Bevegelsesforstyrrelser

Genpanel, versjon v02

Tabellen er sortert på gennavn (HGNC gensymbol)
Navn på gen er iht. HGNC
Kolonnen **>x10** viser andel av genet som vi forventer blir lest med tilfredstillende kvalitet flere enn 10 ganger under sekvensering

* 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: **ACTB, GBA**

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
AAAS	13666	NM_015665.5		1-16	Achalasia-addisonianism-alacrimia syndrome OMIM
ABCB7	48	NM_004299.5		1-16	Anemia, sideroblastic, with ataxia 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
ACAT1	93	NM_000019.3		1-12	Alpha-methylacetoacetic aciduria OMIM

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ACO2	118	NM_001098.2		1-18	?Optic atrophy 9 OMIM Infantile cerebellar-retinal degeneration OMIM
ACOX1	119	NM_004035.6		1-14	Peroxisomal acyl-CoA oxidase deficiency OMIM
ACTB	132	NM_001101.3	2-6	2-6	?Dystonia, juvenile-onset OMIM Baraitser-Winter syndrome 1 OMIM
ADAR	225	NM_001111.4		1-15	Aicardi-Goutieres syndrome 6 OMIM Dyschromatosis symmetrica hereditaria OMIM
ADCK3	16812	NM_020247.4		2-15	Coenzyme Q10 deficiency, primary, 4 OMIM 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	Ataxia, spastic, 5, autosomal recessive OMIM Spinocerebellar ataxia 28 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
AIMP1	10648	NM_004757.3		2-7	Leukodystrophy, hypomyelinating, 3 OMIM
ALDH18A1	9722	NM_002860.3		2-18	Cutis laxa, autosomal dominant 3 OMIM Cutis laxa, autosomal recessive, type IIIA OMIM Spastic paraplegia 9A, autosomal dominant OMIM Spastic paraplegia 9B, autosomal recessive OMIM
ALDH3A2	403	NM_000382.2		1-10	Sjogren-Larsson syndrome OMIM

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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_014324.5		1-5	Alpha-methylacyl-CoA racemase deficiency OMIM Bile acid synthesis defect, congenital, 4 OMIM
AMN	14604	NM_030943.3		1-12	Adrenoleukodystrophy OMIM Adrenomyeloneuropathy, adult OMIM Megaloblastic anemia-1, Norwegian type OMIM
AMPD2	469	NM_001257360.1		2-19	?Spastic paraplegia 63 OMIM Pontocerebellar hypoplasia, type 9 OMIM
AMT	473	NM_000481.3		1-9	Glycine encephalopathy OMIM
ANO10	25519	NM_018075.4		2-13	Spinocerebellar ataxia, autosomal recessive 10 OMIM
ANO3	14004	NM_031418.3		1-27	Dystonia 24 OMIM
AP1S2	560	NM_003916.4		2-5	Mental retardation, X-linked syndromic 5 OMIM
AP4B1	572	NM_006594.4		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

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AP4S1	575	NM_007077.4		2-6	Spastic paraplegia 52, autosomal recessive OMIM
AP5Z1	22197	NM_014855.2		1-17	Spastic paraplegia 48, autosomal recessive OMIM
APOPT1	20492	NM_032374.4		1-5	Mitochondrial complex IV deficiency OMIM
APTX	15984	NM_175073.2		3-9	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia OMIM
ARG1	663	NM_000045.3		1-8	Argininemia OMIM
ARL13B	25419	NM_182896.2		1-10	Joubert syndrome 8 OMIM
ARSA	713	NM_000487.5		1-8	Metachromatic leukodystrophy OMIM
ASPA	756	NM_000049.2		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
ATM	795	NM_000051.3		2-63	Ataxia-telangiectasia OMIM
ATP13A2	30213	NM_022089.3		1-29	Spastic paraplegia 78, autosomal recessive OMIM Kufor-Rakeb syndrome OMIM
ATP1A2	800	NM_000702.3		1-23	Alternating hemiplegia of childhood OMIM Migraine, familial basilar OMIM Migraine, familial hemiplegic, 2 OMIM
ATP1A3	801	NM_152296.4		1-23	Alternating hemiplegia of childhood 2 OMIM CAPOS syndrome OMIM Dystonia-12 OMIM

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ATP2B3	816	NM_001001344.2		1-20	?Spinocerebellar ataxia, X-linked 1 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
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
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_183050.3		1-10	Maple syrup urine disease, type Ib OMIM
BICD2	17208	NM_001003800.1		1-7	Spinal muscular atrophy, lower extremity-predominant, 2, AD OMIM
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_000060.4		1-4	Biotinidase deficiency OMIM
C10orf2	1160	NM_021830.4		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

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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
C5orf42	25801	NM_023073.3		2-52	Joubert syndrome 17 OMIM Orofaciodigital syndrome VI OMIM
CA8	1382	NM_004056.5		1-8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3 OMIM
CACNA1A	1388	NM_001127221.1		1-47	Epileptic encephalopathy, early infantile, 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
CACNB4	1404	NM_000726.4		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	FG syndrome 4 OMIM Mental retardation and microcephaly with pontine and cerebellar hypoplasia OMIM Mental retardation, with or without nystagmus OMIM
CBS	1550	NM_000071.2		3-17	Homocystinuria, B6-responsive and nonresponsive types OMIM Thrombosis, hyperhomocysteinemic OMIM

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CC2D2A	29253	NM_001080522.2		3-38	COACH syndrome OMIM Joubert syndrome 9 OMIM Meckel syndrome 6 OMIM
CCT5	1618	NM_012073.4		1-11	Neuropathy, hereditary sensory, with spastic paraplegia OMIM
CEP290	29021	NM_025114.3	54	2-54	?Bardet-Biedl syndrome 14 OMIM Joubert syndrome 5 OMIM Leber congenital amaurosis 10 OMIM Meckel syndrome 4 OMIM Senior-Loken syndrome 6 OMIM
CEP41	12370	NM_018718.2		1-11	Joubert syndrome 15 OMIM
CHMP1A	8740	NM_002768.4		1-7	Pontocerebellar hypoplasia, type 8 OMIM
CHORDC1	14525	NM_012124.2	9-11	1-11	Autosomal recessive cerebellar ataxia ARCA PubMed
CLCN1	2019	NM_000083.2		1-23	Myotonia congenita, dominant OMIM Myotonia congenita, recessive OMIM Myotonia levior, recessive
CLCN2	2020	NM_004366.5		1-24	Leukoencephalopathy with ataxia OMIM {Epilepsy, idiopathic generalized, susceptibility to, 11} OMIM {Epilepsy, juvenile absence, susceptibility to, 2} OMIM {Epilepsy, juvenile myoclonic, susceptibility to, 8} 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

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CLN8	2079	NM_018941.3		2-3	Ceroid lipofuscinosis, neuronal, 8 OMIM Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant OMIM
CLP1	16999	NM_006831.2		2-3	Pontocerebellar hypoplasia, type 10 OMIM
CLPP	2084	NM_006012.2		1-6	Perrault syndrome 3 OMIM
COASY	29932	NM_025233.6		1-9	Neurodegeneration with brain iron accumulation 6 OMIM
COL4A1	2202	NM_001845.5		1-52	?Retinal arteries, tortuosity of OMIM Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps OMIM Brain small vessel disease with or without ocular anomalies OMIM Porencephaly 1 OMIM Schizencephaly 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_016035.4		1-7	Coenzyme Q10 deficiency, primary, 7 OMIM
COQ9	25302	NM_020312.3		1-9	Coenzyme Q10 deficiency, primary, 5 OMIM
COX10	2260	NM_001303.3	6	1-7	Leigh syndrome due to mitochondrial COX4 deficiency OMIM Mitochondrial complex IV deficiency OMIM
COX15	2263	NM_004376.6		1-9	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2 OMIM Leigh syndrome due to cytochrome c oxidase deficiency OMIM

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COX20	26970	NM_198076.5		1-4	Mitochondrial complex IV deficiency OMIM
CP	2295	NM_000096.3	19	1-19	Cerebellar ataxia OMIM Hemosiderosis, systemic, due to aceruloplasminemia OMIM
CSF1R	2433	NM_005211.3		2-22	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_025099.5		1-23	Cerebroretinal microangiopathy with calcifications and cysts OMIM
CTDP1	2498	NM_004715.4		1-13	Congenital cataracts, facial dysmorphism, and neuropathy OMIM
CTSD	2529	NM_001909.4		1-9	Ceroid lipofuscinosis, neuronal, 10 OMIM
CTSF	2531	NM_003793.3		1-13	Ceroid lipofuscinosis, neuronal, 13, Kufs type OMIM
CUBN	2548	NM_001081.3	41-50, 61-67	1-67	Megaloblastic anemia-1, Finnish type 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_183075.2		1-5	Spastic paraplegia 56, autosomal recessive OMIM
CYP7B1	2652	NM_004820.4		1-6	Spastic paraplegia 5A, autosomal recessive OMIM
DARS2	25538	NM_018122.4		1-17	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation OMIM
DBT	2698	NM_001918.3		1-11	Maple syrup urine disease, type II OMIM

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DCAF17	25784	NM_025000.3		1-14	Woodhouse-Sakati syndrome 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	Neuropathy, distal hereditary motor, type VIIB OMIM Perry syndrome OMIM
DCX	2714	NM_178153.2		2-7	Lissencephaly, X-linked OMIM Subcortical laminal heterotopia, X-linked OMIM
DDC	2719	NM_000790.3		2-14	Aromatic L-amino acid decarboxylase deficiency OMIM
DDHD1	19714	NM_001160147.1		1-13	Spastic paraplegia 28, autosomal recessive OMIM
DDHD2	29106	NM_015214.2		2-17	Spastic paraplegia 54, autosomal recessive OMIM
DHFR	2861	NM_000791.3	6	1-6	Megaloblastic anemia due to dihydrofolate reductase deficiency OMIM
DLAT	2896	NM_001931.4		1-14	Pyruvate dehydrogenase E2 deficiency OMIM
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_025219.2		2-5	Ceroid lipofuscinosis, neuronal, 4, Parry type OMIM

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DNMT1	2976	NM_001130823.2		1-41	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant OMIM Neuropathy, hereditary sensory, type IE OMIM
EARS2	29419	NM_001083614.1		1-9	Combined oxidative phosphorylation deficiency 12 OMIM
EEF2	3214	NM_001961.3		1-15	?Spinocerebellar ataxia 26 OMIM
EIF2B1	3257	NM_001414.3		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_020365.4		2-12	Leukoencephalopathy with vanishing white matter OMIM
EIF2B4	3260	NM_015636.3		1-13	Leukoencephaly with vanishing white matter OMIM Ovarioleukodystrophy OMIM
EIF2B5	3261	NM_003907.2		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 Stargardt disease 3 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 2B (Lafora) OMIM Epilepsy, progressive myoclonic 2A (Lafora) OMIM

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ERCC3	3435	NM_000122.1		1-15	Trichothiodystrophy 2, photosensitive OMIM Xeroderma pigmentosum, group B OMIM
ERCC4	3436	NM_005236.2		1-11	?XFE progeroid syndrome OMIM Fanconi anemia, complementation group Q OMIM Xeroderma pigmentosum, group F OMIM Xeroderma pigmentosum, type F/Cockayne 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 Premature ovarian failure 11 OMIM UV-sensitive syndrome 1 OMIM
ERCC8	3439	NM_000082.3		1-12	Cockayne syndrome, type A OMIM UV-sensitive syndrome 2 OMIM
ERLIN2	1356	NM_007175.6		2-12	Spastic paraplegia 18, autosomal recessive OMIM
EXOSC3	17944	NM_016042.3		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
FAM126A	24587	NM_032581.3		2-11	Leukodystrophy, hypomyelinating, 5 OMIM

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FAR1	26222	NM_032228.5	12	2-12	Peroxisomal fatty acyl-CoA reductase 1 disorder OMIM
FAT1	3595	NM_005245.3		2-27	Spinocerebellar ataxia PubMed
FAT2	3596	NM_001447.2		1-23	Spinocerebellar ataxia 45 OMIM
FBXL4	13601	NM_012160.4		3-9	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type) OMIM
FBXO7	13586	NM_012179.3		1-9	Parkinson disease 15, autosomal recessive OMIM
FGF14	3671	NM_004115.3		1-5	Spinocerebellar ataxia 27 OMIM
FLVCR1	24682	NM_014053.3		1-10	Ataxia, posterior column, with retinitis pigmentosa OMIM
FOLR1	3791	NM_016725.2		2-5	Neurodegeneration due to cerebral folate transport deficiency OMIM
FOXRED1	26927	NM_017547.3		1-11	Leigh syndrome due to mitochondrial complex I deficiency OMIM Mitochondrial complex I deficiency OMIM
FTL	3999	NM_000146.3		1-4	Hyperferritinemia-cataract syndrome OMIM Neurodegeneration with brain iron accumulation 3 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
GAMT	4136	NM_000156.5		1-6	Cerebral creatine deficiency syndrome 2 OMIM
GAN	4137	NM_022041.3		1-11	Giant axonal neuropathy-1 OMIM

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GBA	4177	NM_001005741.2	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
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_000161.2		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
GCSH	4208	NM_004483.4		1-5	Glycine encephalopathy OMIM
GFAP	4235	NM_002055.4		1-9	Alexander disease OMIM
GIF	4268	NM_005142.2		1-9	Intrinsic factor deficiency OMIM
GJC2	17494	NM_020435.3		2	Leukodystrophy, hypomyelinating, 2 OMIM Spastic paraplegia 44, autosomal recessive 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

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GLRA1	4326	NM_000171.3		1-9	Hyperekplexia, hereditary 1, autosomal dominant or recessive OMIM
GNAL	4388	NM_001142339.2		2-13	Dystonia 25 OMIM
GNAO1	4389	NM_020988.2		1-8	Epileptic encephalopathy, early infantile, 17 OMIM Neurodevelopmental disorder with involuntary movements OMIM
GOSR2	4431	NM_004287.4	3-4	1-6	Epilepsy, progressive myoclonic 6 OMIM
GPR56	4512	NM_005682.6		3-15	Polymicrogyria, bilateral frontoparietal OMIM Polymicrogyria, bilateral perisylvian OMIM
GRID2	4576	NM_001510.3		1-16	Spinocerebellar ataxia, autosomal recessive 18 OMIM
GRM1	4593	NM_001278066.1		1-8	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
GSS	4624	NM_000178.3		2-13	Glutathione synthetase deficiency OMIM Hemolytic anemia due to glutathione synthetase deficiency OMIM
HACE1	21033	NM_020771.3		1-24	Spastic paraplegia and psychomotor retardation with or without seizures OMIM

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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
HEXB	4879	NM_000521.3		1-14	Sandhoff disease, infantile, juvenile, and adult forms OMIM
HMGCL	5005	NM_000191.2		1-9	HMG-CoA lyase deficiency OMIM
HMGCS2	5008	NM_005518.3		1-9	HMG-CoA synthase-2 deficiency OMIM
HPRT1	5157	NM_000194.2		1-9	Lesch-Nyhan syndrome OMIM
HSD17B10	4800	NM_004493.2		1-6	?Mental retardation, X-linked syndromic 10 OMIM 17-beta-hydroxysteroid dehydrogenase X deficiency OMIM
HSD17B4	5213	NM_000414.3		1-24	D-bifunctional protein deficiency OMIM Perrault syndrome 1 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 {Macular degeneration, age-related, 7} OMIM {Macular degeneration, age-related, neovascular type} OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
INPP5E	21474	NM_019892.5		1-10	Joubert syndrome 1 OMIM Mental retardation, truncal obesity, retinal dystrophy, and micropenis OMIM
ITM2B	6174	NM_021999.4		1-6	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities OMIM 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
KCNA1	6218	NM_000217.2		2	Episodic ataxia/myokymia syndrome OMIM
KCNC1	6233	NM_001112741.1		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	Brugada syndrome 9 OMIM Spinocerebellar ataxia 19 OMIM
KCNJ10	6256	NM_002241.4		2	Enlarged vestibular aqueduct, digenic OMIM SESAME syndrome OMIM
KCNMA1	6284	NM_002247.3		1-27	Generalized epilepsy and paroxysmal dyskinesia OMIM
KCTD17	25705	NM_024681.3		1-8	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

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KIF1A	888	NM_004321.7		2-47	Mental retardation, autosomal dominant 9 OMIM Neuropathy, hereditary sensory, type IIC OMIM Spastic paraplegia 30, autosomal recessive OMIM
KIF1C	6317	NM_006612.5	22-23	3-23	Spastic ataxia 2, autosomal recessive OMIM
KIF5A	6323	NM_004984.2		1-28	Spastic paraplegia 10, autosomal dominant OMIM
KIF7	30497	NM_198525.2		2-19	?Al-Gazali-Bakalinova syndrome OMIM ?Hydrolethalus syndrome 2 OMIM Acrocallosal syndrome OMIM Joubert syndrome 12 OMIM
KLC2	20716	NM_001134775.1		2-16	Spastic paraplegia, optic atrophy, and neuropathy OMIM
L1CAM	6470	NM_000425.4		1-28	Corpus callosum, partial agenesis of OMIM CRASH syndrome OMIM Hydrocephalus due to aqueductal stenosis OMIM Hydrocephalus with congenital idiopathic intestinal pseudoobstruction OMIM Hydrocephalus with Hirschsprung disease OMIM MASA syndrome OMIM
L2HGDH	20499	NM_024884.2		1-10	L-2-hydroxyglutaric aciduria OMIM
LAMA1	6481	NM_005559.3		1-63	Poretti-Boltshauser syndrome OMIM
LMBRD1	23038	NM_018368.3		1-16	Methylmalonic aciduria and homocystinuria, cblF type OMIM
LMNB1	6637	NM_005573.3		1-11	Leukodystrophy, adult-onset, autosomal dominant OMIM
LRRK2	18618	NM_198578.3		1-51	{Parkinson disease 8} OMIM

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MAG	6783	NM_002361.3		3-11	Spastic paraplegia 75, autosomal recessive OMIM
MARS2	25133	NM_138395.3		1	?Combined oxidative phosphorylation deficiency 25 OMIM Spastic ataxia 3, autosomal recessive OMIM
MFSD8	28486	NM_152778.2		2-13	Ceroid lipofuscinosis, neuronal, 7 OMIM Macular dystrophy with central cone involvement OMIM
MICU1	1530	NM_006077.3		2-12	Myopathy with extrapyramidal signs OMIM
MKS1	7121	NM_017777.3		1-18	Bardet-Biedl syndrome 13 OMIM Joubert syndrome 28 OMIM Meckel syndrome 1 OMIM
MLC1	17082	NM_015166.3		2-12	Megalencephalic leukoencephalopathy with subcortical cysts OMIM
MMAA	18871	NM_172250.2		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
MPV17	7224	NM_002437.4		2-8	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type) OMIM
MR1	4975	NM_001194999.1	3-4	2-7	Paroxysmal nonkinesigenic dyskinesia 1 OMIM

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MRE11A	7230	NM_005591.3		2-20	Ataxia-telangiectasia-like disorder OMIM
MTHFR	7436	NM_005957.4		2-12	Homocystinuria due to MTHFR deficiency OMIM
MTPAP	25532	NM_018109.3		1-9	Ataxia, spastic, 4 OMIM
MTR	7468	NM_000254.2		1-33	Homocystinuria-megaloblastic anemia, cblG complementation type OMIM
MTRR	7473	NM_002454.2		2-15	{Neural tube defects, folate-sensitive, susceptibility to} OMIM Homocystinuria-megaloblastic anemia, cbl E type OMIM
MTTP	7467	NM_000253.3		2-19	Abetalipoproteinemia OMIM
MUT	7526	NM_000255.3		2-13	Methylmalonic aciduria, mut(0) type OMIM
MVK	7530	NM_000431.3		2-11	Hyper-IgD syndrome OMIM Mevalonic aciduria OMIM Porokeratosis 3, multiple types OMIM
NDUFA10	7684	NM_004544.3		1-10	Leigh syndrome OMIM
NDUFAF5	15899	NM_024120.4		1-11	Mitochondrial complex 1 deficiency OMIM
NEU1	7758	NM_000434.3		1-6	Sialidosis, type I OMIM Sialidosis, type II 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	Chorea, hereditary benign OMIM
NOL3	7869	NM_001276312.1		4-6	Myoclonus, familial cortical OMIM

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NOTCH3	7883	NM_000435.2		1-33	?Myofibromatosis, infantile 2 OMIM Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1 OMIM Lateral meningocele syndrome OMIM
NPC1	7897	NM_000271.4		1-25	Niemann-Pick disease, type C1 OMIM Niemann-Pick disease, type D OMIM
NPC2	14537	NM_006432.3		1-5	Niemann-pick disease, type C2 OMIM
NPHP1	7905	NM_000272.3		1-20	Joubert syndrome 4 OMIM Nephronophthisis 1, juvenile OMIM Senior-Loken syndrome-1 OMIM
NT5C2	8022	NM_012229.4		3-19	Spastic paraplegia 45, autosomal recessive OMIM
NUP62	8066	NM_001193357.1		2	Striatonigral degeneration, infantile OMIM
OPA1	8140	NM_015560.2		1-28	?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type) OMIM Behr syndrome OMIM Optic atrophy 1 OMIM Optic atrophy plus syndrome OMIM
OPHN1	8148	NM_002547.2		2-24	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance OMIM
OXCT1	8527	NM_000436.3		1-17	Succinyl CoA:3-oxoacid CoA transferase deficiency OMIM
PANK2	15894	NM_153638.3		1-7	HARP syndrome OMIM Neurodegeneration with brain iron accumulation 1 OMIM
PARK2	8607	NM_004562.2		1-12	Parkinson disease, juvenile, type 2 OMIM
PARK7	16369	NM_007262.4		2-7	Parkinson disease 7, autosomal recessive early-onset OMIM

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PCBD1	8646	NM_000281.3		1-4	Hyperphenylalaninemia, BH4-deficient, D OMIM
PCCA	8653	NM_000282.3		1-24	Propionicacidemia OMIM
PCCB	8654	NM_000532.4		1-15	Propionicacidemia OMIM
PCDH12	8657	NM_016580.3		1-4	Microcephaly, seizures, spasticity, and brain calcification 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 Dyskinesia, limb and orofacial, infantile-onset OMIM Striatal degeneration, autosomal dominant OMIM
PDE8B	8794	NM_003719.3		1-22	Pigmented nodular adrenocortical disease, primary, 3 OMIM Striatal degeneration, autosomal dominant OMIM
PDGFB	8800	NM_002608.3		1-6	Basal ganglia calcification, idiopathic, 5 OMIM
PDGFRB	8804	NM_002609.3		2-23	Basal ganglia calcification, idiopathic, 4 OMIM
PDHA1	8806	NM_000284.3		1-11	Pyruvate dehydrogenase E1-alpha deficiency OMIM
PDHX	21350	NM_003477.2		1-11	Lacticacidemia due to PDX1 deficiency OMIM
PDSS1	17759	NM_014317.4		1-12	Coenzyme Q10 deficiency, primary, 2 OMIM
PDSS2	23041	NM_020381.3		1-8	Coenzyme Q10 deficiency, primary, 3 OMIM

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PDYN	8820	NM_024411.4		3-4	Spinocerebellar ataxia 23 OMIM
PET100	40038	NM_001171155.1		1-4	Mitochondrial complex IV deficiency 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_002618.3		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
PEX16	8857	NM_004813.2		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

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PEX26	22965	NM_017929.5		2-6	Peroxisome biogenesis disorder 7A (Zellweger) OMIM Peroxisome biogenesis disorder 7B OMIM
PEX3	8858	NM_003630.2		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_000288.3		1-10	Peroxisome biogenesis disorder 9B OMIM Rhizomelic chondrodysplasia punctata, type 1 OMIM
PGAP1	25712	NM_024989.3		1-27	Mental retardation, autosomal recessive 42 OMIM
PGK1	8896	NM_000291.3		1-11	Phosphoglycerate kinase 1 deficiency OMIM
PGM3	8907	NM_001199917.1		2-14	Immunodeficiency 23 OMIM
PHYH	8940	NM_006214.3		1-9	Refsum disease OMIM
PIK3R5	30035	NM_001142633.2		2-19	Ataxia-oculomotor apraxia 3 OMIM
PINK1	14581	NM_032409.2		1-8	Parkinson disease 6, early onset OMIM

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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
PLD3	17158	NM_012268.3		3-13	?Spinocerebellar ataxia 46 OMIM
PLP1	9086	NM_000533.4		1-7	Pelizaeus-Merzbacher disease OMIM Spastic paraplegia 2, X-linked OMIM
PNKD	9153	NM_015488.4		1-10	Paroxysmal nonkinesigenic dyskinesia OMIM
PNKP	9154	NM_007254.3		2-17	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

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POLG2	9180	NM_007215.3		1-8	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4 OMIM
POLR1C	20194	NM_203290.3		1-9	Leukodystrophy, hypomyelinating, 11 OMIM Treacher Collins syndrome 3 OMIM
POLR3A	30074	NM_007055.3		1-31	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism OMIM
POLR3B	30348	NM_018082.5		1-28	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism OMIM
PPT1	9325	NM_000310.3		1-9	Ceroid lipofuscinosis, neuronal, 1 OMIM
PRF1	9360	NM_001083116.1		2-3	Aplastic anemia OMIM Hemophagocytic lymphohistiocytosis, familial, 2 OMIM
PRICKLE1	17019	NM_153026.2		2-8	Epilepsy, progressive myoclonic 1B OMIM
PRKCG	9402	NM_002739.4		1-18	Spinocerebellar ataxia 14 OMIM
PRKRA	9438	NM_003690.4		1-8	Dystonia 16 OMIM
PRNP	9449	NM_000311.4		2	Cerebral amyloid angiopathy, PRNP-related OMIM Creutzfeldt-Jakob disease OMIM Gerstmann-Straussler disease OMIM Insomnia, fatal familial OMIM Prion disease with protracted course OMIM {Kuru, susceptibility to} OMIM

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PRPS1	9462	NM_002764.3	7	1-7	Arts syndrome OMIM Charcot-Marie-Tooth disease, X-linked recessive, 5 OMIM Deafness, X-linked 1 OMIM Gout, PRPS-related OMIM Phosphoribosylpyrophosphate synthetase superactivity OMIM
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
PSAP	9498	NM_002778.3		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 Cardiomyopathy, dilated, 1U OMIM Dementia, frontotemporal OMIM Pick disease OMIM
PTF1A	23734	NM_178161.2		1-2	Pancreatic agenesis 2 OMIM Pancreatic and cerebellar agenesis 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
QDPR	9752	NM_000320.2		1-7	Hyperphenylalaninemia, BH4-deficient, C OMIM

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RAD51	9817	NM_002875.4		2-10	Mirror movements 2 OMIM ?Fanconi anemia, complementation group R OMIM
RARS	9870	NM_002887.3		1-15	Leukodystrophy, hypomyelinating, 9 OMIM
RARS2	21406	NM_020320.4		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_152617.3		1-6	RIDDLE syndrome OMIM
RNF170	25358	NM_001160223.1		2-7	Ataxia, sensory, 1, autosomal dominant OMIM
RNF216	21698	NM_207111.3	2, 6-8	2-17	Cerebellar ataxia and hypogonadotropic hypogonadism OMIM
RPGRIP1L	29168	NM_015272.4		2-27	COACH syndrome OMIM Joubert syndrome 7 OMIM Meckel syndrome 5 OMIM

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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_032730.5		1-9	Optic atrophy 10 with or without ataxia, mental retardation, and seizures OMIM
SACS	10519	NM_014363.5		2-10	Spastic ataxia, Charlevoix-Saguenay type OMIM
SAMHD1	15925	NM_015474.3		1-16	Aicardi-Goutieres syndrome 5 OMIM
SCARB2	1665	NM_005506.3		1-12	Epilepsy, progressive myoclonic 4, with or without renal failure OMIM
SCN1A	10585	NM_001165963.1		1-26	Epilepsy, generalized, with febrile seizures plus, type 2 OMIM Epileptic encephalopathy, early infantile, 6 (Dravet syndrome) OMIM Febrile seizures, familial, 3A OMIM Migraine, familial hemiplegic, 3 OMIM
SCN8A	10596	NM_014191.3		2-27	?Cognitive impairment with or without cerebellar ataxia OMIM Epileptic encephalopathy, early infantile, 13 OMIM Seizures, benign familial infantile, 5 OMIM
SCYL1	14372	NM_020680.3		1-18	Spinocerebellar ataxia, autosomal recessive 21 OMIM
SDHAF1	33867	NM_001042631.2		1	Mitochondrial complex II deficiency OMIM
SEPSECS	30605	NM_016955.3		1-11	Pontocerebellar hypoplasia type 2D OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
SERAC1	21061	NM_032861.3		2-17	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome OMIM
SETX	445	NM_015046.6		3-26	Amyotrophic lateral sclerosis 4, juvenile OMIM Spinocerebellar ataxia, autosomal recessive 1 OMIM
SGCE	10808	NM_003919.2		1-11	Dystonia-11, myoclonic 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_006517.4		1-6	Allan-Herndon-Dudley syndrome OMIM
SLC17A5	10933	NM_012434.4		1-11	Salla disease OMIM Sialic acid storage disorder, infantile OMIM
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_006749.4		2-11	Basal ganglia calcification, idiopathic, 1 OMIM
SLC25A15	10985	NM_014252.3	2, 6-7	2-7	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome OMIM

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SLC2A1	11005	NM_006516.2		1-10	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
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
SLC46A1	30521	NM_080669.5		1-6	Folate malabsorption, hereditary OMIM
SLC52A2	30224	NM_024531.4		2-5	Brown-Vialetto-Van Laere syndrome 2 OMIM
SLC52A3	16187	NM_033409.3		2-5	Brown-Vialetto-Van Laere syndrome 1 OMIM Fazio-Londe disease OMIM
SLC6A1	11042	NM_003042.3		3-16	Myoclonic-atonic epilepsy OMIM
SLC6A3	11049	NM_001044.4		2-15	Parkinsonism-dystonia, infantile OMIM
SLC9A6	11079	NM_006359.2		1-16	Mental retardation, X-linked syndromic, Christianson type OMIM
SMPD1	11120	NM_000543.4		1-6	Niemann-Pick disease, type A OMIM Niemann-Pick disease, type B OMIM
SNCA	11138	NM_000345.3		2-6	Dementia, Lewy body OMIM Parkinson disease 1 OMIM Parkinson disease 4 OMIM
SNX14	14977	NM_020468.5		1-26	Spinocerebellar ataxia, autosomal recessive 20 OMIM

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SPAST	11233	NM_014946.3		1-17	Spastic paraplegia 4, autosomal dominant OMIM
SPG11	11226	NM_025137.3		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_015087.4		2-9	Troyer syndrome OMIM Troyer syndrome OMIM
SPG21	20373	NM_016630.6		2-9	Mast syndrome OMIM
SPG7	11237	NM_003119.3		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
STUB1	11427	NM_005861.3		1-7	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_000456.2		4-6	Sulfite oxidase deficiency OMIM
SURF1	11474	NM_003172.3		1-9	Charcot-Marie-Tooth disease, type 4K OMIM Leigh syndrome, due to COX IV deficiency OMIM

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SYNE1	17089	NM_033071.3		2-146	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
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	Deafness , autosomal recessive 86 OMIM Deafness, autosomal dominant 65 OMIM DOORS syndrome OMIM Epileptic encephalopathy, early infantile, 16 OMIM Myoclonic epilepsy, infantile, familial OMIM
TCN2	11653	NM_000355.3		1-9	Transcobalamin II deficiency OMIM
TCTN1	26113	NM_001082538.2		1-14	Joubert syndrome 13 OMIM
TCTN2	25774	NM_024809.4		1-18	?Meckel syndrome 8 OMIM Joubert syndrome 24 OMIM
TCTN3	24519	NM_015631.5		1-14	Joubert syndrome 18 OMIM Orofaciodigital syndrome IV OMIM
TDP1	18884	NM_018319.3		3-17	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy OMIM
TECPR2	19957	NM_014844.4		2-20	Spastic paraplegia 49, autosomal recessive OMIM
TGM6	16255	NM_198994.2		1-13	Spinocerebellar ataxia 35 OMIM
TH	11782	NM_199292.2		1-14	Segawa syndrome, recessive OMIM

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THAP1	20856	NM_018105.2		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 Meckel syndrome 3 OMIM Nephronophthisis 11 OMIM
TOE1	15954	NM_025077.3		1-8	Pontocerebellar hypoplasia, type 7 OMIM
TOR1A	3098	NM_000113.2		1-5	Dystonia-1, torsion OMIM
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
TREX1	12269	NM_033629.4		2	Aicardi-Goutieres syndrome 1, dominant and recessive OMIM Chilblain lupus OMIM Vasculopathy, retinal, with cerebral leukodystrophy 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
TTBK2	19141	NM_173500.3		2-15	Spinocerebellar ataxia 11 OMIM

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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
TUBB4A	20774	NM_006087.3	4	1-4	Dystonia 4, torsion, autosomal dominant OMIM Leukodystrophy, hypomyelinating, 6 OMIM
UBTF	12511	NM_001076683.1		2-20	Neurodegeneration, childhood-onset, with brain atrophy OMIM
UCHL1	12513	NM_004181.4		1-9	Spastic paraplegia 79, autosomal recessive OMIM {?Parkinson disease 5, susceptibility to} OMIM
VAC14	25507	NM_018052.4		1-19	Striatonigral degeneration, childhood-onset OMIM
VAMP1	12642	NM_014231.4		1-5	Spastic ataxia 1, autosomal dominant OMIM
VLDLR	12698	NM_003383.4		1-19	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1 OMIM
VPS11	14583	NM_021729.5		1-17	Leukodystrophy, hypomyelinating, 12 OMIM
VPS13A	1908	NM_033305.2		1-72	Choreoacanthocytosis 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
WDR45	28912	NM_007075.3		3-12	Neurodegeneration with brain iron acculation 5 OMIM
WDR81	26600	NM_001163809.1		1-10	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2 OMIM

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WWOX	12799	NM_016373.3		1-9	Epileptic encephalopathy, early infantile, 28 OMIM Spinocrebellar ataxia, autosomal recessive 12 OMIM
XK	12811	NM_021083.3		1-3	McLeod syndrome with or without chronic granulomatous disease OMIM
XPR1	12827	NM_004736.3		1-15	Basal ganglia calcification, idiopathic, 6 OMIM
ZFYVE26	20761	NM_015346.3		2-42	Spastic paraplegia 15, autosomal recessive OMIM