Whole Exome Sequencing
Gene package Epilepsy, version 4.1, 31-1-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
AARS	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339	601065	59	100	97	86
ABAT	GABA-transaminase deficiency, 613163	137150	65	100	98	87
ABCC8	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800	600509	68	100	100	95
ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	135	100	100	100
ACTL6B	No OMIM phenotype	612458	111	100	100	100
ACY1	Aminoacylase 1 deficiency, 609924	104620	72	100	100	100
ADSL	Adenylosuccinase deficiency, 103050	608222	61	100	99	88
ALDH7A1	Epilepsy, pyridoxine-dependent, 266100	107323	50	100	93	71
ALG1	Congenital disorder of glycosylation, type Ik, 608540	605907	46	90	76	70
ALG11	Congenital disorder of glycosylation, type Ip, 613661	613666	59	100	100	99

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ALG13	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884	300776	42	100	96	73
ALG3	Congenital disorder of glycosylation, type Id, 601110	608750	63	100	100	100
ALG6	Congenital disorder of glycosylation, type Ic, 603147	604566	58	100	99	83
AMACR	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950	604489	53	100	100	89
AMT	Glycine encephalopathy, 605899	238310	73	100	100	98
ANKRD11	KBG syndrome, 148050	611192	111	100	100	98
AP3B2	Epileptic encephalopathy, early infantile, 48, 617276	602166	97	100	100	98
APOPT1	Mitochondrial complex IV deficiency, 220110	616003	51	100	96	76
ARHGEF9	Epileptic encephalopathy, early infantile, 8, 300607	300429	38	100	95	63
ARID1B	Coffin-Siris syndrome 1, 135900	614556	67	99	97	90
ARV1	Epileptic encephalopathy, early infantile, 38, 617020	611647	81	97	89	84
ARX	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly 2, 300215 Mental retardation 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004	300382	42	89	80	65
ASAH1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950	613468	49	100	97	80
ASL	Argininosuccinic aciduria, 207900	608310	70	100	100	99
ATAD1	Hyperekplexia 4, 618011	614452	36	100	93	64
ATP1A2	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481	182340	86	100	100	99
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235	182350	91	100	100	100
ATP6AP2	Mental retardation, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, 300911	300556	45	100	93	64
ATP7A	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal 3, 300489	300011	44	100	98	84

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ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, 309580	300032	40	100	91	76
AUTS2	Mental retardation 26, 615834	607270	60	100	97	91
BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299	613183	46	100	82	81
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498	614506	74	100	100	98
BTD	Biotinidase deficiency, 253260	609019	70	100	100	98
CACNA1A	Epileptic encephalopathy, early infantile, 42, 617106 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086	601011	63	100	98	90
CACNA1E	No OMIM phenotype	601013	63	100	98	91
CACNA2D2	No OMIM phenotype	607082	72	99	97	94
CACNB4	{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 Episodic ataxia, type 5, 613855	601949	50	100	99	86
CASK	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422	300172	42	100	95	69
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672	300203	46	100	93	78
CERS1	?Epilepsy, progressive myoclonic, 8, 616230	606919	100	89	81	77
CHD2	Epileptic encephalopathy, childhood-onset, 615369	602119	50	100	98	85
CHRNA2	Epilepsy, nocturnal frontal lobe, type 4, 610353	118502	117	100	100	100
CHRNA4	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890	118504	108	100	95	95
CHRNB2	Epilepsy, nocturnal frontal lobe, 3, 605375	118507	91	100	100	97
CLCN4	Mental retardation, X-linked 49/15, 300114	302910	64	100	100	92
CLDN16	Hypomagnesemia 3, renal, 248250	603959	51	100	99	89
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190	610036	75	100	100	100
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	76	100	100	94
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	57	100	100	98
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	78	100	98	88

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CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	81	100	100	100
CNKSR2	Mental retardation, X-linked, syndromic, Houge type, 301008	300724	46	100	93	79
CNNM2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418	607803	106	100	100	95
CNTN2	?Epilepsy, myoclonic, familial adult, 5, 615400	190197	66	100	100	98
CNTNAP2	{Autism susceptibility 15}, 612100 Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042	604569	48	100	99	89
COL4A1	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 {Hemorrhage, intracerebral, susceptibility to}, 614519 Porencephaly 1, 175780 ?Retinal arteries, tortuosity of, 180000	120130	61	100	99	91
COL4A3BP	Mental retardation 34, 616351	604677	44	100	92	74
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500	609825	49	100	97	82
COQ4	Coenzyme Q10 deficiency, primary, 7, 616276	612898	74	100	100	100
COQ8A	Coenzyme Q10 deficiency, primary, 4, 612016	606980	86	100	100	100
CPA6	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418	609562	56	100	96	83
CPS1	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}	608307	42	100	96	78
CPT2	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212	600650	72	100	100	98
CSNK2B	No OMIM phenotype	115441	76	100	100	98
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800	601145	62	100	100	98
CTNND2	No OMIM phenotype	604275	81	96	92	85
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	79	100	100	100
CTSF	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362	603539	70	100	98	93
CUL4B	Mental retardation, syndromic 15 (Cabezas type), 300354	300304	49	100	93	77
D2HGDH	D-2-hydroxyglutaric aciduria, 600721	609186	82	100	100	100

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DCX	Lissencephaly, 300067 Subcortical laminal heteropia, 300067	300121	47	100	96	83
DENND5A	Epileptic encephalopathy, early infantile, 49, 617281	617278	54	100	97	89
DEPDC5	Epilepsy, familial focal, with variable foci 1, 604364	614191	56	100	98	90
DLAT	Pyruvate dehydrogenase E2 deficiency, 245348	608770	53	100	94	71
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350	611203	129	100	100	97
DNM1	Epileptic encephalopathy, early infantile, 31, 616346	602377	82	100	99	92
DOCK7	Epileptic encephalopathy, early infantile, 23, 615859	615730	43	100	96	77
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750	191350	54	100	100	95
DPM1	Congenital disorder of glycosylation, type Ie, 608799	603503	54	93	89	77
DPM2	Congenital disorder of glycosylation, type Iu, 615042	603564	54	100	100	93
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	46	100	99	85
DYNC1H1	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600	600112	69	100	99	95
DYRK1A	Mental retardation 7, 614104	600855	54	100	98	87
EEF1A2	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation 38, 616393	602959	93	100	100	98
EGF	Hypomagnesemia 4, renal, 611718	131530	53	100	98	84
EHMT1	Kleefstra syndrome, 610253	607001	81	99	99	98
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora), 254780	607566	45	89	85	77
FA2H	Spastic paraplegia 35, 612319	611026	56	100	100	92
FARS2	Combined oxidative phosphorylation deficiency 14, 614946 ?Spastic paraplegia 77, 617046	611592	68	100	100	90
FASN	No OMIM phenotype	600212	89	100	100	99
FGD1	Aarskog-Scott syndrome, 305400 Mental retardation syndromic 16, 305400	300546	60	100	99	95

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FLNA	Cardiac valvular dysplasia, 314400 Congenital short bowel syndrome, 300048 FG syndrome 2, 300321 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244	300017	79	100	100	100
FOLR1	Neurodegeneration due to cerebral folate transport deficiency, 613068	136430	85	100	100	100
FOXG1	Rett syndrome, congenital variant, 613454	164874	60	99	91	82
FOXRED1	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010 dominant	613622	64	100	100	100
FRRS1L	Epileptic encephalopathy, early infantile, 37, 616981	604574	39	84	71	58
FXYD2	Hypomagnesemia 2, renal, 154020	601814	62	100	100	100
GABRA1	{Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 Epileptic encephalopathy, early infantile, 19, 615744	137160	52	100	100	90
GABRA3	No OMIM phenotype	305660	37	96	82	61
GABRB3	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 Epileptic encephalopathy, early infantile, 43, 617113	137192	64	100	99	95
GABRG2	{Epilepsy, childhood absence, susceptibility to, 2}, 607681 Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures, familial, 8, 611277	137164	50	92	90	78
GAMT	Cerebral creatine deficiency syndrome 2, 612736	601240	60	100	97	90
GCK	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851	138079	73	100	100	100
GCSH	Glycine encephalopathy, 605899	238330	70	100	91	57
GLDC	Glycine encephalopathy, 605899	238300	50	100	94	82
GLRA1	Hyperekplexia, hereditary 1 or recessive, 149400	138491	58	100	97	86
GLRB	Hyperekplexia 2, 614619	138492	48	100	100	89
GLUD1	Hyperinsulinism-hyperammonemia syndrome, 606762	138130	64	100	93	85

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GNAO1	Epileptic encephalopathy, early infantile, 17, 615473	139311	55	100	98	84
GOSR2	Epilepsy, progressive myoclonic 6, 614018	604027	48	100	91	80
GPC3	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070	300037	42	100	94	79
GPHN	Molybdenum cofactor deficiency C, 615501	603930	48	100	99	84
GRIA3	Mental retardation 94, 300699	305915	42	100	97	80
GRIK2	Mental retardation, autosomal recessive, 6, 611092	138244	79	100	100	96
GRIN1	Mental retardation 8, 614254	138249	75	100	100	97
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570	138253	76	100	100	99
GRIN2B	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation 6, 613970	138252	89	100	100	96
GRIN2D	Epileptic encephalopathy, early infantile, 46, 617162	602717	67	87	74	69
GRN	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485	138945	101	100	100	100
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975	601609	47	100	100	92
HCFC1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541	300019	71	100	98	89
HCN1	Epileptic encephalopathy, early infantile, 24, 615871	602780	80	100	99	93
HDAC4	No OMIM phenotype	605314	70	100	100	98
HLCS	Holocarboxylase synthetase deficiency, 253270	609018	63	100	99	91
HNRNPU	Epileptic encephalopathy, early infantile, 54, 617391	602869	51	100	99	84
HSD17B10	HDS10 mitochondrial disease, 300438	300256	63	100	100	95
HSD17B4	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400	601860	49	100	97	80
HUWE1	Mental retardation, X-linked syndromic, Turner type, 300706	300697	45	100	92	72
IDH2	D-2-hydroxyglutaric aciduria 2, 613657	147650	60	100	100	96
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome, 614231	609382	43	100	86	75
IFIH1	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250	606951	65	100	99	90
INTS8	No OMIM phenotype	611351	44	100	94	74
IQSEC2	Mental retardation 1/78, 309530	300522	56	99	97	88
IRF2BPL	No OMIM phenotype	611720	124	100	98	96
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	606871	49	100	97	83
KANSL1	Koolen-De Vries syndrome, 610443	612452	74	100	100	92

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KCNA1	Episodic ataxia/myokymia syndrome, 160120	176260	83	100	100	100
KCNA2	Epileptic encephalopathy, early infantile, 32, 616366	176262	87	100	100	100
KCNB1	Epileptic encephalopathy, early infantile, 26, 616056	600397	92	100	100	100
KCNC1	Epilepsy, progressive myoclonic 7, 616187	176258	90	100	100	97
KCND3	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346	605411	160	100	100	98
KCNH1	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500	603305	76	100	99	96
KCNJ10	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780	602208	133	100	100	100
KCNJ11	Diabetes mellitus, transient neonatal, 3, 610582 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes, permanent neonatal, with or without neurologic features, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329	600937	134	100	100	100
KCNMA1	Generalized epilepsy and paroxysmal dyskinesia, 609446	600150	58	100	98	87
KCNQ2	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200	602235	81	100	100	97
KCNQ3	Seizures, benign neonatal, type 2, 121201	602232	72	100	98	90
KCNQ5	Mental retardation, autosomal dominant 46, 617601	607357	70	100	99	94
KCNT1	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959	608167	63	100	98	95
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726	611725	102	100	100	99
KDM5C	Mental retardation, syndromic, Claes-Jensen type, 300534	314690	66	100	99	94
KMT2A	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130	159555	60	100	100	95
KPNA7	No OMIM phenotype	614107	84	100	100	99
KPTN	Mental retardation 41, 615637	615620	73	100	100	94
LGI1	Epilepsy, familial temporal lobe, 1, 600512	604619	64	100	99	92
LIAS	Hyperglycinemia, lactic acidosis, and seizures, 614462	607031	56	100	99	88
MBD5	Mental retardation 1, 156200	611472	58	100	100	97
MDH2	Epileptic encephalopathy, early infantile, 51, 617339	154100	85	100	100	100

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MECP2	{Autism susceptibility 3}, 300496 Encephalopathy, neonatal severe, 300673 Mental retardation syndromic, Lubs type, 300260 Mental retardation, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750	300005	88	100	100	94
MED12	Lujan-Fryns syndrome, 309520 Ohdo syndrome, 300895 Opitz-Kaveggia syndrome, 305450	300188	57	100	100	97
MEF2C	Chromosome 5q14.3 deletion syndrome, 613443 (4) Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443	600662	67	100	99	96
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	50	100	98	81
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004	605908	49	100	98	86
MOCS1	Molybdenum cofactor deficiency A, 252150	603707	69	100	100	98
MOCS2	Molybdenum cofactor deficiency B, 252160	603708	50	100	100	90
MPDU1	Congenital disorder of glycosylation, type If, 609180	604041	60	100	100	97
MTHFR	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}	607093	65	100	100	98
MTOR	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638	601231	65	100	98	90
NAPB	No OMIM phenotype	611270	50	100	97	87
NBEA	No OMIM phenotype	604889	60	99	95	84
NDUFA1	Mitochondrial complex I deficiency, 252010 dominant	300078	83	100	100	100
NDUFA11	Mitochondrial complex I deficiency, 252010 dominant	612638	72	100	100	100
NDUFAF1	Mitochondrial complex I deficiency, 252010 dominant	606934	49	100	99	86
NDUFAF2	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010 dominant	609653	63	100	90	77
NDUFAF3	Mitochondrial complex I deficiency, 252010 dominant	612911	79	100	100	100
NDUFAF4	Mitochondrial complex I deficiency, 252010 dominant	611776	72	100	100	100
NDUFAF5	Mitochondrial complex 1 deficiency, 252010 dominant	612360	54	100	100	89

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NDUFB3	Mitochondrial complex I deficiency, 252010 dominant	603839	28	100	81	47
NDUFB9	?Mitochondrial complex I deficiency, 252010 dominant	601445	71	100	100	90
NDUFS1	Mitochondrial complex I deficiency, 252010 dominant	157655	49	100	98	77
NDUFS2	Mitochondrial complex I deficiency, 252010 dominant	602985	59	100	100	97
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010 dominant	603846	73	100	100	100
NDUFS4	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010 dominant	602694	56	100	100	90
NDUFS6	Mitochondrial complex I deficiency, 252010 dominant	603848	52	100	100	96
NDUFV1	Mitochondrial complex I deficiency, 252010 dominant	161015	95	100	100	100
NDUFV2	Mitochondrial complex I deficiency, 252010 dominant	600532	47	100	96	78
NECAP1	?Epileptic encephalopathy, early infantile, 21, 615833	611623	54	100	100	99
NEDD4L	Periventricular nodular heterotopia 7, 617201	606384	50	100	98	85
NEXMIF	Mental retardation 98, 300912	300524	51	100	100	98
NGLY1	Congenital disorder of deglycosylation, 615273	610661	55	100	100	89
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora), 254780	608072	93	100	100	100
NPRL2	Epilepsy, familial focal, with variable foci 2, 617116	607072	79	100	100	100
NPRL3	Epilepsy, familial focal, with variable foci 3, 617118	600928	54	100	97	89
NRXN1	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332	600565	67	100	99	93
NSDHL	CHILD syndrome, 308050 CK syndrome, 300831	300275	62	100	98	88
NUBPL	Mitochondrial complex I deficiency, 252010 dominant	613621	53	100	100	89
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	300170	42	100	97	81
OPHN1	Mental retardation, with cerebellar hypoplasia and distinctive facial appearance, 300486	300127	44	100	92	71
PAK3	Mental retardation 30/47, 300558	300142	41	100	94	69
PC	Pyruvate carboxylase deficiency, 266150	608786	90	100	100	100
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088	300460	92	100	99	92
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	42	99	93	79
PDHB	Pyruvate dehydrogenase E1-beta deficiency, 614111	179060	44	100	98	82
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782	605993	72	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PDX1	{Diabetes mellitus, type II, susceptibility to}, 125853 MODY, type IV, 606392 Pancreatic agenesis 1, 260370	600733	49	100	100	95
PET100	Mitochondrial complex IV deficiency, 220110	614770	75	100	100	81
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539	602136	49	100	98	84
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871	602859	64	100	96	89
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510	601758	54	100	100	86
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885	601789	55	100	100	93
PEX14	Peroxisome biogenesis disorder 13A (Zellweger), 614887	601791	74	100	100	97
PEX16	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877	603360	74	100	95	91
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886	600279	46	100	100	93
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873	608666	76	100	100	100
PEX3	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370	603164	44	100	99	84
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716	600414	76	100	100	98
PEX6	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863	601498	62	100	99	91
PGAP1	Mental retardation, autosomal recessive 42, 615802	611655	56	100	97	83
PGAP3	Hyperphosphatasia with mental retardation syndrome 4, 615716	611801	62	100	100	98
PHF6	Borjeson-Forssman-Lehmann syndrome, 301900	300414	48	100	95	73
PHGDH	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815	606879	103	100	100	100
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818	311770	65	100	100	99
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080	606097	45	100	93	72

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PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749	614730	83	100	100	99
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399	610272	88	100	100	100
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953	603604	80	100	100	99
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722	607120	44	100	96	81
PLP1	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, 312920	300401	74	100	99	87
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	53	100	100	91
PNKP	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402	605610	72	100	100	97
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency, 610090	603287	53	100	100	85
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia 1, 157640 Progressive external ophthalmoplegia 1, 258450	174763	71	100	100	99
PPP2R1A	Mental retardation 36, 616362	605983	86	100	100	100
PPP3CA	Epileptic encephalopathy, infantile or early childhood, 1, 617711	114105	65	100	96	83
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	51	100	100	87
PQBP1	Renpenning syndrome, 309500	300463	92	100	100	100
PRICKLE1	Epilepsy, progressive myoclonic 1B, 612437	608500	61	100	100	95
PRICKLE2	No OMIM phenotype	608501	85	100	100	100
PRIMA1	No OMIM phenotype	613851	57	88	85	82
PRRT2	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751	614386	75	100	100	100
PSAT1	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992	610936	57	100	100	91
PSPH	Phosphoserine phosphatase deficiency, 614023	172480	48	100	94	78
PURA	Mental retardation 31, 616158	600473	126	100	100	100
PYCR2	Leukodystrophy, hypomyelinating, 10, 616420	616406	74	100	100	100
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760	603727	75	100	100	96

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RAB39B	Mental retardation 72, 300271 ?Waisman syndrome, 311510	300774	50	100	100	96
RAI1	Smith-Magenis syndrome, 182290	607642	167	100	100	100
RANBP2	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033	601181	109	100	100	98
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	47	100	97	76
RELN	{Epilepsy, familial temporal lobe, 7}, 616436 Lissencephaly 2 (Norman-Roberts type), 257320	600514	64	100	99	92
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333	606034	73	100	100	99
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	48	100	97	81
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	139	100	100	100
ROGDI	Kohlschutter-Tonz syndrome, 226750	614574	76	100	97	94
RPS6KA3	Coffin-Lowry syndrome, 303600 Mental retardation 19, 300844	300075	38	100	85	57
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions 5, 613077	604712	60	100	100	95
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	49	100	93	69
SCARB2	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900	602257	60	100	98	88
SCN1A	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6, 607208 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634	182389	67	100	100	94
SCN1B	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Epileptic encephalopathy, early infantile, 52, 617350	600235	105	100	93	93
SCN2A	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745	182390	73	100	99	95
SCN8A	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080	600702	72	100	99	91
SHANK3	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950	606230	106	98	91	84

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SIK1	Epileptic encephalopathy, early infantile, 30, 616341	605705	84	100	100	100
SLC12A5	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685 Epileptic encephalopathy, early infantile, 34, 616645	606726	99	100	100	98
SLC13A5	Epileptic encephalopathy, early infantile, 25, 615905	608305	78	100	98	92
SLC16A1	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095	600682	61	100	100	96
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483	606152	54	100	100	94
SLC1A3	Episodic ataxia, type 6, 612656	600111	90	100	100	100
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182	190315	66	100	100	100
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	603861	85	100	100	94
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304	609302	81	100	100	100
SLC2A1	Dystonia 9, 601042 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885	138140	94	100	100	100
SLC35A2	Congenital disorder of glycosylation, type IIm, 300896	314375	55	100	100	98
SLC6A1	Myoclonic-atonic epilepsy, 616421	137165	73	100	100	96
SLC6A5	Hyperekplexia 3, 614618	604159	80	100	99	95
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352	300036	79	100	97	92
SLC9A6	Mental retardation syndromic, Christianson type, 300243	300231	54	100	94	82
SMARCA2	Nicolaides-Baraitser syndrome, 601358	600014	61	98	96	85
SMC1A	Cornelia de Lange syndrome 2, 300590	300040	59	100	98	95
SMS	Mental retardation, Snyder-Robinson type, 309583	300105	41	95	80	70
SNAP25	?Myasthenic syndrome, congenital, 18, 616330	600322	78	100	100	92
SON	ZTTK syndrome, 617140	182465	77	99	95	88
SPTAN1	Epileptic encephalopathy, early infantile, 5, 613477	182810	64	100	99	94
SRPX2	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643	300642	47	100	97	85
ST3GAL3	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation 12, 611090	606494	52	100	98	87
ST3GAL5	Salt and pepper developmental regression syndrome, 609056	604402	40	94	84	61
STX1B	Generalized epilepsy with febrile seizures plus, type 9, 616172	601485	81	100	99	91
STXBP1	Epileptic encephalopathy, early infantile, 4, 612164	602926	53	100	99	86

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SUOX	Sulfite oxidase deficiency, 272300	606887	106	100	100	100
SYN1	Epilepsy, with variable learning disabilities and behavior disorders, 300491 dominant	313440	58	100	100	97
SYNGAP1	Mental retardation 5, 612621	603384	93	98	98	97
SYNJ1	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530	604297	53	100	98	88
SYP	Mental retardation 96, 300802	313475	50	100	98	87
SZT2	Epileptic encephalopathy, early infantile, 18, 615476	615463	77	100	100	98
TANGO2	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878	616830	61	100	100	93
TBC1D24	DOOR syndrome, 220500 Deafness 86, 614617 Deafness 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021	613577	87	100	100	98
TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460	604934	44	98	90	70
TBCK	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900	616899	45	100	97	78
TCF4	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954	602272	50	100	97	87
TDP2	Spinocerebellar ataxia 23, 616949	605764	80	100	100	92
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia 7, 609270	607998	70	100	100	99
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 {Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukodystrophy, 192315	606609	116	100	100	100
TRIO	Mental retardation, autosomal dominant 44, 617061	601893	89	99	99	96
TRPM6	Hypomagnesemia 1, intestinal, 602014	607009	55	100	97	89
TSC1	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100	605284	105	100	100	100
TSC2	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254	191092	104	100	100	100

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TSEN54	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204	608755	67	100	96	95
TUBA1A	Lissencephaly 3, 611603	602529	89	100	100	100
TUBB2A	Cortical dysplasia, complex, with other brain malformations 5, 615763	615101	170	100	97	95
TUBG1	Cortical dysplasia, complex, with other brain malformations 4, 615412	191135	99	100	100	100
UBA5	Epileptic encephalopathy, early infantile, 44, 617132 ?Spinocerebellar ataxia, autosomal recessive 24, 617133	610552	41	100	87	62
UBE2A	Mental retardation, X-linked syndromic, Nascimento-type, 300860	312180	52	100	93	75
UBE3A	Angelman syndrome, 105830	601623	53	100	100	93
UGDH	No OMIM phenotype	603370	40	100	97	79
WDR45	Neurodegeneration with brain iron accumulation 5, 300894	300526	80	100	100	99
WWOX	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia 12, 614322	605131	57	100	100	93
XK	McLeod syndrome with or without chronic granulomatous disease, 300842	314850	51	100	100	94
YWHAG	Epileptic encephalopathy, early infantile, 56, 617665	605356	105	100	100	100
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type, 300799	300646	39	100	97	76
ZEB2	Mowat-Wilson syndrome, 235730	605802	61	100	100	98

- Gene symbols according HGCN
- OMIM release used: 2-6-2017
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- The statistics above are based on a set of 96 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x