Oslo universitetssykehus
Avdeling for medisinsk genetikk

Bevegelsesforstyrrelser

Genpanel, versjon v05

* 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: **ACTB, GBA, GJA1, SLC6A8, SORD**

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 affisert av segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
AAAS	13666	NM_015665.5		1-16	Achalasia-addisonianism-alacrimia syndrome OMIM
AARS	20	NM_001605.2		2-21	Charcot-Marie-Tooth disease, axonal, type 2N OMIM Developmental and epileptic encephalopathy 29 OMIM
AARS2	21022	NM_020745.3		1-22	Combined oxidative phosphorylation deficiency 8 OMIM Leukoencephalopathy, progressive, with ovarian failure OMIM
ABCA1	29	NM_005502.3		2-50	Tangier disease OMIM

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ABCB7	48	NM_004299.5		1-16	Anemia, sideroblastic, with ataxia OMIM
ABCC6	57	NM_001171.5	1-9	1-31	Arterial calcification, generalized, of infancy, 2 OMIM Pseudoxanthoma elasticum OMIM Pseudoxanthoma elasticum, forme fruste OMIM
ABCD1	61	NM_000033.3	7-10	1-10	Adrenoleukodystrophy OMIM Adrenomyeloneuropathy, adult OMIM
ABHD12	15868	NM_001042472.2		1-13	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract OMIM
ACADS	90	NM_000017.3		1-10	Acyl-CoA dehydrogenase, short-chain, deficiency of OMIM
ACAT1	93	NM_000019.4		1-12	Alpha-methylacetoacetic aciduria OMIM
ACO2	118	NM_001098.2		1-18	Infantile cerebellar-retinal degeneration OMIM
ACOX1	119	NM_004035.6		1-14	Mitchell syndrome OMIM Peroxisomal acyl-CoA oxidase deficiency OMIM
ACP5	124	NM_001111035.1		4-7	Spondyloenchondrodysplasia with immune dysregulation OMIM
ACTB	132	NM_001101.4	2-6	2-6	?Dystonia, juvenile-onset OMIM Baraitser-Winter syndrome 1 OMIM
ADAR	225	NM_001111.5		1-15	Aicardi-Goutieres syndrome 6 OMIM
ADCK3	16812	NM_020247.5		2-15	Coenzyme Q10 deficiency, primary, 4 OMIM
ADCY5	236	NM_183357.2		1-21	Dyskinesia, familial, with facial myokymia OMIM
AFG3L2	315	NM_006796.2	14	1-17	Spastic ataxia 5, autosomal recessive OMIM Spinocerebellar ataxia 28 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
AGTPBP1	17258	NM_001286715.1		1-25	Neurodegeneration, childhood-onset, with cerebellar atrophy OMIM
AGXT	341	NM_000030.2		1-11	Hyperoxaluria, primary, type 1 OMIM
AHI1	21575	NM_017651.4		3-28	Joubert syndrome 3 OMIM
AIFM1	8768	NM_004208.3		1-16	Combined oxidative phosphorylation deficiency 6 OMIM Cowchock syndrome OMIM Deafness, X-linked 5 OMIM Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy OMIM
AIMP1	10648	NM_004757.3		2-7	Leukodystrophy, hypomyelinating, 3 OMIM
ALDH18A1	9722	NM_002860.4		2-18	Spastic paraplegia 9A, autosomal dominant OMIM Spastic paraplegia 9B, autosomal recessive OMIM
ALDH3A2	403	NM_000382.3		1-10	Sjogren-Larsson syndrome OMIM
ALDH5A1	408	NM_001080.3		1-10	Succinic semialdehyde dehydrogenase deficiency OMIM
ALG6	23157	NM_013339.3		2-15	Congenital disorder of glycosylation, type Ic OMIM
ALS2	443	NM_020919.3		2-34	Amyotrophic lateral sclerosis 2, juvenile OMIM Primary lateral sclerosis, juvenile OMIM Spastic paralysis, infantile onset ascending OMIM
AMACR	451	NM_014324.6		1-5	Alpha-methylacyl-CoA racemase deficiency OMIM
AMPD2	469	NM_001257360.1		2-19	?Spastic paraplegia 63 OMIM Pontocerebellar hypoplasia, type 9 OMIM

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AMT	473	NM_000481.3		1-9	Glycine encephalopathy OMIM
ANO10	25519	NM_018075.5		2-13	Spinocerebellar ataxia, autosomal recessive 10 OMIM
ANO3	14004	NM_031418.4		1-27	Dystonia 24 OMIM
AP1S1	559	NM_001283.4		1-5	MEDNIK syndrome OMIM
AP1S2	560	NM_003916.4		2-5	Mental retardation, X-linked syndromic 5 OMIM
AP4B1	572	NM_006594.3		2-11	Spastic paraplegia 47, autosomal recessive OMIM
AP4E1	573	NM_007347.4		1-21	Spastic paraplegia 51, autosomal recessive OMIM
AP4M1	574	NM_004722.3		1-15	Spastic paraplegia 50, autosomal recessive OMIM
AP4S1	575	NM_007077.4		2-6	Spastic paraplegia 52, autosomal recessive OMIM
AP5Z1	22197	NM_014855.3		1-17	Spastic paraplegia 48, autosomal recessive OMIM
APOA1	600	NM_000039.2		2-4	Amyloidosis, 3 or more types OMIM
APP	620	NM_000484.4		1-18	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants OMIM
APTX	15984	NM_175073.2		3-9	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia OMIM
AR	644	NM_000044.4		1-8	Spinal and bulbar muscular atrophy of Kennedy OMIM
ARG1	663	NM_000045.4		1-8	Argininemia OMIM
ARHGEF10	14103	NM_014629.3		2-29	?Slowed nerve conduction velocity, AD OMIM

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ARL13B	25419	NM_182896.3		1-10	Joubert syndrome 8 OMIM
ARSA	713	NM_000487.5		1-8	Metachromatic leukodystrophy OMIM
ASPA	756	NM_000049.4		1-6	Canavan disease OMIM
ATCAY	779	NM_033064.4		2-13	Ataxia, cerebellar, Cayman type OMIM
ATL1	11231	NM_015915.4		1-14	Neuropathy, hereditary sensory, type ID OMIM Spastic paraplegia 3A, autosomal dominant OMIM
ATL3	24526	NM_015459.4		1-13	Neuropathy, hereditary sensory, type IF OMIM
ATM	795	NM_000051.3		2-63	Ataxia-telangiectasia OMIM
ATP13A2	30213	NM_022089.4		1-29	Kufor-Rakeb syndrome OMIM Spastic paraplegia 78, autosomal recessive OMIM
ATP1A1	799	NM_000701.8		1-23	Charcot-Marie-Tooth disease, axonal, type 2DD OMIM Hypomagnesemia, seizures, and mental retardation 2 OMIM
ATP1A2	800	NM_000702.4		1-23	Alternating hemiplegia of childhood 1 OMIM Migraine, familial basilar OMIM Migraine, familial hemiplegic, 2 OMIM
ATP1A3	801	NM_152296.5		1-23	Alternating hemiplegia of childhood 2 OMIM CAPOS syndrome OMIM Dystonia-12 OMIM
ATP2B3	816	NM_001001344.2		1-20	?Spinocerebellar ataxia, X-linked 1 OMIM

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ATP7A	869	NM_000052.7		2-23	Menkes disease OMIM Occipital horn syndrome OMIM Spinal muscular atrophy, distal, X-linked 3 OMIM
ATP7B	870	NM_000053.3		1-21	Wilson disease OMIM
ATP8A2	13533	NM_016529.5		1-37	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4 OMIM
ATPAF2	18802	NM_145691.4		1-8	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1 OMIM
ATXN3	7106	NM_004993.5		1-11	Machado-Joseph disease OMIM
AUH	890	NM_001698.2		1-10	3-methylglutaconic aciduria, type I OMIM
B4GALNT1	4117	NM_001478.4		2-11	Spastic paraplegia 26, autosomal recessive OMIM
BAG3	939	NM_004281.3		1-4	Cardiomyopathy, dilated, 1HH OMIM Myopathy, myofibrillar, 6 OMIM
BCAP31	16695	NM_001139441.1	5-8	2-8	Deafness, dystonia, and cerebral hypomyelination OMIM
BCKDHA	986	NM_000709.3		1-9	Maple syrup urine disease, type Ia OMIM
BCKDHB	987	NM_183050.2		1-10	Maple syrup urine disease, type Ib OMIM
BCS1L	1020	NM_004328.5		3-9	Bjornstad syndrome OMIM GRACILE syndrome OMIM Mitochondrial complex III deficiency, nuclear type 1 OMIM
BICD2	17208	NM_001003800.1		1-7	Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant OMIM Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant OMIM

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BSCL2	15832	NM_032667.6		2-11	Encephalopathy, progressive, with or without lipodystrophy OMIM Lipodystrophy, congenital generalized, type 2 OMIM Neuropathy, distal hereditary motor, type VA OMIM Silver spastic paraplegia syndrome OMIM
BTD	1122	NM_000060.3		1-4	Biotinidase deficiency OMIM
C10orf2	1160	NM_021830.5		1-5	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type) OMIM Perrault syndrome 5 OMIM Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3 OMIM
C12orf65	26784	NM_152269.4		2-3	Combined oxidative phosphorylation deficiency 7 OMIM Spastic paraplegia 55, autosomal recessive OMIM
C19orf12	25443	NM_001031726.3		1-3	?Spastic paraplegia 43, autosomal recessive OMIM Neurodegeneration with brain iron accumulation 4 OMIM
C1orf194	32331	NM_001122961.1		1-5	Charcot-Marie-Tooth disease PubMed
C5orf42	25801	NM_023073.3		2-52	Joubert syndrome 17 OMIM
CA8	1382	NM_004056.4		1-8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3 OMIM

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CACNA1A	1388	NM_001127221.1		1-47	Developemental and epileptic encephalopahty 42 OMIM Episodic ataxia, type 2 OMIM Migraine, familial hemiplegic, 1 OMIM Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia OMIM Spinocerebellar ataxia 6 OMIM
CACNA1G	1394	NM_018896.4		1-38	Spinocerebellar ataxia 42 OMIM Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits OMIM
CACNB4	1404	NM_000726.3		1-14	Episodic ataxia, type 5 OMIM
CAMTA1	18806	NM_015215.3		1-23	Cerebellar ataxia, nonprogressive, with mental retardation OMIM
CAPN1	1476	NM_001198868.1		2-22	Spastic paraplegia 76, autosomal recessive OMIM
CASK	1497	NM_003688.3		1-27	Mental retardation and microcephaly with pontine and cerebellar hypoplasia OMIM
CBS	1550	NM_000071.2		3-17	Homocystinuria, B6-responsive and nonresponsive types OMIM Thrombosis, hyperhomocysteinemic OMIM
CC2D2A	29253	NM_001080522.2		3-38	COACH syndrome OMIM Joubert syndrome 9 OMIM Meckel syndrome 6 OMIM
CCDC88C	19967	NM_001080414.4		1-30	?Spinocerebellar ataxia 40 OMIM
CCT5	1618	NM_012073.4		1-11	Neuropathy, hereditary sensory, with spastic paraplegia OMIM
CD59	1689	NM_203330.2		4-6	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy OMIM
CEP290	29021	NM_025114.3	54	2-54	Joubert syndrome 5 OMIM

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CEP41	12370	NM_018718.2		1-11	Joubert syndrome 15 OMIM
CHCHD10	15559	NM_213720.2		1-4	?Myopathy, isolated mitochondrial, autosomal dominant OMIM Spinal muscular atrophy, Jokela type OMIM
CHMP1A	8740	NM_002768.4		1-7	Pontocerebellar hypoplasia, type 8 OMIM
CIC	14214	NM_015125.4		1-20	Mental retardation, autosomal dominant 45 OMIM
CLCN1	2019	NM_000083.3		1-23	Myotonia congenita, dominant OMIM Myotonia congenita, recessive OMIM
CLCN2	2020	NM_004366.5		1-24	Leukoencephalopathy with ataxia OMIM
CLN3	2074	NM_001042432.1		2-16	Ceroid lipofuscinosis, neuronal, 3 OMIM
CLN5	2076	NM_006493.2		1-4	Ceroid lipofuscinosis, neuronal, 5 OMIM
CLN6	2077	NM_017882.2		1-7	Ceroid lipofuscinosis, neuronal, 6 OMIM Ceroid lipofuscinosis, neuronal, Kufs type, adult onset OMIM
CLN8	2079	NM_018941.4		2-3	Ceroid lipofuscinosis, neuronal, 8 OMIM Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant OMIM
CLP1	16999	NM_006831.3		2-3	Pontocerebellar hypoplasia, type 10 OMIM
CLPP	2084	NM_006012.2		1-6	Perrault syndrome 3 OMIM
CNTNAP1	8011	NM_003632.2		1-24	Hypomyelinating neuropathy, congenital, 3 OMIM Lethal congenital contracture syndrome 7 OMIM
COA7	25716	NM_023077.2		1-3	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3 OMIM

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COA8	20492	NM_032374.4		1-5	Mitochondrial complex IV deficiency, nuclear type 17 OMIM
COASY	29932	NM_025233.7		1-9	Neurodegeneration with brain iron accumulation 6 OMIM Pontocerebellar hypoplasia, type 12 OMIM
COL3A1	2201	NM_000090.3		1-51	Ehlers-Danlos syndrome, vascular type OMIM Polymicrogyria with or without vascular-type EDS OMIM
COL4A1	2202	NM_001845.6		1-52	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps OMIM Brain small vessel disease with or without ocular anomalies OMIM Microangiopathy and leukoencephalopathy, pontine, autosomal dominant OMIM
COL4A2	2203	NM_001846.2		2-48	Brain small vessel disease 2 OMIM
COL6A3	2213	NM_004369.3		2-44	Bethlem myopathy 1 OMIM Dystonia 27 OMIM Ullrich congenital muscular dystrophy 1 OMIM
COQ2	25223	NM_015697.7		1-7	Coenzyme Q10 deficiency, primary, 1 OMIM
COQ4	19693	NM_016035.3		1-7	Coenzyme Q10 deficiency, primary, 7 OMIM
COQ9	25302	NM_020312.3		1-9	Coenzyme Q10 deficiency, primary, 5 OMIM
COX10	2260	NM_001303.4	6	1-7	Mitochondrial complex IV deficiency, nuclear type 3 OMIM
COX15	2263	NM_004376.7		1-9	Mitochondrial complex IV deficiency, nuclear type 6 OMIM
COX20	26970	NM_198076.5		1-4	Mitochondrial complex IV deficiency, nuclear type 11 OMIM

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COX6A1	2277	NM_004373.3		1-3	Charcot-Marie-Tooth disease, recessive intermediate D OMIM
CP	2295	NM_000096.3	19	1-19	Cerebellar ataxia OMIM Hemosiderosis, systemic, due to aceruloplasminemia OMIM
CPOX	2321	NM_000097.7		1-7	Coproporphyria OMIM
CSF1R	2433	NM_005211.3		2-22	Brain abnormalities, neurodegeneration, and dysosteosclerosis OMIM Leukoencephalopathy, diffuse hereditary, with spheroids OMIM
CSPP1	26193	NM_024790.6		1-29	Joubert syndrome 21 OMIM
CSTB	2482	NM_000100.3		1-3	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg) OMIM
CTC1	26169	NM_025099.6		1-23	Cerebroretinal microangiopathy with calcifications and cysts OMIM
CTDP1	2498	NM_004715.4		1-13	Congenital cataracts, facial dysmorphism, and neuropathy OMIM
CTSA	9251	NM_000308.2		1-15	Galactosialidosis OMIM
CTSD	2529	NM_001909.4		1-9	Ceroid lipofuscinosis, neuronal, 10 OMIM
CTSF	2531	NM_003793.4		1-13	Ceroid lipofuscinosis, neuronal, 13, Kufs type OMIM
CUBN	2548	NM_001081.3	41-50, 61-67	1-67	Imerslund-Grasbeck syndrome 1 OMIM
CWF19L1	25613	NM_018294.5		1-14	Spinocerebellar ataxia, autosomal recessive 17 OMIM
CYP27A1	2605	NM_000784.3		1-9	Cerebrotendinous xanthomatosis OMIM
CYP2U1	20582	NM_183075.3		1-5	Spastic paraplegia 56, autosomal recessive OMIM

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CYP7B1	2652	NM_004820.3		1-6	Spastic paraplegia 5A, autosomal recessive OMIM
D2HGDH	28358	NM_152783.4		2-10	D-2-hydroxyglutaric aciduria 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	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation OMIM
DBT	2698	NM_001918.4		1-11	Maple syrup urine disease, type II OMIM
DCAF17	25784	NM_025000.3		1-14	Woodhouse-Sakati syndrome OMIM
DCAF8	24891	NM_015726.4		3-14	?Giant axonal neuropathy 2, autosomal dominant OMIM
DCC	2701	NM_005215.3		1-29	Gaze palsy, familial horizontal, with progressive scoliosis, 2 OMIM Mirror movements 1 and/or agenesis of the corpus callosum OMIM
DCTN1	2711	NM_004082.4		1-32	Neuronopathy, distal hereditary motor, type VIIB OMIM Perry syndrome OMIM
DCTN2	2712	NM_006400.4		1-14	Charcot-Marie-Tooth disease PubMed
DDB2	2718	NM_000107.2		1-10	Xeroderma pigmentosum, group E, DDB-negative subtype OMIM
DDC	2719	NM_000790.3		2-14	Aromatic L-amino acid decarboxylase deficiency OMIM
DDHD1	19714	NM_001160147.2		1-13	Spastic paraplegia 28, autosomal recessive OMIM
DDHD2	29106	NM_015214.2		2-17	Spastic paraplegia 54, autosomal recessive OMIM

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DEGS1	13709	NM_003676.3		1-3	Leukodystrophy, hypomyelinating, 18 OMIM
DGUOK	2858	NM_080916.2		1-7	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type) OMIM Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4 OMIM
DHFR	2861	NM_000791.4	6	1-6	Megaloblastic anemia due to dihydrofolate reductase deficiency OMIM
DHTKD1	23537	NM_018706.7		1-17	?Charcot-Marie-Tooth disease, axonal, type 2Q OMIM
DLAT	2896	NM_001931.4		1-14	Pyruvate dehydrogenase E2 deficiency OMIM
DNAJB2	5228	NM_001039550.2		2-10	Spinal muscular atrophy, distal, autosomal recessive, 5 OMIM
DNAJB5	14887	NM_001135004.2		1-5	Charcot-Marie-Tooth disease PubMed
DNAJC12	28908	NM_021800.2		1-5	Hyperphenylalaninemia, mild, non-BH4-deficient OMIM
DNAJC19	30528	NM_145261.3		1-6	3-methylglutaconic aciduria, type V OMIM
DNAJC3	9439	NM_006260.4		1-12	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus OMIM
DNAJC5	16235	NM_025219.3		2-5	Ceroid lipofuscinosis, neuronal, 4, Parry type OMIM
DNM2	2974	NM_001005360.2		1-21	Centronuclear myopathy 1 OMIM Charcot-Marie-Tooth disease, axonal type 2M OMIM Charcot-Marie-Tooth disease, dominant intermediate B OMIM Lethal congenital contracture syndrome 5 OMIM

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DNMT1	2976	NM_001130823.1		1-41	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant OMIM Neuropathy, hereditary sensory, type IE OMIM
DPYD	3012	NM_000110.4		1-23	5-fluorouracil toxicity OMIM Dihydropyrimidine dehydrogenase deficiency OMIM
DRP2	3032	NM_001939.2		3-24	Charcot-Marie-Tooth disease PubMed
DST	1090	NM_001723.5		1-24	?Neuropathy, hereditary sensory and autonomic, type VI OMIM
DYNC1H1	2961	NM_001376.4		1-78	Charcot-Marie-Tooth disease, axonal, type 20 OMIM Mental retardation, autosomal dominant 13 OMIM Spinal muscular atrophy, lower extremity-predominant 1, AD OMIM
DYSF	3097	NM_003494.3		1-55	Miyoshi muscular dystrophy 1 OMIM Muscular dystrophy, limb-girdle, autosomal recessive 2 OMIM Myopathy, distal, with anterior tibial onset OMIM
EARS2	29419	NM_001083614.1		1-9	Combined oxidative phosphorylation deficiency 12 OMIM
ECHS1	3151	NM_004092.3		1-8	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency OMIM
EEF2	3214	NM_001961.3		1-15	?Spinocerebellar ataxia 26 OMIM
EGR2	3239	NM_000399.3		1-2	Charcot-Marie-Tooth disease, type 1D OMIM Dejerine-Sottas disease OMIM Hypomyelinating neuropathy, congenital, 1 OMIM
EIF2AK2	9437	NM_001135651.3		3-17	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome OMIM

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EIF2B1	3257	NM_001414.4		1-9	Leukoencephalopathy with vanishing white matter OMIM
EIF2B2	3258	NM_014239.3		1-8	Leukoencephalopathy with vanishing white matter OMIM Ovarioleukodystrophy OMIM
EIF2B3	3259	NM_020365.5		2-12	Leukoencephalopathy with vanishing white matter OMIM
EIF2B4	3260	NM_015636.3		1-13	Leukoencephalopathy with vanishing white matter OMIM Ovarioleukodystrophy OMIM
EIF2B5	3261	NM_003907.3		1-16	Leukoencephalopathy with vanishing white matter OMIM Ovarioleukodystrophy OMIM
ELOVL4	14415	NM_022726.3		1-6	Ichthyosis, spastic quadriplegia, and mental retardation OMIM Spinocerebellar ataxia 34 OMIM
ELOVL5	21308	NM_021814.4		2-8	Spinocerebellar ataxia 38 OMIM
ENTPD1	3363	NM_001776.5		1-10	Spastic paraplegia 64, autosomal recessive OMIM
EPM2A	3413	NM_005670.3		1-4	Epilepsy, progressive myoclonic 2A (Lafora) OMIM Epilepsy, progressive myoclonic 2B (Lafora) OMIM
ERCC1	3433	NM_202001.2		1-8	Cerebrooculofacioskeletal syndrome 4 OMIM
ERCC2	3434	NM_000400.3		1-23	?Cerebrooculofacioskeletal syndrome 2 OMIM Trichothiodystrophy 1, photosensitive OMIM Xeroderma pigmentosum, group D OMIM
ERCC3	3435	NM_000122.1		1-15	Trichothiodystrophy 2, photosensitive OMIM Xeroderma pigmentosum, group B OMIM

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ERCC4	3436	NM_005236.2		1-11	Fanconi anemia, complementation group Q OMIM Xeroderma pigmentosum, group F OMIM Xeroderma pigmentosum, type F/Cockayne syndrome OMIM XFE progeroid syndrome OMIM
ERCC5	3437	NM_000123.3		1-15	Cerebrooculofacioskeletal syndrome 3 OMIM Xeroderma pigmentosum, group G OMIM Xeroderma pigmentosum, group G/Cockayne syndrome OMIM
ERCC6	3438	NM_000124.3		2-21	Cerebrooculofacioskeletal syndrome 1 OMIM Cockayne syndrome, type B OMIM De Sanctis-Cacchione syndrome OMIM UV-sensitive syndrome 1 OMIM
ERCC8	3439	NM_000082.3		1-12	Cockayne syndrome, type A OMIM
ERLIN2	1356	NM_007175.8		2-12	Spastic paraplegia 18, autosomal recessive OMIM
ETFDH	3483	NM_004453.4		1-13	Glutaric acidemia IIC OMIM
EXOSC3	17944	NM_016042.4		1-4	Pontocerebellar hypoplasia, type 1B OMIM
EXOSC8	17035	NM_181503.2		1-11	Pontocerebellar hypoplasia, type 1C OMIM
FA2H	21197	NM_024306.4		1-7	Spastic paraplegia 35, autosomal recessive OMIM
FAH	3579	NM_000137.2		1-14	Tyrosinemia, type I OMIM
FAM126A	24587	NM_032581.3		2-11	Leukodystrophy, hypomyelinating, 5 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
FAM134B	25964	NM_001034850.2		1-9	Neuropathy, hereditary sensory and autonomic, type IIB OMIM
FAR1	26222	NM_032228.6	12	2-12	Peroxisomal fatty acyl-CoA reductase 1 disorder OMIM
FARSB	17800	NM_005687.4		1-17	Rajab interstitial lung disease with brain calcifications 1 OMIM
FAT2	3596	NM_001447.2		1-23	Spinocerebellar ataxia 45 OMIM
FBLN5	3602	NM_006329.3		1-11	Neuropathy, hereditary, with or without age-related macular degeneration OMIM
FBXL4	13601	NM_012160.4		3-9	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type) OMIM
FBXO38	28844	NM_030793.4		2-22	Neuronopathy, distal hereditary motor, type IID OMIM
FBXO7	13586	NM_012179.3		1-9	Parkinson disease 15, autosomal recessive OMIM
FGD4	19125	NM_139241.3		3-17	Charcot-Marie-Tooth disease, type 4H OMIM
FGF14	3671	NM_004115.3		1-5	Spinocerebellar ataxia 27 OMIM
FIG4	16873	NM_014845.5		1-23	?Polymicrogyria, bilateral temporooccipital OMIM Charcot-Marie-Tooth disease, type 4J OMIM
FLVCR1	24682	NM_014053.3		1-10	Ataxia, posterior column, with retinitis pigmentosa OMIM
FLVCR2	20105	NM_017791.3		1-10	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome OMIM
FMR1	3775	NM_002024.5		1-17	Fragile X tremor/ataxia syndrome OMIM
FOLR1	3791	NM_016725.2		2-5	Neurodegeneration due to cerebral folate transport deficiency OMIM

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FOXC1	3800	NM_001453.2		1	Axenfeld-Rieger syndrome, type 3 OMIM
FOXRED1	26927	NM_017547.4		1-11	Mitochondrial complex I deficiency, nuclear type 19 OMIM
FTL	3999	NM_000146.4		1-4	Neurodegeneration with brain iron accumulation 3 OMIM
FUCA1	4006	NM_000147.4		1-8	Fucosidosis OMIM
FXN	3951	NM_000144.4	5	1-5	Friedreich ataxia OMIM Friedreich ataxia with retained reflexes OMIM
GALC	4115	NM_000153.3		1-17	Krabbe disease OMIM
GALNT2	4124	NM_004481.4		1-16	Congenital disorder of glycosylation, type IIt OMIM
GAMT	4136	NM_000156.6		1-6	Cerebral creatine deficiency syndrome 2 OMIM
GAN	4137	NM_022041.3		1-11	Giant axonal neuropathy-1 OMIM
GARS	4162	NM_002047.2		1-17	Charcot-Marie-Tooth disease, type 2D OMIM Neuronopathy, distal hereditary motor, type VA OMIM Spinal muscular atrophy, infantile, James type OMIM
GATM	4175	NM_001482.2		1-9	Cerebral creatine deficiency syndrome 3 OMIM
GBA	4177	NM_001005741.3	2-12	2-12	Gaucher disease, perinatal lethal OMIM Gaucher disease, type I OMIM Gaucher disease, type II OMIM Gaucher disease, type III OMIM Gaucher disease, type IIIC OMIM
GBA2	18986	NM_020944.2		1-17	Spastic paraplegia 46, autosomal recessive OMIM

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GBE1	4180	NM_000158.3		1-16	Glycogen storage disease IV OMIM Polyglucosan body disease, adult form OMIM
GCDH	4189	NM_000159.3		2-12	Glutaricaciduria, type I OMIM
GCH1	4193	NM_000161.3		1-6	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia OMIM Hyperphenylalaninemia, BH4-deficient, B OMIM
GCLC	4311	NM_001498.3		1-16	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency OMIM
GDAP1	15968	NM_018972.3		1-6	Charcot-Marie-Tooth disease, axonal, type 2K OMIM Charcot-Marie-Tooth disease, axonal, with vocal cord paresis OMIM Charcot-Marie-Tooth disease, recessive intermediate, A OMIM Charcot-Marie-Tooth disease, type 4A OMIM
GDAP2	18010	NM_017686.4		2-14	Spinocerebellar ataxia, autosomal recessive 27 OMIM
GFAP	4235	NM_002055.4		1-9	Alexander disease OMIM
GFM1	13780	NM_024996.6		1-18	Combined oxidative phosphorylation deficiency 1 OMIM
GJA1	4274	NM_000165.5	2	2	Oculodentodigital dysplasia OMIM Oculodentodigital dysplasia, autosomal recessive OMIM
GJB1	4283	NM_000166.5		2	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1 OMIM
GJB3	4285	NM_024009.2		2	Deafness, autosomal dominant, with peripheral neuropathy

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GJC2	17494	NM_020435.4		2	Leukodystrophy, hypomyelinating, 2 OMIM Spastic paraplegia 44, autosomal recessive OMIM
GLA	4296	NM_000169.2		1-7	Fabry disease OMIM Fabry disease, cardiac variant OMIM
GLB1	4298	NM_000404.3		1-16	GM1-gangliosidosis, type I OMIM GM1-gangliosidosis, type II OMIM GM1-gangliosidosis, type III OMIM Mucopolysaccharidosis type IVB (Morquio) OMIM
GLDC	4313	NM_000170.2		1-25	Glycine encephalopathy OMIM
GLRA1	4326	NM_000171.3		1-9	Hyperekplexia 1 OMIM
GM2A	4367	NM_000405.4		1-4	GM2-gangliosidosis, AB variant OMIM
GMPPA	22923	NM_205847.3		2-13	Alacrima, achalasia, and mental retardation syndrome OMIM
GNAL	4388	NM_001142339.2		2-13	Dystonia 25 OMIM
GNAO1	4389	NM_020988.2		1-8	Developmental and epileptic encephalopathy 17 OMIM Neurodevelopmental disorder with involuntary movements OMIM
GNB4	20731	NM_021629.3		2-10	Charcot-Marie-Tooth disease, dominant intermediate F OMIM
GOSR2	4431	NM_004287.3	3-4	1-6	Epilepsy, progressive myoclonic 6 OMIM
GPR56	4512	NM_005682.6		3-15	Polymicrogyria, bilateral frontoparietal OMIM
GRID2	4576	NM_001510.3		1-16	Spinocerebellar ataxia, autosomal recessive 18 OMIM

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GRM1	4593	NM_001278064.1		1-8	Spinocerebellar ataxia 44 OMIM Spinocerebellar ataxia, autosomal recessive 13 OMIM
GRN	4601	NM_002087.3		2-13	Aphasia, primary progressive OMIM Ceroid lipofuscinosis, neuronal, 11 OMIM Frontotemporal lobar degeneration with ubiquitin-positive inclusions OMIM
GSN	4620	NM_000177.4		1-17	Amyloidosis, Finnish type OMIM
GSS	4624	NM_000178.3		2-13	Glutathione synthetase deficiency OMIM
HACE1	21033	NM_020771.4		1-24	Spastic paraplegia and psychomotor retardation with or without seizures OMIM
HADHA	4801	NM_000182.4		1-20	LCHAD deficiency OMIM Mitochondrial trifunctional protein deficiency OMIM
HADHB	4803	NM_000183.3		2-16	Trifunctional protein deficiency OMIM
HARS	4816	NM_002109.5		1-13	Charcot-Marie-Tooth disease, axonal, type 2W OMIM
HEPACAM	26361	NM_152722.4		1-7	Megalencephalic leukoencephalopathy with subcortical cysts 2A OMIM Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation OMIM
HEXA	4878	NM_000520.5		1-14	GM2-gangliosidosis, several forms OMIM Tay-Sachs disease OMIM
HEXB	4879	NM_000521.3		1-14	Sandhoff disease, infantile, juvenile, and adult forms OMIM
HINT1	4912	NM_005340.7		1-3	Neuromyotonia and axonal neuropathy, autosomal recessive OMIM

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HK1	4922	NM_000188.2		1-18	Neurodevelopmental disorder with visual defects and brain anomalies OMIM Neuropathy, hereditary motor and sensory, Russe type OMIM
HMBS	4982	NM_000190.4		1-14	Porphyria, acute intermittent OMIM Porphyria, acute intermittent, nonerythroid variant OMIM
HMGCL	5005	NM_000191.2		1-9	HMG-CoA lyase deficiency OMIM
HPCA	5144	NM_002143.3		2-4	Dystonia 2, torsion, autosomal recessive OMIM
HPRT1	5157	NM_000194.2		1-9	Hyperuricemia, HRPT-related OMIM Lesch-Nyhan syndrome OMIM
HSD17B10	4800	NM_004493.2		1-6	HSD10 mitochondrial disease OMIM
HSD17B4	5213	NM_000414.3		1-24	D-bifunctional protein deficiency OMIM Perrault syndrome 1 OMIM
HSPB1	5246	NM_001540.3		1-3	Charcot-Marie-Tooth disease, axonal, type 2F OMIM Neuronopathy, distal hereditary motor, type IIB OMIM
HSPB3	5248	NM_006308.2		1	?Neuronopathy, distal hereditary motor, type IIC OMIM
HSPB8	30171	NM_014365.2		1-3	Charcot-Marie-Tooth disease, axonal, type 2L OMIM Neuronopathy, distal hereditary motor, type IIA OMIM
HSPD1	5261	NM_002156.4	9-12	2-12	Leukodystrophy, hypomyelinating, 4 OMIM Spastic paraplegia 13, autosomal dominant OMIM
HTRA1	9476	NM_002775.4		1-9	CARASIL syndrome OMIM Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2 OMIM

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IARS2	29685	NM_018060.3		1-23	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia OMIM
IBA57	27302	NM_001010867.3		1-3	?Spastic paraplegia 74, autosomal recessive OMIM Multiple mitochondrial dysfunctions syndrome 3 OMIM
IFIH1	18873	NM_022168.3		1-16	Aicardi-Goutieres syndrome 7 OMIM
IGHMBP2	5542	NM_002180.2		1-15	Charcot-Marie-Tooth disease, axonal, type 2S OMIM Neuronopathy, distal hereditary motor, type VI OMIM
INF2	23791	NM_022489.3		2-22	Charcot-Marie-Tooth disease, dominant intermediate E OMIM
ISCA2	19857	NM_194279.4		1-4	Multiple mitochondrial dysfunctions syndrome 4 OMIM
ITM2B	6174	NM_021999.4		1-6	Dementia, familial British OMIM Dementia, familial Danish OMIM
ITPR1	6180	NM_002222.5		3-58	Gillespie syndrome OMIM Spinocerebellar ataxia 15 OMIM Spinocerebellar ataxia 29, congenital nonprogressive OMIM
JAM2	14686	NM_001270407.1		1-9	Basal ganglia calcification, idiopathic, 8, autosomal recessive OMIM
JAM3	15532	NM_032801.4		1-9	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts OMIM
JPH1	14201	NM_020647.2		1-5	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K OMIM
KARS	6215	NM_001130089.1		2-15	?Charcot-Marie-Tooth disease, recessive intermediate, B OMIM
KCNA1	6218	NM_000217.3		2	Episodic ataxia/myokymia syndrome OMIM

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KCNA2	6220	NM_004974.3		3	Developmental and epileptic encephalopathy 32 OMIM
KCNC1	6233	NM_001112741.2		1-4	Epilepsy, progressive myoclonic 7 OMIM
KCNC3	6235	NM_004977.2		1-4	Spinocerebellar ataxia 13 OMIM
KCND3	6239	NM_004980.4		2-8	Spinocerebellar ataxia 19 OMIM
KCNJ10	6256	NM_002241.4		2	SESAME syndrome OMIM
KCNMA1	6284	NM_002247.3		1-27	Cerebellar atrophy, developmental delay, and seizures OMIM Liang-Wang syndrome OMIM Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy OMIM
KCTD17	25705	NM_001282684.1		1-9	Dystonia 26, myoclonic OMIM
KCTD7	21957	NM_153033.4		1-4	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions OMIM
KIAA0196	28984	NM_014846.3		2-29	Ritscher-Schinzel syndrome 1 OMIM Spastic paraplegia 8, autosomal dominant OMIM
KIF1A	888	NM_004321.7		2-47	NESCAV syndrome OMIM Neuropathy, hereditary sensory, type IIC OMIM Spastic paraplegia 30, autosomal dominant OMIM Spastic paraplegia 30, autosomal recessive OMIM
KIF1B	16636	NM_015074.3		2-47	?Charcot-Marie-Tooth disease, type 2A1 OMIM
KIF1C	6317	NM_006612.6	22-23	3-23	Spastic ataxia 2, autosomal recessive OMIM

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KIF5A	6323	NM_004984.3		1-28	Myoclonus, intractable, neonatal OMIM Spastic paraplegia 10, autosomal dominant OMIM
KLC2	20716	NM_022822.3		2-16	Spastic paraplegia, optic atrophy, and neuropathy OMIM
KMT2B	15840	NM_014727.2		1-37	Dystonia 28, childhood-onset OMIM
L1CAM	6470	NM_000425.5		1-28	Hydrocephalus with congenital idiopathic intestinal pseudoobstruction OMIM
L2HGDH	20499	NM_024884.3		1-10	L-2-hydroxyglutaric aciduria OMIM
LAMA1	6481	NM_005559.3		1-63	Poretti-Boltshauser syndrome OMIM
LAMA2	6482	NM_000426.3		1-65	Muscular dystrophy, congenital, merosin deficient or partially deficient OMIM Muscular dystrophy, limb-girdle, autosomal recessive 23 OMIM
LAMB1	6486	NM_002291.3		2-34	Lissencephaly 5 OMIM
LARS2	17095	NM_015340.3		3-22	?Hydrops, lactic acidosis, and sideroblastic anemia OMIM Perrault syndrome 4 OMIM
LITAF	16841	NM_004862.3		2-4	Charcot-Marie-Tooth disease, type 1C OMIM
LMBRD1	23038	NM_018368.3		1-16	Methylmalonic aciduria and homocystinuria, cblF type OMIM
LMNA	6636	NM_170707.4		1-12	Charcot-Marie-Tooth disease, type 2B1 OMIM Emery-Dreifuss muscular dystrophy 2, autosomal dominant OMIM Emery-Dreifuss muscular dystrophy 3, autosomal recessive OMIM Muscular dystrophy, congenital OMIM Restrictive dermopathy, lethal OMIM

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LMNB1	6637	NM_005573.4		1-11	Leukodystrophy, adult-onset, autosomal dominant OMIM
LRRK2	18618	NM_198578.3		1-51	{Parkinson disease 8} OMIM
LRSAM1	25135	NM_138361.5		2-25	Charcot-Marie-Tooth disease, axonal, type 2P OMIM
LYRM7	28072	NM_181705.3		1-5	Mitochondrial complex III deficiency, nuclear type 8 OMIM
LYST	1968	NM_000081.4		3-53	Chediak-Higashi syndrome OMIM
MAG	6783	NM_002361.4		3-11	Spastic paraplegia 75, autosomal recessive OMIM
MARS	6898	NM_004990.3		1-21	Charcot-Marie-Tooth disease, axonal, type 2U OMIM
MARS2	25133	NM_138395.4		1	?Combined oxidative phosphorylation deficiency 25 OMIM Spastic ataxia 3, autosomal recessive OMIM
MCM3AP	6946	NM_003906.4		1-28	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development OMIM
MCOLN1	13356	NM_020533.3		1-14	Mucolipidosis IV OMIM
MECR	19691	NM_016011.5		1-10	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities OMIM
MEF2C	6996	NM_002397.4		2-11	Chromosome 5q14.3 deletion syndrome OMIM Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations OMIM

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MFN2	16877	NM_014874.4		3-19	Charcot-Marie-Tooth disease, axonal, type 2A2A OMIM Charcot-Marie-Tooth disease, axonal, type 2A2B OMIM Hereditary motor and sensory neuropathy VIA OMIM
MFSD8	28486	NM_152778.2		2-13	Ceroid lipofuscinosis, neuronal, 7 OMIM
MICU1	1530	NM_006077.3		2-12	Myopathy with extrapyramidal signs OMIM
MKS1	7121	NM_017777.3		1-18	Joubert syndrome 28 OMIM
MLC1	17082	NM_015166.3		2-12	Megalencephalic leukoencephalopathy with subcortical cysts OMIM
MMAA	18871	NM_172250.3		2-7	Methylmalonic aciduria, vitamin B12-responsive OMIM
MMAB	19331	NM_052845.3		1-9	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type OMIM
MMACHC	24525	NM_015506.2		1-4	Methylmalonic aciduria and homocystinuria, cblC type OMIM
MMADHC	25221	NM_015702.2		2-8	Homocystinuria, cblD type, variant 1 OMIM Methylmalonic aciduria and homocystinuria, cblD type OMIM Methylmalonic aciduria, cblD type, variant 2 OMIM
MME	7154	NM_007289.2		2-23	?Spinocerebellar ataxia 43 OMIM Charcot-Marie-Tooth disease, axonal, type 2T OMIM
MORC2	23573	NM_014941.2		5-27	Charcot-Marie-Tooth disease, axonal, type 2Z OMIM
MPLKIP	16002	NM_138701.4		1-2	Trichothiodystrophy 4, nonphotosensitive OMIM

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MPV17	7224	NM_002437.4		2-8	Charcot-Marie-Tooth disease, axonal, type 2EE OMIM Mitochondrial DNA depletion syndrome 6 (hepatocerebral type) OMIM
MPZ	7225	NM_000530.7		1-6	Charcot-Marie-Tooth disease, dominant intermediate D OMIM Charcot-Marie-Tooth disease, type 1B OMIM Charcot-Marie-Tooth disease, type 2I OMIM Charcot-Marie-Tooth disease, type 2J OMIM Dejerine-Sottas disease OMIM Hypomyelinating neuropathy, congenital, 2 OMIM Roussy-Levy syndrome OMIM
MRE11A	7230	NM_005591.3		2-20	Ataxia-telangiectasia-like disorder 1 OMIM
MRPS16	14048	NM_016065.3		1-3	Combined oxidative phosphorylation deficiency 2 OMIM
MTFMT	29666	NM_139242.3		1-9	Combined oxidative phosphorylation deficiency 15 OMIM Mitochondrial complex I deficiency, nuclear type 27 OMIM
MTHFR	7436	NM_005957.4		2-12	Homocystinuria due to MTHFR deficiency OMIM
MTMR2	7450	NM_016156.5		1-15	Charcot-Marie-Tooth disease, type 4B1 OMIM
MTPAP	25532	NM_018109.3		1-9	?Spastic ataxia 4, autosomal recessive OMIM
MTR	7468	NM_000254.2		1-33	Homocystinuria-megaloblastic anemia, cblG complementation type OMIM
MTRR	7473	NM_002454.2		2-15	Homocystinuria-megaloblastic anemia, cbl E type OMIM
MTTP	7467	NM_000253.3		2-19	Abetalipoproteinemia OMIM

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MUT	7526	NM_000255.4		2-13	Methylmalonic aciduria, mut(0) type OMIM
MVK	7530	NM_000431.3		2-11	Mevalonic aciduria OMIM
MYH14	23212	NM_024729.3		2-41	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss OMIM Deafness, autosomal dominant 4A OMIM
MYORG	19918	NM_020702.4		2	Basal ganglia calcification, idiopathic, 7, autosomal recessive OMIM
NAGA	7631	NM_000262.2		1-9	Kanzaki disease OMIM Schindler disease, type I OMIM Schindler disease, type III OMIM
NAGLU	7632	NM_000263.4		1-6	?Charcot-Marie-Tooth disease, axonal, type 2V OMIM Mucopolysaccharidosis type IIIB (Sanfilippo B) OMIM
NDRG1	7679	NM_006096.4		2-16	Charcot-Marie-Tooth disease, type 4D OMIM
NDUFA10	7684	NM_004544.3		1-10	Mitochondrial complex I deficiency, nuclear type 22 OMIM
NDUFA2	7685	NM_002488.4		1-3	Mitochondrial complex I deficiency, nuclear type 13 OMIM
NDUFAF1	18828	NM_016013.3		2-5	Mitochondrial complex I deficiency, nuclear type 11 OMIM
NDUFAF3	29918	NM_199069.1		1-5	Mitochondrial complex I deficiency, nuclear type 18 OMIM
NDUFAF5	15899	NM_024120.4		1-11	Mitochondrial complex I deficiency, nuclear type 16 OMIM
NDUFS1	7707	NM_005006.7		2-19	Mitochondrial complex I deficiency, nuclear type 5 OMIM
NDUFS2	7708	NM_004550.4		2-15	Mitochondrial complex I deficiency, nuclear type 6 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
NDUFS4	7711	NM_002495.4		1-5	Mitochondrial complex I deficiency, nuclear type 1 OMIM
NDUFS7	7714	NM_024407.4		1-8	Mitochondrial complex I deficiency, nuclear type 3 OMIM
NDUFS8	7715	NM_002496.3		2-7	Mitochondrial complex I deficiency, nuclear type 2 OMIM
NDUFV1	7716	NM_007103.3		1-10	Mitochondrial complex I deficiency, nuclear type 4 OMIM
NEFH	7737	NM_021076.4	4	1-4	Charcot-Marie-Tooth disease, axonal, type 2CC OMIM
NEFL	7739	NM_006158.4		1-5	Charcot-Marie-Tooth disease, dominant intermediate G OMIM Charcot-Marie-Tooth disease, type 1F OMIM Charcot-Marie-Tooth disease, type 2E OMIM
NEU1	7758	NM_000434.3		1-6	Sialidosis, type I OMIM Sialidosis, type II OMIM
NGF	7808	NM_002506.2		3	Neuropathy, hereditary sensory and autonomic, type V OMIM
NHLRC1	21576	NM_198586.2		1	Epilepsy, progressive myoclonic 2B (Lafora) OMIM
NIPA1	17043	NM_144599.4		1-5	Spastic paraplegia 6, autosomal dominant OMIM
NKX2-1	11825	NM_001079668.2		1-3	Choreoathetosis, hypothyroidism, and neonatal respiratory distress OMIM
NKX6-2	19321	NM_177400.2		1-3	Spastic ataxia 8 with hypomyelinating leukodystrophy OMIM
NOL3	7869	NM_001276312.1		4-6	?Myoclonus, familial, 1 OMIM
NOTCH3	7883	NM_000435.2		1-33	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1 OMIM

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NPC1	7897	NM_000271.4		1-25	Niemann-Pick disease, type C1 OMIM Niemann-Pick disease, type D OMIM
NPC2	14537	NM_006432.4		1-5	Niemann-pick disease, type C2 OMIM
NPHP1	7905	NM_000272.3		1-20	Joubert syndrome 4 OMIM
NRROS	24613	NM_198565.3		2-3	Seizures, early-onset, with neurodegeneration and brain calcification OMIM
NT5C2	8022	NM_012229.4		3-19	Spastic paraplegia 45, autosomal recessive OMIM
NTRK1	8031	NM_001012331.1		1-16	Insensitivity to pain, congenital, with anhidrosis OMIM
NUBPL	20278	NM_025152.3		1-11	Mitochondrial complex I deficiency, nuclear type 21 OMIM
NUP188	17859	NM_015354.3		1-44	Sandestig-Stefanova syndrome OMIM
NUP62	8066	NM_001193357.1		2	Striatonigral degeneration, infantile OMIM
OCLN	8104	NM_002538.3	5-9	2-9	Pseudo-TORCH syndrome 1 OMIM
OCRL	8108	NM_000276.3		1-24	Dent disease 2 OMIM Lowe syndrome OMIM
OPA1	8140	NM_015560.2		1-28	?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type) OMIM Behr syndrome OMIM Optic atrophy plus syndrome OMIM
OPA3	8142	NM_025136.4		1-2	3-methylglutaconic aciduria, type III OMIM
OPHN1	8148	NM_002547.3		2-24	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
PAFAH1B1	8574	NM_000430.4		2-11	Lissencephaly 1 OMIM Subcortical laminar heterotopia OMIM
PAH	8582	NM_000277.2		1-13	[Hyperphenylalaninemia, non-PKU mild] OMIM Phenylketonuria OMIM
PANK2	15894	NM_153638.2		1-7	HARP syndrome OMIM Neurodegeneration with brain iron accumulation 1 OMIM
PARK2	8607	NM_004562.3		1-12	Parkinson disease, juvenile, type 2 OMIM
PARK7	16369	NM_007262.4		2-7	Parkinson disease 7, autosomal recessive early-onset OMIM
PCCA	8653	NM_000282.4		1-24	Propionicacidemia OMIM
PCCB	8654	NM_000532.4		1-15	Propionicacidemia OMIM
PCDH12	8657	NM_016580.3		1-4	Diencephalic-mesencephalic junction dysplasia syndrome 1 OMIM
PCNA	8729	NM_002592.2		2-7	?Ataxia-telangiectasia-like disorder 2 OMIM
PDE10A	8772	NM_001130690.2		1-22	Dyskinesia, limb and orofacial, infantile-onset OMIM Striatal degeneration, autosomal dominant OMIM
PDE8B	8794	NM_003719.4		1-22	Striatal degeneration, autosomal dominant OMIM
PDGFB	8800	NM_002608.4		1-6	Basal ganglia calcification, idiopathic, 5 OMIM
PDGFRB	8804	NM_002609.3		2-23	Basal ganglia calcification, idiopathic, 4 OMIM Kosaki overgrowth syndrome OMIM
PDHA1	8806	NM_000284.3		1-11	Pyruvate dehydrogenase E1-alpha deficiency OMIM

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PDHX	21350	NM_003477.2		1-11	Lacticacidemia due to PDX1 deficiency OMIM
PDK3	8811	NM_001142386.3		1-12	?Charcot-Marie-Tooth disease, X-linked dominant, 6 OMIM
PDSS1	17759	NM_014317.3		1-12	Coenzyme Q10 deficiency, primary, 2 OMIM
PDSS2	23041	NM_020381.4		1-8	Coenzyme Q10 deficiency, primary, 3 OMIM
PDYN	8820	NM_024411.5		3-4	Spinocerebellar ataxia 23 OMIM
PET100	40038	NM_001171155.2		1-4	Mitochondrial complex IV deficiency, nuclear type 12 OMIM
PEX1	8850	NM_000466.2		1-24	Heimler syndrome 1 OMIM Peroxisome biogenesis disorder 1A (Zellweger) OMIM Peroxisome biogenesis disorder 1B (NALD/IRD) OMIM
PEX10	8851	NM_153818.1		1-6	Peroxisome biogenesis disorder 6A (Zellweger) OMIM Peroxisome biogenesis disorder 6B OMIM
PEX11B	8853	NM_003846.2		1-4	?Peroxisome biogenesis disorder 14B OMIM
PEX12	8854	NM_000286.2		1-3	Peroxisome biogenesis disorder 3A (Zellweger) OMIM Peroxisome biogenesis disorder 3B OMIM
PEX13	8855	NM_002618.4		1-4	Peroxisome biogenesis disorder 11A (Zellweger) OMIM Peroxisome biogenesis disorder 11B OMIM
PEX14	8856	NM_004565.2		1-9	Peroxisome biogenesis disorder 13A (Zellweger) OMIM

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PEX16	8857	NM_004813.3		1-11	Peroxisome biogenesis disorder 8A (Zellweger) OMIM Peroxisome biogenesis disorder 8B OMIM
PEX19	9713	NM_002857.3		1-8	Peroxisome biogenesis disorder 12A (Zellweger) OMIM
PEX2	9717	NM_000318.2		4	Peroxisome biogenesis disorder 5A (Zellweger) OMIM Peroxisome biogenesis disorder 5B OMIM
PEX26	22965	NM_017929.6		2-6	Peroxisome biogenesis disorder 7A (Zellweger) OMIM Peroxisome biogenesis disorder 7B OMIM
PEX3	8858	NM_003630.3		1-12	?Peroxisome biogenesis disorder 10B OMIM Peroxisome biogenesis disorder 10A (Zellweger) OMIM
PEX5	9719	NM_001131025.1		2-16	Peroxisome biogenesis disorder 2A (Zellweger) OMIM Peroxisome biogenesis disorder 2B OMIM Rhizomelic chondrodysplasia punctata, type 5 OMIM
PEX6	8859	NM_000287.3		1-17	Heimler syndrome 2 OMIM Peroxisome biogenesis disorder 4A (Zellweger) OMIM Peroxisome biogenesis disorder 4B OMIM
PEX7	8860	NM_000288.4		1-10	Peroxisome biogenesis disorder 9B OMIM Rhizomelic chondrodysplasia punctata, type 1 OMIM
PGK1	8896	NM_000291.4		1-11	Phosphoglycerate kinase 1 deficiency OMIM
PGM3	8907	NM_001199917.2		2-14	Immunodeficiency 23 OMIM

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PHOX2B	9143	NM_003924.3		1-3	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease OMIM
PHYH	8940	NM_006214.3		1-9	Refsum disease OMIM
PINK1	14581	NM_032409.2		1-8	Parkinson disease 6, early onset OMIM
PLA2G6	9039	NM_003560.3		2-17	Infantile neuroaxonal dystrophy 1 OMIM Neurodegeneration with brain iron accumulation 2B OMIM Parkinson disease 14, autosomal recessive OMIM
PLEKHG5	29105	NM_020631.4		2-21	Charcot-Marie-Tooth disease, recessive intermediate C OMIM Spinal muscular atrophy, distal, autosomal recessive, 4 OMIM
PLP1	9086	NM_000533.5		1-7	Pelizaeus-Merzbacher disease OMIM Spastic paraplegia 2, X-linked OMIM
PMM2	9115	NM_000303.2		1-8	Congenital disorder of glycosylation, type Ia OMIM
PMP2	9117	NM_002677.4		1-4	Charcot-Marie-Tooth disease, demyelinating, type 1G OMIM
PMP22	9118	NM_000304.4		2-5	?Neuropathy, inflammatory demyelinating OMIM Charcot-Marie-Tooth disease, type 1A OMIM Charcot-Marie-Tooth disease, type 1E OMIM Dejerine-Sottas disease OMIM Neuropathy, recurrent, with pressure palsies OMIM Roussy-Levy syndrome OMIM
PMPCA	18667	NM_015160.3		1-13	Spinocerebellar ataxia, autosomal recessive 2 OMIM
PNKD	9153	NM_015488.5		1-10	Paroxysmal nonkinesigenic dyskinesia 1 OMIM

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PNKP	9154	NM_007254.3		2-17	?Charcot-Marie-Tooth disease, type 2B2 OMIM Ataxia-oculomotor apraxia 4 OMIM Microcephaly, seizures, and developmental delay OMIM
PNP	7892	NM_000270.3		1-6	Immunodeficiency due to purine nucleoside phosphorylase deficiency OMIM
PNPLA6	16268	NM_006702.4		3-35	?Laurence-Moon syndrome OMIM Boucher-Neuhauser syndrome OMIM Oliver-McFarlane syndrome OMIM Spastic paraplegia 39, autosomal recessive OMIM
POLG	9179	NM_002693.2		2-23	Mitochondrial DNA depletion syndrome 4A (Alpers type) OMIM Mitochondrial DNA depletion syndrome 4B (MNGIE type) OMIM Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE) OMIM Progressive external ophthalmoplegia, autosomal dominant 1 OMIM Progressive external ophthalmoplegia, autosomal recessive 1 OMIM
POLG2	9180	NM_007215.4		1-8	Mitochondrial DNA depletion syndrome 16 (hepatic type) OMIM Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4 OMIM
POLR1C	20194	NM_203290.3		1-9	Leukodystrophy, hypomyelinating, 11 OMIM
POLR3A	30074	NM_007055.3		1-31	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism OMIM Wiedemann-Rautenstrauch syndrome OMIM

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POLR3B	30348	NM_018082.5		1-28	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism OMIM
PPOX	9280	NM_000309.5		2-13	Porphyria variegata OMIM
PPT1	9325	NM_000310.3		1-9	Ceroid lipofuscinosis, neuronal, 1 OMIM
PRDM12	13997	NM_021619.3		1-5	Neuropathy, hereditary sensory and autonomic, type VIII OMIM
PRF1	9360	NM_001083116.1		2-3	Hemophagocytic lymphohistiocytosis, familial, 2 OMIM Lymphoma, non-Hodgkin OMIM
PRICKLE1	17019	NM_153026.2		2-8	Epilepsy, progressive myoclonic 1B OMIM
PRKCG	9402	NM_002739.3		1-18	Spinocerebellar ataxia 14 OMIM
PRKRA	9438	NM_003690.4		1-8	Dystonia 16 OMIM
PRNP	9449	NM_000311.3		2	Cerebral amyloid angiopathy, PRNP-related OMIM Creutzfeldt-Jakob disease OMIM Gerstmann-Straussler disease OMIM Huntington disease-like 1 OMIM Insomnia, fatal familial OMIM Prion disease with protracted course OMIM
PRPS1	9462	NM_002764.4	7	1-7	Arts syndrome OMIM Charcot-Marie-Tooth disease, X-linked recessive, 5 OMIM Gout, PRPS-related OMIM Phosphoribosylpyrophosphate synthetase superactivity OMIM

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PRRT2	30500	NM_145239.2		2-4	Convulsions, familial infantile, with paroxysmal choreoathetosis OMIM Episodic kinesigenic dyskinesia 1 OMIM Seizures, benign familial infantile, 2 OMIM
PRX	13797	NM_181882.2		4-7	Charcot-Marie-Tooth disease, type 4F OMIM Dejerine-Sottas disease OMIM
PSAP	9498	NM_002778.2		1-14	Combined SAP deficiency OMIM Gaucher disease, atypical OMIM Krabbe disease, atypical OMIM Metachromatic leukodystrophy due to SAP-b deficiency OMIM
PSEN1	9508	NM_000021.3		3-12	Alzheimer disease, type 3 OMIM Alzheimer disease, type 3, with spastic paraparesis and apraxia OMIM Alzheimer disease, type 3, with spastic paraparesis and unusual plaques OMIM Dementia, frontotemporal OMIM Pick disease OMIM
PTEN	9588	NM_000314.6	9	1-9	Cowden syndrome 1 OMIM Lhermitte-Duclos syndrome OMIM Macrocephaly/autism syndrome OMIM
PTF1A	23734	NM_178161.3		1-2	Pancreatic and cerebellar agenesis OMIM
PTPN11	9644	NM_002834.4		1-15	Noonan syndrome 1 OMIM
PTRH2	24265	NM_016077.4		2	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease OMIM
PTS	9689	NM_000317.2		1-6	Hyperphenylalaninemia, BH4-deficient, A OMIM
PUM1	14957	NM_001020658.1		2-22	Spinocerebellar ataxia 47 OMIM
PYCR2	30262	NM_013328.4		1-7	Leukodystrophy, hypomyelinating, 10 OMIM

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QDPR	9752	NM_000320.2		1-7	Hyperphenylalaninemia, BH4-deficient, C OMIM
RAB7A	9788	NM_004637.5		2-6	Charcot-Marie-Tooth disease OMIM
RAD51	9817	NM_002875.4		2-10	Mirror movements 2 OMIM
RARS	9870	NM_002887.3		1-15	Leukodystrophy, hypomyelinating, 9 OMIM
RARS2	21406	NM_020320.3		1-20	Pontocerebellar hypoplasia, type 6 OMIM
REEP1	25786	NM_022912.2		1-7	?Neuronopathy, distal hereditary motor, type VB OMIM Spastic paraplegia 31, autosomal dominant OMIM
RNASEH1	18466	NM_002936.4		1-8	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2 OMIM
RNASEH2A	18518	NM_006397.2		1-8	Aicardi-Goutieres syndrome 4 OMIM
RNASEH2B	25671	NM_024570.3		1-11	Aicardi-Goutieres syndrome 2 OMIM
RNASEH2C	24116	NM_032193.3		1-4	Aicardi-Goutieres syndrome 3 OMIM
RNASET2	21686	NM_003730.4		1-9	Leukoencephalopathy, cystic, without megalencephaly OMIM
RNF168	26661	NM_152617.4		1-6	RIDDLE syndrome OMIM
RNF170	25358	NM_001160223.1		2-7	Ataxia, sensory, 1, autosomal dominant OMIM
RNF216	21698	NM_207111.4	2, 6-8	2-17	Cerebellar ataxia and hypogonadotropic hypogonadism OMIM
RPGRIP1L	29168	NM_015272.2		2-27	COACH syndrome OMIM Joubert syndrome 7 OMIM

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RPS6KA3	10432	NM_004586.3		1-22	Coffin-Lowry syndrome OMIM Mental retardation, X-linked 19 OMIM
RRM2B	17296	NM_015713.4		1-9	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy) OMIM Mitochondrial DNA depletion syndrome 8B (MNGIE type) OMIM Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5 OMIM
RTN2	10468	NM_005619.4		1-11	Spastic paraplegia 12, autosomal dominant OMIM
RTN4IP1	18647	NM_032730.4		1-9	Optic atrophy 10 with or without ataxia, mental retardation, and seizures OMIM
SACS	10519	NM_014363.6		2-10	Spastic ataxia, Charlevoix-Saguenay type OMIM
SAMHD1	15925	NM_015474.3		1-16	Aicardi-Goutieres syndrome 5 OMIM
SBF1	10542	NM_002972.4		1-41	Charcot-Marie-Tooth disease, type 4B3 OMIM
SBF2	2135	NM_030962.3		1-40	Charcot-Marie-Tooth disease, type 4B2 OMIM
SCARB2	1665	NM_005506.3		1-12	Epilepsy, progressive myoclonic 4, with or without renal failure OMIM
SCN10A	10582	NM_006514.3		1-27	Episodic pain syndrome, familial, 2 OMIM
SCN11A	10583	NM_014139.2		1-26	Episodic pain syndrome, familial, 3 OMIM Neuropathy, hereditary sensory and autonomic, type VII OMIM
SCN1A	10585	NM_001165963.1		1-26	Dravet syndrome OMIM Epilepsy, generalized, with febrile seizures plus, type 2 OMIM Febrile seizures, familial, 3A OMIM Migraine, familial hemiplegic, 3 OMIM

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SCN4A	10591	NM_000334.4		1-24	Hyperkalemic periodic paralysis, type 2 OMIM Hypokalemic periodic paralysis, type 2 OMIM Myasthenic syndrome, congenital, 16 OMIM Myotonia congenita, atypical, acetazolamide-responsive OMIM Paramyotonia congenita OMIM
SCN8A	10596	NM_014191.3		2-27	?Myoclonus, familial, 2 OMIM Cognitive impairment with or without cerebellar ataxia OMIM Developmental and epileptic encephalopathy 13 OMIM Seizures, benign familial infantile, 5 OMIM
SCN9A	10597	NM_002977.3		2-27	Erythermalgia, primary OMIM Febrile seizures, familial, 3B OMIM Generalized epilepsy with febrile seizures plus, type 7 OMIM Insensitivity to pain, congenital OMIM Neuropathy, hereditary sensory and autonomic, type IID OMIM Paroxysmal extreme pain disorder OMIM Small fiber neuropathy OMIM
SCO1	10603	NM_004589.3		1-6	Mitochondrial complex IV deficiency, nuclear type 4 OMIM
SCO2	10604	NM_005138.2		2	Mitochondrial complex IV deficiency, nuclear type 2 OMIM
SCP2	10606	NM_002979.5		1-16	?Leukoencephalopathy with dystonia and motor neuropathy OMIM
SCYL1	14372	NM_020680.4		1-18	Spinocerebellar ataxia, autosomal recessive 21 OMIM
SDHAF1	33867	NM_001042631.2		1	Mitochondrial complex II deficiency OMIM

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SDHB	10681	NM_003000.2		1-8	Mitochondrial leukencephalopathy and complex II deficiency
SEPSECS	30605	NM_016955.4		1-11	Pontocerebellar hypoplasia type 2D OMIM
SEPT9	7323	NM_006640.4		1-11	Amyotrophy, hereditary neuralgic OMIM
SERAC1	21061	NM_032861.4		2-17	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome OMIM
SETX	445	NM_015046.7		3-26	Amyotrophic lateral sclerosis 4, juvenile OMIM Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2 OMIM
SGCE	10808	NM_003919.2		1-11	Dystonia-11, myoclonic OMIM
SGPL1	10817	NM_003901.4		2-15	Nephrotic syndrome, type 14 OMIM
SH3TC2	29427	NM_024577.3		1-17	Charcot-Marie-Tooth disease, type 4C OMIM Mononeuropathy of the median nerve, mild OMIM
SIGMAR1	8157	NM_005866.4		1-4	?Amyotrophic lateral sclerosis 16, juvenile OMIM ?Spinal muscular atrophy, distal, autosomal recessive, 2 OMIM
SIL1	24624	NM_022464.4		2-10	Marinesco-Sjogren syndrome OMIM
SLC12A6	10914	NM_133647.1		1-25	Agenesis of the corpus callosum with peripheral neuropathy OMIM
SLC16A2	10923	NM_006517.5		1-6	Allan-Herndon-Dudley syndrome OMIM
SLC17A5	10933	NM_012434.4		1-11	Salla disease OMIM Sialic acid storage disorder, infantile OMIM

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SLC19A2	10938	NM_006996.2		1-6	Thiamine-responsive megaloblastic anemia syndrome OMIM
SLC19A3	16266	NM_025243.3		2-6	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2) OMIM
SLC1A3	10941	NM_004172.4		2-10	Episodic ataxia, type 6 OMIM
SLC20A2	10947	NM_006749.5		2-11	Basal ganglia calcification, idiopathic, 1 OMIM
SLC25A12	10982	NM_003705.5	9	1-18	Developmental and epileptic encephalopathy 39 OMIM
SLC25A15	10985	NM_014252.3	2, 6-7	2-7	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome OMIM
SLC25A19	14409	NM_021734.4		3-8	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type) OMIM
SLC25A4	10990	NM_001151.3		1-4	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD OMIM Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR OMIM Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2 OMIM
SLC25A46	25198	NM_138773.2		1-8	Neuropathy, hereditary motor and sensory, type VIB OMIM
SLC2A1	11005	NM_006516.3		1-10	{Epilepsy, idiopathic generalized, susceptibility to, 12} OMIM Dystonia 9 OMIM GLUT1 deficiency syndrome 1, infantile onset, severe OMIM GLUT1 deficiency syndrome 2, childhood onset OMIM Stomatin-deficient cryohydrocytosis with neurologic defects OMIM

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SLC30A10	25355	NM_018713.2		1-4	Hypermanganesemia with dystonia 1 OMIM
SLC33A1	95	NM_004733.3	6	1-6	Congenital cataracts, hearing loss, and neurodegeneration OMIM Spastic paraplegia 42, autosomal dominant OMIM
SLC39A4	17129	NM_130849.3		1-12	Acrodermatitis enteropathica OMIM
SLC52A2	30224	NM_024531.5		2-5	Brown-Vialetto-Van Laere syndrome 2 OMIM
SLC52A3	16187	NM_033409.4		2-5	?Fazio-Londe disease OMIM Brown-Vialetto-Van Laere syndrome 1 OMIM
SLC5A7	14025	NM_021815.5		2-9	Myasthenic syndrome, congenital, 20, presynaptic OMIM Neuronopathy, distal hereditary motor, type VIIA OMIM
SLC6A1	11042	NM_003042.3		3-16	Myoclonic-atonic epilepsy OMIM
SLC6A3	11049	NM_001044.4		2-15	Parkinsonism-dystonia, infantile, 1 OMIM
SLC6A8	11055	NM_005629.4	1-13	1-13	Cerebral creatine deficiency syndrome 1 OMIM
SLC9A6	11079	NM_006359.3		1-16	Mental retardation, X-linked syndromic, Christianson type OMIM
SNCA	11138	NM_000345.3		2-6	Dementia, Lewy body OMIM Parkinson disease 1 OMIM Parkinson disease 4 OMIM
SNX14	14977	NM_153816.6		1-29	Spinocerebellar ataxia, autosomal recessive 20 OMIM
SOD1	11179	NM_000454.4		1-5	Amyotrophic lateral sclerosis 1 OMIM Spastic tetraplegia and axial hypotonia, progressive OMIM
SORD	11184	NM_003104.6	1-9	1-9	Sorbitol dehydrogenase deficiency with peripheral neuropathy OMIM

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SOX10	11190	NM_006941.3		2-4	PCWH syndrome OMIM Waardenburg syndrome, type 2E, with or without neurologic involvement OMIM Waardenburg syndrome, type 4C OMIM
SPAST	11233	NM_014946.3		1-17	Spastic paraplegia 4, autosomal dominant OMIM
SPG11	11226	NM_025137.4		1-40	Amyotrophic lateral sclerosis 5, juvenile OMIM Charcot-Marie-Tooth disease, axonal, type 2X OMIM Spastic paraplegia 11, autosomal recessive OMIM
SPG20	18514	NM_001142294.1		2-9	Troyer syndrome OMIM
SPG21	20373	NM_016630.6		2-9	Mast syndrome OMIM
SPG7	11237	NM_003119.4		1-17	Spastic paraplegia 7, autosomal recessive OMIM
SPR	11257	NM_003124.4		1-3	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency OMIM
SPTBN2	11276	NM_006946.2		2-37	Spinocerebellar ataxia 5 OMIM Spinocerebellar ataxia, autosomal recessive 14 OMIM
SPTLC1	11277	NM_006415.4	3	1-15	Neuropathy, hereditary sensory and autonomic, type IA OMIM
SPTLC2	11278	NM_004863.3		1-12	Neuropathy, hereditary sensory and autonomic, type IC OMIM
SQSTM1	11280	NM_003900.4		1-8	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3 OMIM Myopathy, distal, with rimmed vacuoles OMIM Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset OMIM

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STUB1	11427	NM_005861.4		1-7	?Spinocerebellar ataxia 48 OMIM Spinocerebellar ataxia, autosomal recessive 16 OMIM
SUCLA2	11448	NM_003850.2		1-11	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria) OMIM
SUMF1	20376	NM_182760.3		1-9	Multiple sulfatase deficiency OMIM
SUOX	11460	NM_000456.3		4-6	Sulfite oxidase deficiency OMIM
SURF1	11474	NM_003172.4		1-9	Charcot-Marie-Tooth disease, type 4K OMIM Mitochondrial complex IV deficiency, nuclear type 1 OMIM
SYNE1	17089	NM_033071.3		2-146	Arthrogryposis multiplex congenita 3, myogenic type OMIM Emery-Dreifuss muscular dystrophy 4, autosomal dominant OMIM Spinocerebellar ataxia, autosomal recessive 8 OMIM
SYT14	23143	NM_001146261.2	10	1-10	?Spinocerebellar ataxia, autosomal recessive 11 OMIM
SYT2	11510	NM_177402.4	2	2-9	Myasthenic syndrome, congenital, 7, presynaptic OMIM
TACO1	24316	NM_016360.3		1-5	Mitochondrial complex IV deficiency, nuclear type 8 OMIM
TAF1	11535	NM_004606.4		1-38	Dystonia-Parkinsonism, X-linked OMIM Mental retardation, X-linked, syndromic 33 OMIM
TBC1D24	29203	NM_001199107.1		2-8	Developmental and epileptic encephalopathy 16 OMIM Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp OMIM Myoclonic epilepsy, infantile, familial OMIM

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TCN2	11653	NM_000355.3		1-9	Transcobalamin II deficiency OMIM
TCTN2	25774	NM_024809.4		1-18	Joubert syndrome 24 OMIM
TCTN3	24519	NM_015631.5		1-14	Joubert syndrome 18 OMIM
TDP1	18884	NM_018319.4		3-17	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1 OMIM
TDP2	17768	NM_016614.3		1-7	Spinocerebellar ataxia, autosomal recessive 23 OMIM
TECPR2	19957	NM_014844.3		2-20	Spastic paraplegia 49, autosomal recessive OMIM
TFG	11758	NM_006070.6		2-8	?Spastic paraplegia 57, autosomal recessive OMIM Hereditary motor and sensory neuropathy, Okinawa type OMIM
TGM6	16255	NM_198994.2		1-13	Spinocerebellar ataxia 35 OMIM
TH	11782	NM_199292.2		1-14	Segawa syndrome, recessive OMIM
THAP1	20856	NM_018105.3		1-3	Dystonia 6, torsion OMIM
TIMM8A	11817	NM_004085.3	2	1-2	Mohr-Tranebjaerg syndrome OMIM
TMEM240	25186	NM_001114748.1		1-4	Spinocerebellar ataxia 21 OMIM
TMEM67	28396	NM_153704.5		1-28	COACH syndrome OMIM Joubert syndrome 6 OMIM
TOE1	15954	NM_025077.3		1-8	Pontocerebellar hypoplasia, type 7 OMIM
TOR1A	3098	NM_000113.3		1-5	Arthrogryposis multiplex congenita 5 OMIM Dystonia-1, torsion OMIM

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TPK1	17358	NM_022445.3		2-9	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type) OMIM
TPP1	2073	NM_000391.3		1-13	Ceroid lipofuscinosis, neuronal, 2 OMIM Spinocerebellar ataxia, autosomal recessive 7 OMIM
TREM2	17761	NM_018965.3		1-5	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2 OMIM
TREX1	12269	NM_033629.4		2	{Systemic lupus erythematosus, susceptibility to} OMIM Aicardi-Goutieres syndrome 1, dominant and recessive OMIM Vasculopathy, retinal, with cerebral leukodystrophy OMIM
TRIM2	15974	NM_001130067.1		2-12	Charcot-Marie-Tooth disease, type 2R OMIM
TRPA1	497	NM_007332.2		1-27	?Episodic pain syndrome, familial, 1 OMIM
TRPV4	18083	NM_021625.5		2-16	Brachyolmia type 3 OMIM Digital arthropathy-brachydactyly, familial OMIM Hereditary motor and sensory neuropathy, type IIc OMIM Metatropic dysplasia OMIM Neuronopathy, distal hereditary motor, type VIII OMIM Scapuloperoneal spinal muscular atrophy OMIM
TSEN2	28422	NM_025265.3		2-12	Pontocerebellar hypoplasia type 2B OMIM
TSEN54	27561	NM_207346.2		1-11	?Pontocerebellar hypoplasia type 5 OMIM Pontocerebellar hypoplasia type 2A OMIM Pontocerebellar hypoplasia type 4 OMIM

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TTBK2	19141	NM_173500.4		2-15	Spinocerebellar ataxia 11 OMIM
TTC19	26006	NM_017775.3		1-10	Mitochondrial complex III deficiency, nuclear type 2 OMIM
TTPA	12404	NM_000370.3		1-5	Ataxia with isolated vitamin E deficiency OMIM
TTR	12405	NM_000371.3		1-4	Amyloidosis, hereditary, transthyretin-related OMIM Carpal tunnel syndrome, familial OMIM
TUBB3	20772	NM_001197181.1	4	3-4	Cortical dysplasia, complex, with other brain malformations 1 OMIM Fibrosis of extraocular muscles, congenital, 3A OMIM
TUBB4A	20774	NM_006087.4	4	1-4	Dystonia 4, torsion, autosomal dominant OMIM Leukodystrophy, hypomyelinating, 6 OMIM
TUFM	12420	NM_003321.4		1-10	Combined oxidative phosphorylation deficiency 4 OMIM
TYMP	3148	NM_001953.4		2-10	Mitochondrial DNA depletion syndrome 1 (MNGIE type) OMIM
TYROBP	12449	NM_003332.4		1-5	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1 OMIM
UBA5	23230	NM_024818.4	9-12	1-12	?Spinocerebellar ataxia, autosomal recessive 24 OMIM Developmental and epileptic encephalopathy 44 OMIM
UBTF	12511	NM_014233.4		2-21	Neurodegeneration, childhood-onset, with brain atrophy OMIM
UCHL1	12513	NM_004181.4		1-9	Spastic paraplegia 79, autosomal recessive OMIM
USP18	12616	NM_017414.3	3-11	2-11	Pseudo-TORCH syndrome 2 OMIM

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VAC14	25507	NM_018052.3		1-19	Striatonigral degeneration, childhood-onset OMIM
VAMP1	12642	NM_014231.5		1-5	Myasthenic syndrome, congenital, 25 OMIM Spastic ataxia 1, autosomal dominant OMIM
VAPB	12649	NM_004738.4		1-6	Spinal muscular atrophy, late-onset, Finkel type OMIM
VCP	12666	NM_007126.4		1-17	Charcot-Marie-Tooth disease, type 2Y OMIM Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1 OMIM
VLDLR	12698	NM_003383.5		1-19	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1 OMIM
VPS11	14583	NM_021729.5		1-16	Leukodystrophy, hypomyelinating, 12 OMIM
VPS13A	1908	NM_033305.3		1-72	Choreoacanthocytosis OMIM
VPS13D	23595	NM_015378.3		2-70	Spinocerebellar ataxia, autosomal recessive 4 OMIM
VPS37A	24928	NM_152415.2		1-11	Spastic paraplegia 53, autosomal recessive OMIM
VPS53	25608	NM_001128159.2		1-22	Pontocerebellar hypoplasia, type 2E OMIM
VRK1	12718	NM_003384.2		2-13	Pontocerebellar hypoplasia type 1A OMIM
WARS	12729	NM_004184.3		2-11	Neuronopathy, distal hereditary motor, type IX OMIM
WDR45	28912	NM_007075.3		3-12	Neurodegeneration with brain iron accumulation 5 OMIM

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WDR81	26600	NM_001163809.1		1-10	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2 OMIM Hydrocephalus, congenital, 3, with brain anomalies OMIM
WNK1	14540	NM_018979.3		1-28	Neuropathy, hereditary sensory and autonomic, type II OMIM
WWOX	12799	NM_016373.3		1-9	Developmental and epileptic encephalopathy 28 OMIM Spinocerebellar ataxia, autosomal recessive 12 OMIM
XK	12811	NM_021083.3		1-3	McLeod syndrome with or without chronic granulomatous disease OMIM
XPA	12814	NM_000380.3		1-6	Xeroderma pigmentosum, group A OMIM
XPC	12816	NM_004628.4		1-16	Xeroderma pigmentosum, group C OMIM
XPR1	12827	NM_004736.3		1-15	Basal ganglia calcification, idiopathic, 6 OMIM
XRCC1	12828	NM_006297.2		1-17	?Spinocerebellar ataxia, autosomal recessive 26 OMIM
YARS	12840	NM_003680.3		1-13	Charcot-Marie-Tooth disease, dominant intermediate C OMIM
ZFYVE26	20761	NM_015346.3		2-42	Spastic paraplegia 15, autosomal recessive OMIM