaine syndrome OMIM
SLC25A26	20661	NM_173471.3		2-11	S-adenosylmethionine carrier deficiency OMIM Combined oxidative phosphorylation deficiency type 28 OMIM
SLC25A3	10989	NM_005888.3		2-8	Mitochondrial phosphate carrier deficiency OMIM
SLC25A38	26054	NM_017875.4		1-7	Mitochondrial glycine transporter deficiency OMIM Congenital sideroblastic anemia type 2 OMIM
SLC25A4	10990	NM_001151.3		1-4	Adenine nucleotide translocator deficiency OMIM Mitochondrial DNA depletion syndrome type 12 (cardiomyopathic type); adPEO with mitochondrial DNA deletions type 2 OMIM
SLC25A42	28380	NM_178526.4		2-8	Mitochondrial coenzyme A transporter deficiency OMIM
SLC25A46	25198	NM_138773.3		1-8	UGO-1 like protein deficiency OMIM Hereditary motor and sensory neuropathy type 6B OMIM
SPG7	11237	NM_003119.3		1-17	Paraplegin deficiency OMIM Spastic paraplegia type 7 OMIM
SUCLA2	11448	NM_003850.2		1-11	ATP-specific succinyl-CoA ligase ? subunit deficiency OMIM Mitochondrial DNA depletion syndrome type 5 OMIM

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SUCLG1	11449	NM_003849.3		1-9	GTP-specific succinyl-CoA ligase ? subunit deficiency OMIM Mitochondrial DNA depletion syndrome type 9 OMIM
SURF1	11474	NM_003172.3		1-9	SURF1 deficiency OMIM
TACO1	24316	NM_016360.3		1-5	TACO1 deficiency OMIM
TARS2	30740	NM_025150.4		1-18	Mitochondrial threonyl-tRNA synthetase deficiency OMIM
TAZ	24042	NM_000116.4		1-11	Tafazin deficiency OMIM Barth syndrome OMIM
TFAM	11741	NM_003201.2	7	1-7	Mitochondrial transcription factor A deficiency OMIM
TIMM50	23656	NM_001001563.3		1-11	TIMM50 deficiency OMIM 3-methylglutaconic aciduria type 9 OMIM
TIMM8A	11817	NM_004085.3	2	1-2	TIMM8A deficiency OMIM Mohr-Tranebjaerg syndrome OMIM
TIMMDC1	1321	NM_016589.3		1-7	TIMMDC1 deficiency OMIM
TK2	11831	NM_004614.4		1-10	Mitochondrial thymidine kinase deficiency OMIM Mitochondrial DNA depletion syndrome+type 2 OMIM
TMEM126A	25382	NM_032273.3		2-5	Transmembrane protein 126A deficiency OMIM Optic atrophy type 7 OMIM
TMEM126B	30883	NM_018480.5		1-5	Transmembrane protein 126B deficiency OMIM
TMEM70	26050	NM_017866.5		1-3	Transmembrane protein 70 deficiency OMIM
TPK1	17358	NM_022445.3		2-9	Thiamine pyrophosphokinase+deficiency OMIM Thiamine metabolism dysfunction syndrome type 5 OMIM

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TRAK1	29947	NM_001042646.2		1-16	Trafficking kinesin-binding protein 1 deficiency OMIM
TRIT1	20286	NM_017646.5		1-11	tRNA isopentenyl transferase deficiency OMIM
TRMT10C	26022	NM_017819.3		2	Ribonuclease P 5' tRNA processing enzyme deficiency OMIM Combined oxidative phosphorylation+deficiency+30 OMIM
TRMT5	23141	NM_020810.3		1-5	tRNA methyltransferase 5 deficiency OMIM Combined oxidative phosphorylation deficiency type 26 OMIM
TRMU	25481	NM_018006.4		1-11	tRNA 5-methylaminomethyl-2-thiouridylate-methyltransferase deficiency OMIM Transient infantile liver failure OMIM
TRNT1	17341	NM_182916.2		2-8	CCA-adding tRNA-nucleotidyltransferase deficiency OMIM Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay (severe); retinitis pigmentosa and erythrocytic microcytosis (milder) OMIM
TSFM	12367	NM_001172696.1		1-7	Mitochondrial elongation factor Ts deficiency OMIM Combined oxidative phosphorylation+deficiency+type 3 OMIM
TTC19	26006	NM_017775.3		1-10	TTC19 deficiency OMIM
TUFM	12420	NM_003321.4		1-10	Mitochondrial elongation factor Tu deficiency OMIM Combined oxidative phosphorylation+deficiency+type 4 OMIM
TXN2	17772	NM_012473.3		2-4	Mitochondrial thioredoxin 2 deficiency OMIM

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TYMP	3148	NM_001953.4		2-10	Thymidine phosphorylase deficiency OMIM Mitochondrial neurogastrointestinal encephalopathy syndrome OMIM
UCP2	12518	NM_003355.2		3-8	Uncoupling protein 2 deficiency OMIM
UQCC2	21237	NM_032340.3		1-4	UQCC2 deficiency OMIM
UQCRB	12582	NM_006294.4	3-4	1-4	UQCRB deficiency OMIM
UQCRC2	12586	NM_003366.3		1-14	UQCRC2 deficiency OMIM
UQCRQ	29594	NM_014402.4		2-3	UQCRQ deficiency OMIM
USMG5	30889	NM_001206426.1		2-3	DAPIT deficiency OMIM
VARS2	21642	NM_001167734.1		1-30	Mitochondrial valyl-tRNA synthetase deficiency OMIM Combined oxidative phosphorylation deficiency type 20 OMIM
WARS2	12730	NM_201263.2		1-6	Mitochondrial tryptophanyl-tRNA synthetase deficiency OMIM Mitochondrial neurodevelopmental disorder with abnormal movements and lactic acidosis, with or without seizures+ OMIM
WDR45	28912	NM_007075.3		3-12	WDR45 deficiency OMIM Neurodegeneration with brain iron accumulation type 5; static encephalopathy of childhood with neurodegeneration in adulthood (SENDA); ?-propeller protein-associated neurodegeneration+ (BPAN) OMIM

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WFS1	12762	NM_006005.3		2-8	Deafness, autosomal dominant 6/14/38 OMIM Wolfram syndrome OMIM Wolfram-like syndrome, autosomal dominant OMIM
WWOX	12799	NM_016373.3		1-9	Epileptic encephalopathy, early infantile, 28 OMIM Spinocerebellar ataxia, autosomal recessive 12 OMIM
YARS2	24249	NM_001040436.2		1-5	Mitochondrial tyrosyl-tRNA synthetase deficiency OMIM Myopathy, lactic acidosis, and sideroblastic anemia type 2 OMIM
YME1L1	12843	NM_014263.3	6-8	1-19	YME1L1 deficiency OMIM