Epilepsy Panel Genes and Disorders

| GENE | Transcript | *Covered 10X (%) | OMIM ID | OMIM Phenotype |
|---|---|---|---|---|
| *ABAT* | NM_020686.5 | 100 | 137150 | GABA-transaminase deficiency |
| *ADGRG1* | NM_001145771.2 | 100 | 604110 | Polymicrogyria, bilateral frontoparietal; Polymicrogyria, bilateral perisylvian |
| *ADGRV1* | NM_032119.3 | 99.93 | 602851 | Febrile seizures, familial, 4; Usher syndrome, type 2C |
| *ADRA2B* | NM_000682.5 | 100 | 104260 | Autosomal dominant cortical myoclonus and epilepsy (ADCME) |
| *ADSL* | NM_000026.2 | 100 | 608222 | Adenylosuccinase deficiency |
| *AFG3L2* | NM_006796.2 | 98.16 | 604581 | Ataxia, spastic, 5, autosomal recessive; Spinocerebellar ataxia 28 |
| *AKT3* | NM_005465.4 | 99.97 | 611223 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome |
| *ALDH7A1*** | NM_001182.3 | 93.05 (100% with Sanger Gap fill) | 107323 | Epilepsy, pyridoxine-dependent |
| *ALG13* | NM_018466.5 | 98.76 | 300776 | Congenital disorder of glycosylation, type Is |
| *ARFGEF2* | NM_006420.2 | 100 | 605371 | Periventricular heterotopia with microcephaly |
| *ARHGEF9* | NM_015185.3 | 99.96 | 300430 | Epileptic encephalopathy, early infantile, 8 |
| *ARX*** | NM_139058.2 | 98.96 (100% with Sanger Gap fill) | 300382 | Epileptic encephalopathy, early infantile, 1; Hydranencephaly with abnormal genitalia; Lissencephaly, X-linked 2; Mental retardation, X-linked; Partington syndrome; Proud syndrome |
| *ASAH1* | NM_177924.3 | 100 | 613468 | Farber lipogranulomatosis; Spinal muscular atrophy with progressive myoclonic epilepsy |
| *ASPM* | NM_018136.4 | 99.88 | 605481 | Microcephaly 5, primary, autosomal recessive |
| *ATP1A2* | NM_000702.3 | 100 | 182340 | Alternating hemiplegia of childhood; Migraine, familial basilar; Migraine, familial hemiplegic, 2 |
| *BCKDK* | NM_005881.2 | 100 | 614901 | Branched-chain ketoacid dehydrogenase kinase deficiency |
| *C12orf57* | NM_138425.2 | 100 | 615140 | Temtamy syndrome |
| *CACNA1A* | NM_001127221.1 | 99.15 | 601011 | Migraine, familial hemiplegic; Migraine, familial hemiplegic with progressive cerebellar ataxia |
| *CACNA1H* | NM_021098.2 | 99.57 | 607904 | Epilepsy, childhood absence, susceptibility to, 6; Epilepsy, idiopathic generalized, susceptibility to, 6 |
| *CACNB4* | NM_000726.3 | 100 | 601949 | Episodic ataxia, type 5; Epilepsy, idiopathic generalized, susceptibility to, 9; Epilepsy, juvenile myoclonic, susceptibility to, 6 |
| *CASK* | NM_003688.3 | 99.9 | 300172 | FG syndrome 4; Mental retardation and microcephaly with pontine and cerebellar hypoplasia; Mental retardation, with or without nystagmus |
| *CASR* | NM_000388.3 | 100 | 601199 | Hyperparathyroidism, neonatal; Hypocalcemia, AD; Hypocalcemia, AD with Bartter syndrome; Hypocalciuric hypercalcemia, type I; Epilepsy idiopathic generalized, susceptibility to, 8 |
| *CDK5RAP2* | NM_018249.5 | 99.99 | 608201 | Microcephaly 3, primary, autosomal recessive |
| *CDKL5*** | NM_003159.2 | 99.97 | 300203 | Epileptic encephalopathy, early infantile, 2 |

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| *CHD2* | NM_001271.3 | 100 | 602119 | Epileptic encephalopathy, childhood-onset |
| *CHRNA2* | NM_000742.3 | 100 | 118502 | Epilepsy, nocturnal frontal lobe, type 4 |
| *CHRNA4* | NM_000744.6 | 99.86 | 118504 | Epilepsy, nocturnal frontal lobe, 1; Nicotine addiction, susceptibility to |
| *CHRNA7* | NM_000746.5 | 99.21 | 118511 | Schizophrenia, neurophysiologic defect; Epilepsy, idiopathic generalized, susceptibility to, 7 |
| *CHRNB2* | NM_000748.2 | 100 | 118507 | Epilepsy, nocturnal frontal lobe, 3 |
| *CLCN2* | NM_004366.5 | 100 | 600570 | Leukoencephalopathy with ataxia; Epilepsy, idiopathic generalized, susceptibility to, 11; Epilepsy, juvenile absence, susceptibility to, 2; Epilepsy, juvenile myoclonic, susceptibility to, 8 |
| *CLN3* | NM_001042432.1 | 100 | 607042 | Ceroid lipofuscinosis, neuronal, 3 |
| *CLN5* | NM_006493.2 | 100 | 608102 | Ceroid lipofuscinosis, neuronal, 5 |
| *CLN6* | NM_017882.2 | 99.95 | 606725 | Ceroid lipofuscinosis, neuronal, 6; Ceroid lipofuscinosis, neuronal, adult onset |
| *CLN8* | NM_018941.3 | 100 | 607837 | Ceroid lipofuscinosis, neuronal, 8; Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant |
| *CNTNAP2* | NM_014141.5 | 100 | 604569 | Cortical dysplasia-focal epilepsy syndrome; Pitt-Hopkins like syndrome 1; Autism susceptibility 15 |
| *COL4A1* | NM_001845.4 | 99.91 | 120130 | Angiopathy, hereditary, with nephropathy, aneurysms, and muscle; Brain small vessel disease with Axenfeld-Rieger anomaly; Brain small vessel disease with hemorrhage; Porencephaly 1; Hemorrhage, intracerebral, susceptibility to |
| *CPA6* | NM_020361.4 | 100 | 609562 | Epilepsy, familial temporal lobe, 5; Febrile seizures, familial, 11 |
| *CSTB* | NM_000100.3 | 100 | 601145 | Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg) |
| *CTSD* | NM_001909.4 | 100 | 116840 | Ceroid lipofuscinosis, neuronal, 10 |
| *CUL4B* | NM_003588.3 | 99.58 | 300304 | Mental retardation, X-linked, syndromic 15 (Cabezas type) |
| *DCX* | NM_178151.2 | 100 | 300121 | Lissencephaly, X-linked; Subcortical laminal heteropia, X-linked |
| *DEPDC5* | NM_001242896.1 | 99.6 | 614191 | Epilepsy, familial focal, with variable foci |
| *DNAJC5* | NM_025219.2 | 100 | 611203 | Ceroid lipofuscinosis, neuronal, 4, Parry type |
| *DNM1* | NM_004408.3 | 99.96 | 616346 | Epileptic encephalopathy, early infantile, 31 |
| *DOCK7* | NM_033407.2 | 99.96 | 615730 | Epileptic encephalopathy, early infantile, 23 |
| *DYNC1H1* | NM_001376.4 | 100 | 600112 | Charcot-Marie-Tooth disease, axonal, type 20; Mental retardation, autosomal dominant 13; Spinal muscular atrophy, lower extremity-predominant, AD |
| *DYRK1A* | NM_001396.3 | 100 | 600855 | Mental retardation, autosomal dominant 7 |
| *EEF1A2* | NM_001958.3 | 100 | 602959 | Epileptic encephalopathy |
| *EFHC1* | NM_018100.3 | 100 | 608815 | Epilepsy, juvenile absence, susceptibility to, 1; Myoclonic epilepsy, juvenile, susceptibility to, 1 |
| *EPM2A* | NM_005670.3 | 90.5 | 607566 | Epilepsy, progressive myoclonic 2A (Lafora) |
| *EXOSC3* | NM_016042.2 | 99.38 | 606489 | Pontocerebellar hypoplasia, type 1B |

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| *FIG4* | NM_014845.5 | 99.97 | 609390 | Polymicrogyria, bilateral temporooccipital (also allelic to multiple other conditions) |
| *FLNA* | NM_001456.3 | 100 | 300017 | Heterotopia, periventricular; Heterotopia, periventricular, ED variant (also allelic to multiple other conditions) |
| *FOLR1*** | NM_016725.2 | 100 | 136430 | Neurodegeneration due to cerebral folate transport deficiency |
| *FOXG1* | NM_005249.4 | 100 | 164874 | Rett syndrome, congenital variant |
| *GABRA1* | NM_000806.5 | 95.98 | 137160 | Epileptic encephalopathy, early infantile, 19; Epilepsy, childhood absence, susceptibility to, 4; Epilepsy, childhood absence, susceptibility to, 5 |
| *GABRB3* | NM_000814.5 | 100 | 137192 | Epilepsy, childhood absence, susceptibility to, 5 |
| *GABRG2* | NM_000816.3 | 99.93 | 137164 | Epilepsy, generalized, with febrile seizures plus, type 3; Febrile seizures, familial, 8; Epilepsy, childhood absence, susceptibility to, 2 |
| *GAMT* | NM_000156.5 | 96.56 | 601240 | Cerebral creatine deficiency syndrome 2 |
| *GATM* | NM_001482.2 | 99.86 | 602360 | Cerebral creatine deficiency syndrome 3 |
| *GLI2* | NM_005270.4 | 100 | 165230 | Holoprosencephaly-9; Culler-Jones syndrome |
| *GNAO1* | NM_020988.2 | 100 | 139311 | Epileptic encephalopathy, early infantile, 17 |
| *GOSR2* | NM_004287.3 | 100 | 604027 | Epilepsy, progressive myoclonic 6 |
| *GPHN* | NM_020806.4 | 99.97 | 603930 | Molybdenum cofactor deficiency C |
| *GRIK2* | NM_021956.4 | 100 | 611092 | Mental retardation, autosomal recessive 6 |
| *GRIN1* | NM_007327.3 | 99.92 | 138249 | Mental retardation, autosomal dominant 8 |
| *GRIN2A* | NM_000833.3 | 100 | 138253 | Epilepsy, focal, with speech disorder and with or without mental retardation; |
| *GRIN2B* | NM_000834.3 | 99.87 | 138252 | Epileptic encephalopathy, early infantile, 27; Mental retardation, autosomal dominant 6 |
| *GRN* | NM_002087.2 | 100 | 138945 | Aphasia, primary progressive; Ceroid lipofuscinosis, neuronal, 11; Frontotemporal lobar degeneration with ubiquitin-positive inclusions |
| *HCN1* | NM_021072.2 | 99.99 | 602780 | Epileptic encephalopathy, early infantile, 24 |
| *HNRNPU* | NM_031844.2 | 99.97 | 602869 | Epileptic encephalopathy; Intellectual disabilities, CNS anomalies and seizures |
| *IER3IP1* | NM_016097.3 | 100 | 609382 | Microcephaly, epilepsy, and diabetes syndrome |
| *IQSEC2* | NM_001111125.2 | 96.97 | 300522 | Mental retardation, X-linked 1 |
| *JRK* | NM_003724.2 | 99.96 | 603210 | Childhood absence epilepsy |
| *KANSL1* | NM_001193466.1 | 99.98 | 612452 | Koolen-De Vries syndrome |
| *KCNA2* | NM_001204269.1 | 100 | 176262 | Epileptic encephalopathy |
| *KCNC1* | NM_001112741.1 | 100 | 176258 | Epilepsy, progressive myoclonic 7 |
| *KCNH5* | NM_139318.3 | 100 | 605716 | Epileptic encephalopathy |
| *KCNJ10* | NM_002241.4 | 100 | 602208 | Enlarged vestibular aqueduct, digenic; SESAME syndrome (includes seizures) |
| *KCNMA1* | NM_002247.3 | 100 | 600150 | Generalized epilepsy and paroxysmal dyskinesia |

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| *KCNQ2*** | NM_172107.2 | 100 | 613720 | Epileptic encephalopathy, early infantile, 7 |
| *KCNQ3*** | NM_004519.2 | 100 | 602235 | Seizures, benign neonatal, type 2 |
| *KCNT1*** | NM_020822.2 | 99.79 | 608167 | Epilepsy, nocturnal frontal lobe, 5; Epileptic encephalopathy, early infantile, 14 |
| *KCTD7* | NM_153033.4 | 100 | 611725 | Epilepsy, progressive myoclonic 3, with or without intracellular inclusions |
| *KPNA7* | NM_001145715.1 | 100 | 614107 | EIEE candidate |
| *KPTN* | NM_007059.2 | 99.95 | 615620 | Mental retardation, autosomal recessive 41 |
| *LGI1* | NM_005097.2 | 100 | 604619 | Epilepsy, familial temporal lobe, 1 |
| *LIAS* | NM_006859.2 | 97.08 | 607031 | Pyruvate dehydrogenase lipoic acid synthetase deficiency |
| *MAGI2* | NM_012301.3 | 100 | 606382 | Deletions including MAGI2 reported in patients with seizuers (PMID: 18565486) |
| *MBD5* | NM_018328.4 | 99.99 | 611472 | Mental retardation, autosomal dominant 1 |
| *MCPH1* | NM_024596.4 | 100 | 607117 | Microcephaly 1, primary, autosomal recessive |
| *MECP2*** | NM_004992.3 | 100 | 300005 | Encephalopathy, neonatal severe, Mental retardation, X-linked syndromic, Lubs type, Mental retardation, X-linked, syndromic 13, Rett syndrome<br>Rett syndrome, atypical Rett syndrome, preserved speech variant |
| *MEF2C*** | NM_002397.4 | 100 | 600662 | Chromosome 5q14.3 deletion syndrome;Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations; |
| *MFSD8* | NM_152778.2 | 100 | 611124 | Ceroid lipofuscinosis, neuronal, 7; |
| *MTOR* | NM_004958.3 | 99.94 | 601231 | Smith-Kingsmore syndrome |
| *NDE1* | NM_001143979.1 | 100 | 609449 | Microhydranencephaly; Lissencephaly 4 (with microcephaly); |
| *NEDD4L* | NM_001144967.2 | 100 | 606384 | Epileptic encephalopathy candidate (PMID: 23934111; limited evidance) |
| *NHLRC1* | NM_198586.2 | 99.96 | 608072 | Epilepsy, progressive myoclonic 2B (Lafora) |
| *NRXN1* | NM_001135659.1 | 98.75 | 600565 | Pitt-Hopkins-like syndrome 2; Schizophrenia, susceptibility to, 17 |
| *OPHN1* | NM_002547.2 | 99.71 | 300127 | Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance |
| *PAFAH1B1* | NM_000430.3 | 99.59 | 601545 | Lissencephaly 1;Subcortical laminar heterotopia |
| *PCDH19*** | NM_001105243.1 | 100 | 300460 | Epileptic encephalopathy, early infantile, 9 |
| *PHF6* | NM_032458.2 | 99.73 | 300414 | Borjeson-Forssman-Lehmann syndrome |
| *PIGA* | NM_002641.3 | 100 | 311770 | Multiple congenital anomalies-hypotonia-seizures syndrome 2;Paroxysmal nocturnal hemoglobinuria, somatic |
| *PIGN* | NM_176787.4 | 100 | 606097 | Multiple congenital anomalies-hypotonia-seizures syndrome 1 |
| *PIGO* | NM_032634.3 | 99.87 | 614749 | Hyperphosphatasia with mental retardation syndrome 2 |
| *PIGT* | NM_015937.5 | 100 | 610272 | Multiple congenital anomalies-hypotonia-seizures syndrome 3; ?Paroxysmal nocturnal hemoglobinuria 2 |
| *PLCB1* | NM_015192.2 | 100 | 607120 | Epileptic encephalopathy, early infantile, 12 |

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| *PNKP* | NM_007254.3 | 100 | 605610 | Epileptic encephalopathy, early infantile, 10 |
| *PNPO* | NM_018129.3 | 100 | 610090 | Pyridoxamine 5'-phosphate oxidase deficiency |
| *POLG*** | NM_002693.2 | 100 | 174763 | Mitochondrial DNA depletion syndrome 4A (Alpers type), Mitochondrial DNA depletion syndrome 4B (MNGIE type), Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), Progressive external ophthalmoplegia, autosomal dominant 1, Progressive external ophthalmoplegia, autosomal recessive 1 |
| *PPT1* | NM_000310.3 | 100 | 600722 | Ceroid lipofuscinosis, neuronal, 1 |
| *PRICKLE1* | NM_153026.2 | 100 | 608500 | Epilepsy, progressive myoclonic 1B |
| *PRICKLE2* | NM_198859.3 | 100 | 608501 | Epilepsy, progressive myoclonic 5 |
| *PRRT2* | NM_145239.2 | 100 | 614386 | Convulsions, familial infantile, with paroxysmal choreoathetosis; Episodic kinesigenic dyskinesia 1; Seizures, benign familial infantile, 2 |
| *PTCH1* | NM_000264.3 | 100 | 601309 | Basal cell carcinoma, somatic; Basal cell nevus syndrome; Holoprosencephaly-7 |
| *PURA* | NM_005859.4 | 99.95 | 600473 | Mental retardation, autosomal dominant 31 |
| *QARS* | NM_005051.2 | 100 | 603727 | Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy |
| *ROGDI* | NM_024589.2 | 100 | 614574 | Kohlschutter-Tonz syndrome; |
| *SCARB2* | NM_005506.3 | 99.77 | 602257 | Epilepsy, progressive myoclonic 4, with or without renal failure |
| *SCN1A*** | NM_001165963.1 | 99.91 | 182389 | Dravet syndrome, Epilepsy, generalized, with febrile seizures plus, type 2, Febrile seizures, familial, 3A, Migraine, familial hemiplegic, 3 |
| *SCN1B*** | NM_001037.4 | 100 | 600235 | Atrial fibrillation, familial, 13; Brugada syndrome 5; Cardiac conduction defect, nonspecific; Epilepsy, generalized, with febrile seizures plus, type |
| *SCN2A*** | NM_021007.2 | 99.8 | 182390 | Epileptic encephalopathy, early infantile, 11, Seizures, benign familial infantile, 3 |
| *SCN3A* | NM_006922.3 | 100 | 182391 | Pediatric partial epilepsy; Focal epilepsy |
| *SCN8A*** | NM_014191.3 | 100 | 600702 | ?Cognitive impairment with or without cerebellar ataxia, Epileptic encephalopathy, early infantile, 13, Seizures, benign familial infantile, 5 |
| *SCN9A* | NM_002977.3 | 99.67 | 603415 | Epilepsy, generalized, with febrile seizures plus, 7; Erythermalgia, primary; Febrile seizures, familial, 3B; Insensitivity to pain; Paroxysmal extreme pain disorder; Small fiber neuropathy; Dravet syndrome, modifier of |
| *SERPINI1* | NM_005025.4 | 100 | 602445 | Encephalopathy, familial, with neuroserpin inclusion bodies |
| *SHANK3* | NM_001080420 | 95.08 | 606230 | Phelan-McDermid syndrome; Schizophrenia susceptibility to 15 |
| *SHH* | NM_000193.2 | 99.99 | 600725 | Holoprosencephaly-3; Microphthalmia with coloboma 5; Schizencephaly; Single median maxillary central incisor |
| *SIX3* | NM_005413.3 | 100 | 603714 | Holoprosencephaly-2; Schizencephaly |
| *SLC12A5* | NM_020708.4 | 100 | 606726 | Epileptic encephalopathy, early infantile, 34 |
| *SLC13A5* | NM_001143838.1 | 100 | 608305 | Epileptic encephalopathy, early infantile, 25 |
| *SLC19A3* | NM_025243.3 | 99.98 | 606152 | Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2) |
| *SLC1A2* | NM_004171.3 | 100 | 600300 | Epileptic encephalopathy, early infantile, 41 |

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| *SLC1A3* | NM_004172.4 | 100 | 600111 | Episodic ataxia, type 6 |
| *SLC25A12* | NM_003705.4 | 100 | 603667 | Hypomyelination, global cerebral |
| *SLC25A22* | NM_024698.5 | 100 | 609302 | Epileptic encephalopathy, early infantile, 3 |
| *SLC2A1*** | NM_006516.2 | 100 | 138140 | Dystonia 9, GLUT1 deficiency syndrome 1, infantile onset, severe, GLUT1 deficiency syndrome 2, childhood onset, Stomatin-deficient cryohydrocytosis with neurologic defects, {Epilepsy, idiopathic generalized, susceptibility to, 12} |
| *SLC35A2* | NM_005660.2 | 100 | 314375 | Congenital disorder of glycosylation, type IIm |
| *SLC35A3* | NM_012243.2 | 99.68 | 615553 | Arthrogryposis, mental retardation, and seizures |
| *SLC6A8*** | NM_005629.3 | 99.94 | 300036 | Cerebral creatine deficiency syndrome 1 |
| *SLC9A6* | NM_001042537.1 | 99.87 | 300231 | Mental retardation, X-linked syndromic, Christianson type |
| *SPTAN1*** | NM_001130438.1 | 100 | 182810 | Epileptic encephalopathy, early infantile, 5 |
| *SRPX2* | NM_014467.2 | 99.9 | 300643 | Rolandic epilepsy, mental retardation, and speech dyspraxia |
| *ST3GAL3* | NM_006279.3 | 99.99 | 606494 | Epileptic encephalopathy, early infantile, 15; Mental retardation, autosomal recessive 12 |
| *ST3GAL5* | NM_003896.3 | 100 | 604402 | Amish infantile epilepsy syndrome |
| *STX1B* | NM_052874.3 | 97.5 | 601485 | Generalized epilepsy with febrile seizures plus, type 9 |
| *STXBP1* | NM_003165.3 | 100 | 602926 | Epileptic encephalopathy, early infantile, 4 |
| *SYN1* | NM_133499.2 | 100 | 313440 | Epilepsy, X-linked, with variable learning disabilities and behavior disorders |
| *SYNGAP1* | NM_006772.2 | 99.16 | 603384 | Mental retardation, autosomal dominant 5 |
| *SZT2* | NM_015284.3 | 100 | 615463 | Epileptic encephalopathy, early infantile, 18 |
| *TBC1D24* | NM_020705.2 | 100 | 613577 | Deafness , autosomal recessive 86; DOOR syndrome; Epileptic encephalopathy, early infantile, 16; Myoclonic epilepsy, infantile, familial |
| *TCF4* | NM_001083962.1 | 100 | 602272 | Pitt-Hopkins syndrome |
| *TNK2* | NM_001010938.1 | 99.83 | 606994 | Infantile epilepsy candidate (case report PMID: 23686771) |
| *TPP1* | NM_000391.3 | 100 | 607998 | Ceroid lipofuscinosis, neuronal, 2;Spinocerebellar ataxia, autosomal recessive 7 |
| *TSC1* | NM_000368.4 | 99.99 | 605284 | Focal cortical dysplasia, Taylor balloon cell type; Lymphangioleiomyomatosis; Tuberous sclerosis-1 |
| *TSC2* | NM_000548.3 | 100 | 191092 | Lymphangioleiomyomatosis, somatic; Tuberous sclerosis-2 |
| *TSEN2* | NM_025265.3 | 99.98 | 608753 | Pontocerebellar hypoplasia type 2B |
| *TSEN54* | NM_207346.2 | 97.72 | 608755 | Pontocerebellar hypoplasia type 2A; Pontocerebellar hypoplasia type 4 |
| *TUBA1A* | NM_006009.3 | 100 | 602529 | Lissencephaly 3 |
| *TUBB2A* | NM_001069.2 | 99.96 | 615101 | Cortical dysplasia, complex, with other brain malformations 5 |
| *UBE3A* | NM_130838.1 | 99.91 | 601623 | Angelman syndrome |
| *WDR45* | NM_007075.3 | 99.41 | 300526 | Neurodegeneration with brain iron accumulation 5 |

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| *WDR62* | NM_001083961.1 | 100 | 613583 | Microcephaly 2, primary, autosomal recessive, with or without cortical malformations |
| *WWOX* | NM_016373.3 | 100 | 605131 | Epileptic encephalopathy, early infantile, 28 |
| *ZEB2* | NM_014795.3 | 100 | 605802 | Mowat-Wilson syndrome |

**The column 'Covered 10X (%)' indicates the percent of bases in the coding regions of the gene that were covered at the minimum of 10X in all the samples tested during assay validation.*
***For these genes, regions not covered at minimum 10X are additionally tested by Sanger sequencing. This includes genes associated with Early Infantile Epileptic Encephalopathy, as well as genes where mutations may directly affect treatment.*