Whole Exome Sequencing
Gene package Epilepsy, version 1, 8-4-2015

Technical information
After DNA was enriched using Agilent Sureselect Clinical Research Exome (CRE) Capture, samples were run on the Illumina Hiseq platform. The aim is to obtain 50 million total reads per exome with a mapped fraction >0.98. The average coverage of the exome is ~50x. Data are demultiplexed by Illumina software bcl2fastq. Reads are mapped to the genome using BWA (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by Genome Analysis Toolkit (reference: http://www.broadinstitute.org/gatk/). Analysis is performed in Cartagenia using The Variant Calling File (VCF) followed by filtering. 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
ABAT	GABA-transaminase deficiency, 613163	137150	77	100	99
ABCC8	Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800 Diabetes mellitus, transient neonatal 2, 610374 Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176	600509	83	100	100
ACY1	Aminoacylase 1 deficiency, 609924	104620	84	100	100
ADSL	Adenylosuccinase deficiency, 103050	608222	86	100	100
ALDH7A1	Epilepsy, pyridoxine-dependent, 266100	107323	76	100	99
ALG13	Epileptic encephalopathy, early infantile, 36, 300884	300776	30	88	60
AMACR	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950	604489	81	100	98
AMT	Glycine encephalopathy, 605899	238310	91	100	100
ARHGEF9	Epileptic encephalopathy, early infantile, 8, 300607	300429	47	100	87
ARX	Epileptic encephalopathy, early infantile, 1, 308350 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Proud syndrome, 300004 Partington syndrome, 309510 Hydranencephaly with abnormal genitalia, 300215	300382	24	84	61

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ASAH1	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950	613468	77	100	99
ATP1A2	Migraine, familial hemiplegic, 2, 602481 Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481	182340	109	100	100
ATP6AP2	?Mental retardation, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911	300556	39	100	90
ATP7A	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489	300011	53	99	94
ATRX	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580	300032	38	98	83
BOLA3	Multiple mitochondrial dysfunctions syndrome 2, 614299	613183	38	83	63
BTD	Biotinidase deficiency, 253260	609019	105	100	100
CACNA1A	Migraine, familial hemiplegic, 1, 141500 Episodic ataxia, type 2, 108500 Spinocerebellar ataxia 6, 183086 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500	601011	74	100	95
CASK	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422 Mental retardation, with or without nystagmus, 300422	300172	39	98	87
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672	300203	47	98	94
CHD2	Epileptic encephalopathy, childhood-onset, 615369	602119	75	100	98
CHRNA2	Epilepsy, nocturnal frontal lobe, type 4, 610353	118502	119	100	100
CHRNA4	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890	118504	91	97	94
CHRNB2	Epilepsy, nocturnal frontal lobe, 3, 605375	118507	102	100	97
CLDN16	Hypomagnesemia 3, renal, 248250	603959	92	100	100
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190	610036	79	100	97
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	68	100	100
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	73	100	94
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	72	100	91
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	100	100	100
CNNM2	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418	607803	113	100	100
CNTN2	?Epilepsy, myoclonic, familial adult, 5, 615400	190197	60	100	95

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CNTNAP2	Cortical dysplasia-focal epilepsy syndrome, 610042 {Autism susceptibility 15}, 612100 Pitt-Hopkins like syndrome 1, 610042	604569	85	100	100
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500	609825	60	100	94
CPA6	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418	609562	104	100	100
CPS1	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}	608307	78	100	99
CPT2	Myopathy due to CPT II deficiency, 255110 CPT deficiency, hepatic, type II, 600649 CPT II deficiency, lethal neonatal, 608836 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212	600650	120	99	96
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800	601145	124	100	97
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	83	100	96
CTSF	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362	603539	43	82	70
CUL4B	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354	300304	39	100	92
D2HGDH	D-2-hydroxyglutaric aciduria, 600721	609186	91	100	100
DCX	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067	300121	47	98	85
DEPDC5	Epilepsy, familial focal, with variable foci, 604364	614191	53	99	90
DLAT	Pyruvate dehydrogenase E2 deficiency, 245348	608770	62	100	95
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350	611203	111	100	100
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	79	98	95
DYNC1H1	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600	600112	86	100	99
DYRK1A	Mental retardation, autosomal dominant 7, 614104	600855	99	100	100
EEF1A2	Mental retardation, autosomal dominant 38, 616393 Epileptic encephalopathy, early infantile, 33, 616409	602959	78	100	96
EGF	Hypomagnesemia 4, renal, 611718	131530	80	100	100
EHMT1	Kleefstra syndrome, 610253	607001	97	99	99
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora), 254780	607566	66	87	83
FARS2	Combined oxidative phosphorylation deficiency 14, 614946	611592	101	100	100
FGD1	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400	300546	43	99	87

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FLNA	Heterotopia, periventricular, 300049 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, ED variant, 300537 FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048	300017	55	100	99
FOLR1	Neurodegeneration due to cerebral folate transport deficiency, 613068	136430	94	100	100
FOXG1	Rett syndrome, congenital variant, 613454	164874	80	83	77
FOXRED1	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010	613622	74	100	100
FXYD2	Hypomagnesemia 2, renal, 154020	601814	49	100	91
GABRA1	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 Epileptic encephalopathy, early infantile, 19, 615744	137160	95	100	100
GABRG2	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681 Febrile seizures, familial, 8, 611277	137164	107	99	92
GAMT	Cerebral creatine deficiency syndrome 2, 612736	601240	59	99	89
GCK	MODY, type II, 125851 Diabetes mellitus, noninsulin-dependent, late onset, 125853 Hyperinsulinemic hypoglycemia, familial, 3, 602485 Diabetes mellitus, permanent neonatal, 606176	138079	84	100	100
GCSH	Glycine encephalopathy, 605899	238330	14	61	18
GLDC	Glycine encephalopathy, 605899	238300	36	81	62
GLRA1	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400	138491	73	100	99
GLRB	Hyperekplexia 2, autosomal recessive, 614619	138492	65	100	99
GLUD1	Hyperinsulinism-hyperammonemia syndrome, 606762	138130	25	80	50
GNAO1	Epileptic encephalopathy, early infantile, 17, 615473	139311	66	100	99
GPC3	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070	300037	38	98	92
GPHN	Molybdenum cofactor deficiency C, 615501	603930	80	100	98
GRIA3	Mental retardation, X-linked 94, 300699	305915	54	100	98
GRIN1	Mental retardation, autosomal dominant 8, 614254	138249	96	100	99
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570	138253	80	100	100

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GRIN2B	Mental retardation, autosomal dominant 6, 613970 Epileptic encephalopathy, early infantile, 27, 616139	138252	95	100	100
GRN	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706	138945	95	100	100
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975	601609	85	100	100
HDAC4	No OMIM phenotype	605314	73	100	100
HLCS	Holocarboxylase synthetase deficiency, 253270	609018	88	100	100
HNRNPU	No OMIM phenotype	602869	73	100	97
HSD17B10	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220	300256	63	100	100
HSD17B4	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400	601860	63	99	96
IDH2	D-2-hydroxyglutaric aciduria 2, 613657	147650	79	100	98
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome, 614231	609382	60	78	78
IQSEC2	Mental retardation, X-linked 1, 309530	300522	38	94	84
KANSL1	Koolen-De Vries syndrome, 610443	612452	65	97	86
KCNA1	Episodic ataxia/myokymia syndrome, 160120	176260	81	100	100
KCNJ10	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791	602208	122	100	100
KCNJ11	Hyperinsulinemic hypoglycemia, familial, 2, 601820 Diabetes, permanent neonatal, 606176 Diabetes mellitus, permanent neonatal, with neurologic features, 606176 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes mellitus, transient neonatal, 3, 610582 Maturity-onset diabetes of the young, type 13, 616329	600937	104	100	100
KCNMA1	Generalized epilepsy and paroxysmal dyskinesia, 609446	600150	73	100	97
KCNQ2	Seizures, benign neonatal, 1, 121200 Myokymia, 121200 Epileptic encephalopathy, early infantile, 7, 613720	602235	70	100	100
KCNQ3	Seizures, benign neonatal, type 2, 121201	602232	85	100	96
KCNT1	Epileptic encephalopathy, early infantile, 14, 614959 Epilepsy, nocturnal frontal lobe, 5, 615005	608167	86	100	99
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726	611725	90	100	100
KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534	314690	52	100	96
LGI1	Epilepsy, familial temporal lobe, 1, 600512	604619	86	100	100
LIAS	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462	607031	85	100	97
MBD5	Mental retardation, autosomal dominant 1, 156200	611472	82	100	100

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MECP2	Rett syndrome, 312750 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, preserved speech variant, 312750 Encephalopathy, neonatal severe, 300673 {Autism susceptibility, X-linked 3}, 300496 Mental retardation, X-linked syndromic, Lubs type, 300260 Rett syndrome, atypical, 312750	300005	52	99	81
MED12	Opitz-Kaveggia syndrome, 305450 Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895	300188	47	100	91
MEF2C	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 Chromosome 5q14.3 deletion syndrome, 613443	600662	66	100	98
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	75	100	97
MOCS1	Molybdenum cofactor deficiency A, 252150	603707	67	93	77
MOCS2	Molybdenum cofactor deficiency B, 252160	603708	73	100	100
MTHFR	Homocystinuria due to MTHFR deficiency, 236250 {Schizophrenia, susceptibility to}, 181500 {Vascular disease, susceptibility to} {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050	607093	94	100	100
MTOR	Smith-Kingsmore syndrome, 616638	601231	47	98	86
NDUFA1	Mitochondrial complex I deficiency, 252010	300078	85	100	90
NDUFA11	Mitochondrial complex I deficiency, 252010	612638	57	100	96
NDUFAF1	Mitochondrial complex I deficiency, 252010	606934	81	100	100
NDUFAF2	Mitochondrial complex I deficiency, 252010 Leigh syndrome, 256000	609653	14	65	23
NDUFAF3	Mitochondrial complex I deficiency, 252010	612911	83	100	100
NDUFAF4	Mitochondrial complex I deficiency, 252010	611776	71	100	100
NDUFAF5	Mitochondrial complex 1 deficiency, 252010	612360	67	100	95
NDUFB3	Mitochondrial complex I deficiency, 252010	603839	47	65	48
NDUFB9	?Mitochondrial complex I deficiency, 252010	601445	90	100	100
NDUFS1	Mitochondrial complex I deficiency, 252010	157655	76	100	98
NDUFS2	Mitochondrial complex I deficiency, 252010	602985	89	100	100
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010	603846	126	100	100
NDUFS4	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010	602694	81	100	100
NDUFS6	Mitochondrial complex I deficiency, 252010	603848	65	100	85

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NDUFV1	Mitochondrial complex I deficiency, 252010	161015	92	100	100
NDUFV2	Mitochondrial complex I deficiency, 252010	600532	30	68	36
NECAP1	?Epileptic encephalopathy, early infantile, 21, 615833	611623	45	100	93
NEDD4L	No OMIM phenotype	606384	84	100	97
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora), 254780	608072	86	100	100
NRXN1	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332	600565	102	100	99
NUBPL	Mitochondrial complex I deficiency, 252010	613621	55	100	92
OFD1	Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804 ?Retinitis pigmentosa 23, 300424	300170	43	99	88
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486	300127	42	100	97
PAK3	Mental retardation, X-linked 30/47, 300558	300142	41	99	86
PC	Pyruvate carboxylase deficiency, 266150	608786	84	100	100
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088	300460	57	100	97
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	64	99	92
PDHB	Pyruvate dehydrogenase E1-beta deficiency, 614111	179060	79	100	100
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782	605993	83	100	100
PEX1	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Heimler syndrome 1, 234580	602136	67	100	98
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871	602859	60	95	89
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510	601758	104	100	100
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885	601789	91	100	100
PEX14	Peroxisome biogenesis disorder 13A (Zellweger), 614887	601791	85	100	100
PEX16	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877	603360	85	100	95
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886	600279	75	100	100
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873	608666	94	100	100
PEX3	Peroxisome biogenesis disorder 10A (Zellweger), 614882	603164	58	100	98
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716	600414	93	100	100

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PEX6	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617	601498	71	95	91
PHF6	Borjeson-Forssman-Lehmann syndrome, 301900	300414	31	98	73
PHGDH	Phosphoglycerate dehydrogenase deficiency, 601815 Neu-Laxova syndrome 1, 256520	606879	86	100	99
PIGA	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868	311770	41	93	83
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080	606097	66	98	93
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749	614730	82	100	100
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953	603604	88	100	99
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722	607120	76	100	99
PLP1	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920	300401	71	100	99
PNKP	Microcephaly, seizures, and developmental delay, 613402 Ataxia-oculomotor apraxia 4, 616267	605610	61	100	98
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency, 610090	603287	66	100	99
POLG	Progressive external ophthalmoplegia, autosomal recessive 1, 258450 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459	174763	86	100	100
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	81	100	100
PQBP1	Renpenning syndrome, 309500	300463	62	100	100
PRICKLE1	Epilepsy, progressive myoclonic 1B, 612437	608500	73	100	97
PRRT2	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066	614386	58	100	100
RAB39B	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510	300774	48	100	97
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	71	100	95
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333	606034	89	100	100
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	50	100	88
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	112	100	100
ROGDI	Kohlschutter-Tonz syndrome, 226750	614574	63	100	98
RPS6KA3	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844	300075	40	96	81

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RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075	604712	87	100	100
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	76	100	94
SCARB2	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900	602257	88	100	100
SCN1A	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Dravet syndrome, 607208 Migraine, familial hemiplegic, 3, 609634 Febrile seizures, familial, 3A, 604403	182389	100	100	99
SCN1B	Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Atrial fibrillation, familial, 13, 615377	600235	102	97	95
SCN2A	Seizures, benign familial infantile, 3, 607745 Epileptic encephalopathy, early infantile, 11, 613721	182390	100	100	99
SCN8A	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558	600702	97	100	99
SCN9A	Erythermalgia, primary, 133020 Paroxysmal extreme pain disorder, 167400, Insensitivity to pain, congenital, 243000 Febrile seizures, familial, 3B, 613863 Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Small fiber neuropathy, 133020 {Dravet syndrome, modifier of}, 607208 HSAN2D, autosomal recessive, 243000	603415	98	100	98
SLC16A1	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095	600682	76	100	100
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483	606152	118	100	100
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182	190315	40	99	90
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	603861	106	90	86
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304	609302	63	100	100

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SLC2A1	GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 Dystonia 9, 601042 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885	138140	104	100	100
SLC35A2	Congenital disorder of glycosylation, type IIm, 300896	314375	29	98	80
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352	300036	19	81	42
SLC9A6	Mental retardation, X-linked syndromic, Christianson type, 300243	300231	50	100	96
SMS	Mental retardation, X-linked, Snyder-Robinson type, 309583	300105	39	95	94
SPTAN1	Epileptic encephalopathy, early infantile, 5, 613477	182810	87	100	100
SRPX2	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643	300642	50	99	91
ST3GAL3	Mental retardation, autosomal recessive 12, 611090 Epileptic encephalopathy, early infantile, 15, 615006	606494	111	100	100
STXBP1	Epileptic encephalopathy, early infantile, 4, 612164	602926	92	100	100
SUOX	Sulfite oxidase deficiency, 272300	606887	104	100	100
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491	313440	37	86	67
SYNGAP1	Mental retardation, autosomal dominant 5, 612621	603384	55	95	82
SYP	Mental retardation, X-linked 96, 300802	313475	48	100	100
SZT2	Epileptic encephalopathy, early infantile, 18, 615476	615463	57	99	93
TBC1D24	Myoclonic epilepsy, infantile, familial, 605021 Epileptic encephalopathy, early infantile, 16, 615338 DOOR syndrome, 220500 Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044	613577	92	100	100
TBCE	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410	604934	80	99	91
TCF4	Pitt-Hopkins syndrome, 610954 Corneal dystrophy, Fuchs endothelial, 3, 613267	602272	73	100	98
TDP2	No OMIM phenotype	605764	73	100	91
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270	607998	79	100	98
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700	606609	129	100	100
TRPM6	Hypomagnesemia 1, intestinal, 602014	607009	81	100	99
UBE3A	Angelman syndrome, 105830	601623	61	100	99
ZEB2	Mowat-Wilson syndrome, 235730	605802	81	100	100

- Gene symbols according HGCN

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x

- Gene symbols according HGCN
- OMIM release used: 17-3-2016
- "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 50 samples
- Median depth is the median of the mean sequence depth over the protein coding exons of the transcript
- % Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x