EPILEPSY GENE PANEL DG 3.4.0 (378 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
AARS1	100,0%	100,0%	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287 ?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661 Trichothiodystrophy 8, nonphotosensitive, 619691
ABAT	100,0%	100,0%	GABA-transaminase deficiency, 613163
ABCC8	100,0%	100,0%	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 Diabetes mellitus, transient neonatal 2, 610374 Diabetes mellitus, noninsulin-dependent, 125853 Hypoglycemia of infancy, leucine-sensitive, 240800 Hyperinsulinemic hypoglycemia, familial, 1, 256450
ACTB	100,0%	100,0%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACTL6B	100,0%	100,0%	Developmental and epileptic encephalopathy 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACY1	100,0%	100,0%	Aminoacylase 1 deficiency, 609924
ADSL	100,0%	100,0%	Adenylosuccinase deficiency, 103050
AGA	100,0%	100,0%	Aspartylglucosaminuria, 208400
ALDH5A1	100,0%	100,0%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH7A1	100,0%	100,0%	Epilepsy, pyridoxine-dependent, 266100
ALG1	100,0%	100,0%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	96,8%	96,8%	Congenital disorder of glycosylation, type Ip, 613661
ALG13	100,0%	99,9%	Developmental and epileptic encephalopathy 36, 300884
ALG3	100,0%	100,0%	Congenital disorder of glycosylation, type Id, 601110
ALG6	100,0%	100,0%	Congenital disorder of glycosylation, type Ic, 603147
AMACR	100,0%	100,0%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMPD2	100,0%	100,0%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	100,0%	100,0%	Glycine encephalopathy, 605899

ANKRD11	100,0%	100,0%	KBG syndrome, 148050
AP1G1	100,0%	100,0%	Usmani-Riazuddin syndrome, autosomal recessive, 619548 Usmani-Riazuddin syndrome, autosomal dominant, 619467
AP3B2	100,0%	99,7%	Developmental and epileptic encephalopathy 48, 617276
ARHGEF9	97,2%	97,2%	Developmental and epileptic encephalopathy 8, 300607
ARID1B	98,6%	98,3%	Coffin-Siris syndrome 1, 135900
ARX	99,0%	96,8%	Proud syndrome, 300004 Hydranencephaly with abnormal genitalia, 300215 Partington syndrome, 309510 Developmental and epileptic encephalopathy 1, 308350 Lissencephaly, X-linked 2, 300215 Intellectual developmental disorder, X-linked 29, 300419
ASAH1	100,0%	100,0%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 Farber lipogranulomatosis, 228000
ASL	100,0%	100,0%	Argininosuccinic aciduria, 207900
ASNS	100,0%	100,0%	Asparagine synthetase deficiency, 615574
ASXL3	100,0%	100,0%	Bainbridge-Ropers syndrome, 615485
ATP1A1	100,0%	100,0%	Hypomagnesemia, seizures, and mental retardation 2, 618314 Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP1A2	100,0%	100,0%	Developmental and epileptic encephalopathy 98, 619605 Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	100,0%	100,0%	Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235 CAPOS syndrome, 601338 Developmental and epileptic encephalopathy 99, 619606
ATP6AP2	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911 Congenital disorder of glycosylation, type IIr, 301045
ATP7A	100,0%	100,0%	Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489 Menkes disease, 309400
ATRX	100,0%	100,0%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Intellectual disability-hypotonic facies syndrome, X-linked, 309580
AUTS2	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 26, 615834

BOLA3	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BRAT1	100,0%	100,0%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BTD	83,1%	83,1%	Biotinidase deficiency, 253260
CACNA1A	100,0%	100,0%	Spinocerebellar ataxia 6, 183086 Episodic ataxia, type 2, 108500 Developmental and epileptic encephalopathy 42, 617106 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Migraine, familial hemiplegic, 1, 141500
CACNA1E	100,0%	100,0%	Developmental and epileptic encephalopathy 69, 618285
CACNA2D2	100,0%	100,0%	Cerebellar atrophy with seizures and variable developmental delay, 618501
CACNB4	100,0%	100,0%	Episodic ataxia, type 5, 613855
CAD	100,0%	100,0%	Developmental and epileptic encephalopathy 50, 616457
CASK	100,0%	100,0%	Intellectual developmental disorder, with or without nystagmus, 300422 Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422
CASQ2	100,0%	100,0%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CDKL5	92,3%	92,2%	Developmental and epileptic encephalopathy 2, 300672
CERT1	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 34, 616351
CHD2	100,0%	100,0%	Developmental and epileptic encephalopathy 94, 615369
CHD5	100,0%	100,0%	No OMIM Disease ID
CHRNA2	100,0%	100,0%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	100,0%	100,0%	Epilepsy, nocturnal frontal lobe, 1, 600513
CHRNB2	100,0%	100,0%	Epilepsy, nocturnal frontal lobe, 3, 605375
CIC	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 45, 617600
CILK1	100,0%	100,0%	Endocrine-cerebroosteodysplasia, 612651
CLCN4	100,0%	100,0%	Raynaud-Claes syndrome, 300114
CLDN16	100,0%	100,0%	Hypomagnesemia 3, renal, 248250
CLDN19	100,0%	100,0%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	92,7%	92,5%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	71,7%	71,6%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 Ceroid lipofuscinosis, neuronal, 8, 600143
CNNM2	100,0%	100,0%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418

CNTN2	100,0%	100,0%	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP2	100,0%	100,0%	Pitt-Hopkins like syndrome 1, 610042
COA8	93,5%	93,5%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COL4A1	100,0%	100,0%	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 Brain small vessel disease with or without ocular anomalies, 175780
COLGALT1	100,0%	100,0%	Brain small vessel disease 3, 618360
COQ2	97,2%	97,2%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 4, 612016
CPA6	100,0%	100,0%	Febrile seizures, familial, 11, 614418 Epilepsy, familial temporal lobe, 5, 614417
CPS1	100,0%	100,0%	Carbamoylphosphate synthetase I deficiency, 237300
CPT2	100,0%	100,0%	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110
CSNK2B	100,0%	100,0%	Poirier-Bienvenu neurodevelopmental syndrome, 618732
CSTB	100,0%	100,0%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362
CUL4B	100,0%	99,9%	Intellectual developmental disorder, X-linked, syndromic, Cabezas type, 300354
CUX2	100,0%	100,0%	Developmental and epileptic encephalopathy 67, 618141
D2HGDH	100,0%	100,0%	D-2-hydroxyglutaric aciduria, 600721
DARS1	100,0%	100,0%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	100,0%	100,0%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DCX	100,0%	100,0%	Subcortical laminal heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067
DDX3X	99,2%	97,6%	Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958
DENND5A	100,0%	100,0%	Developmental and epileptic encephalopathy 49, 617281
DEPDC5	100,0%	100,0%	Epilepsy, familial focal, with variable foci 1, 604364
DHDDS	95,2%	95,2%	Developmental delay and seizures with or without movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861 Retinitis pigmentosa 59, 613861
DIAPH1	100,0%	100,0%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DLAT	100,0%	100,0%	Pyruvate dehydrogenase E2 deficiency, 245348

DNAJC5	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350
DNM1	97,7%	97,4%	Developmental and epileptic encephalopathy 31, 616346
DNM1L	100,0%	100,0%	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DOCK7	100,0%	100,0%	Developmental and epileptic encephalopathy 23, 615859
DPAGT1	100,0%	100,0%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 Congenital disorder of glycosylation, type Ij, 608093
DPM1	99,8%	97,8%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100,0%	100,0%	Congenital disorder of glycosylation, type Iu, 615042
DPYD	100,0%	100,0%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	100,0%	100,0%	Dihydropyrimidinuria, 222748
DTYMK	100,0%	100,0%	No OMIM Disease ID
DYNC1H1	100,0%	100,0%	Charcot-Marie-Tooth disease, axonal, type 2O, 614228 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Intellectual developmental disorder, autosomal dominant 13, 614563
DYRK1A	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 7, 614104
EBP	100,0%	100,0%	MEND syndrome, 300960 Chondrodysplasia punctata, X-linked dominant, 302960
EEF1A2	100,0%	100,0%	Developmental and epileptic encephalopathy 33, 616409 Intellectual developmental disorder, autosomal dominant 38, 616393
EFHC1	98,0%	98,0%	No OMIM Disease ID
EGF	100,0%	100,0%	?Hypomagnesemia 4, renal, 611718
EHMT1	99,9%	99,8%	Kleefstra syndrome 1, 610253
EIF2B1	100,0%	100,0%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	100,0%	100,0%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	100,0%	100,0%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	100,0%	100,0%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
EIF2B5	100,0%	100,0%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
EPM2A	100,0%	100,0%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
ETHE1	100,0%	100,0%	Ethylmalonic encephalopathy, 602473
EXOC7	100,0%	100,0%	Neurodevelopmental disorder with seizures and brain atrophy, 619072
EXOSC3	100,0%	100,0%	Pontocerebellar hypoplasia, type 1B, 614678
FA2H	100,0%	100,0%	Spastic paraplegia 35, autosomal recessive, 612319

FARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FGD1	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF12	100,0%	100,0%	Developmental and epileptic encephalopathy 47, 617166
FLNA	100,0%	100,0%	Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Melnick-Needles syndrome, 309350 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type I, 311300 Heterotopia, periventricular, 1, 300049 Frontometaphyseal dysplasia 1, 305620
FOLR1	100,0%	100,0%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	100,0%	100,0%	Rett syndrome, congenital variant, 613454
FOXRED1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRMPD4	98,3%	98,3%	Intellectual developmental disorder, X-linked 104, 300983
FRRS1L	100,0%	100,0%	Developmental and epileptic encephalopathy 37, 616981
FXYD2	100,0%	100,0%	Hypomagnesemia 2, renal, 154020
GABRA1	100,0%	100,0%	Developmental and epileptic encephalopathy 19, 615744
GABRB3	100,0%	100,0%	Developmental and epileptic encephalopathy 43, 617113
GABRG2	93,0%	93,0%	Developmental and epileptic encephalopathy 74, 618396 Febrile seizures, familial, 8, 607681 Generalized epilepsy with febrile seizures plus, type 3, 607681
GAMT	100,0%	100,0%	Cerebral creatine deficiency syndrome 2, 612736
GCK	97,0%	93,1%	MODY, type II, 125851 Diabetes mellitus, permanent neonatal 1, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 Diabetes mellitus, noninsulin-dependent, late onset, 125853
GCSH	100,0%	100,0%	?Glycine encephalopathy, 605899
GLDC	100,0%	100,0%	Glycine encephalopathy, 605899
GLRA1	100,0%	100,0%	Hyperekplexia 1, 149400
GLRB	100,0%	100,0%	Hyperekplexia 2, 614619
GLUD1	100,0%	100,0%	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	100,0%	100,0%	Developmental and epileptic encephalopathy 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493

GOSR2	100,0%	100,0%	Epilepsy, progressive myoclonic 6, 614018
GPC3	100,0%	99,9%	Wilms tumor, somatic, 194070 Simpson-Golabi-Behmel syndrome, type 1, 312870
GPHN	100,0%	100,0%	Molybdenum cofactor deficiency C, 615501
GRIA3	99,9%	99,7%	Intellectual developmental disorder, X-linked, syndromic, Wu type, 300699
GRIN1	100,0%	100,0%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 Developmental and epileptic encephalopathy 101, 619814 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GRIN2A	100,0%	100,0%	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570
GRIN2B	100,0%	100,0%	Developmental and epileptic encephalopathy 27, 616139 Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970
GRIN2D	99,9%	99,3%	Developmental and epileptic encephalopathy 46, 617162
GRN	100,0%	100,0%	Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706
HACE1	100,0%	100,0%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	100,0%	100,0%	Hyperinsulinemic hypoglycemia, familial, 4, 609975 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HCFC1	100,0%	100,0%	Methylmalonic aciduria and homocysteinemia, cblX type, 309541
HCN1	98,5%	98,5%	Developmental and epileptic encephalopathy 24, 615871 Generalized epilepsy with febrile seizures plus, type 10, 618482
HECW2	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HLCS	100,0%	100,0%	Holocarboxylase synthetase deficiency, 253270
HNRNPU	100,0%	100,0%	Developmental and epileptic encephalopathy 54, 617391
HSD17B10	100,0%	100,0%	HSD10 mitochondrial disease, 300438
HSD17B4	96,6%	96,6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
IDH2	100,0%	100,0%	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	100,0%	100,0%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	100,0%	100,0%	Immunodeficiency 95, 619773 Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IQSEC2	99,8%	99,1%	Intellectual developmental disorder, X-linked 1, 309530
IRF2BPL	100,0%	100,0%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
ITPA	100,0%	100,0%	Developmental and epileptic encephalopathy 35, 616647
JAM3	100,0%	100,0%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANSL1	100,0%	100,0%	Koolen-De Vries syndrome, 610443

KATNB1	100,0%	100,0%	Lissencephaly 6, with microcephaly, 616212
KCNA1	100,0%	100,0%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	100,0%	100,0%	Developmental and epileptic encephalopathy 32, 616366
KCNB1	100,0%	100,0%	Developmental and epileptic encephalopathy 26, 616056
KCNC1	100,0%	100,0%	Epilepsy, progressive myoclonic 7, 616187
KCNH1	98,7%	98,7%	Zimmermann-Laband syndrome 1, 135500 Temple-Baraitser syndrome, 611816
KCNJ10	100,0%	100,0%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	100,0%	100,0%	Diabetes, permanent neonatal 2, with or without neurologic features, 618856 Maturity-onset diabetes of the young, type 13, 616329 Diabetes mellitus, transient neonatal 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNMA1	100,0%	100,0%	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 Cerebellar atrophy, developmental delay, and seizures, 617643 Liang-Wang syndrome, 618729
KCNQ2	100,0%	100,0%	Developmental and epileptic encephalopathy 7, 613720 Seizures, benign neonatal, 1, 121200 Myokymia, 121200
KCNQ3	100,0%	100,0%	Seizures, benign neonatal, 2, 121201
KCNT1	99,9%	99,6%	Developmental and epileptic encephalopathy 14, 614959 Epilepsy nocturnal frontal lobe, 5, 615005
KCNT2	100,0%	100,0%	Developmental and epileptic encephalopathy 57, 617771
KCTD7	100,0%	100,0%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534
KDM6B	100,0%	100,0%	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505
KIF5A	100,0%	100,0%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187
KMT5B	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 51, 617788
KPTN	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 41, 615637
LAMB1	100,0%	100,0%	Lissencephaly 5, 615191
LGI1	100,0%	100,0%	Epilepsy, familial temporal lobe, 1, 600512
LIAS	100,0%	100,0%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPT2	100,0%	100,0%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
MAPK8IP3	100,0%	100,0%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MBD5	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 1, 156200

MECP2	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic 13, 300055 Rett syndrome, atypical, 312750 Encephalopathy, neonatal severe, 300673 Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260 Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750
MED12	100,0%	100,0%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Hardikar syndrome, 301068 Opitz-Kaveggia syndrome, 305450
MEF2C	100,0%	100,0%	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443
MFF	100,0%	100,0%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD8	100,0%	100,0%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MLC1	100,0%	100,0%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MOCS1	100,0%	100,0%	Molybdenum cofactor deficiency A, 252150
MOCS2	100,0%	100,0%	Molybdenum cofactor deficiency B, 252160
MPDU1	100,0%	100,0%	Congenital disorder of glycosylation, type If, 609180
MPDZ	100,0%	100,0%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MTFMT	100,0%	100,0%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFR	100,0%	100,0%	Homocystinuria due to MTHFR deficiency, 236250
MTOR	100,0%	100,0%	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638
MTRR	100,0%	100,0%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
NACC1	100,0%	100,0%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NANS	100,0%	100,0%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 24, 616239 ?Deafness, autosomal recessive 94, 618434
NBEA	100,0%	100,0%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NCDN	100,0%	100,0%	Neurodevelopmental disorder with infantile epileptic spasms, 619373
NDUFA1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA11	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFAF1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 15, 618237

NDUFAF5	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFB3	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB9	98,7%	98,7%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFS1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	95,3%	91,3%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFV1	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NECAP1	100,0%	100,0%	Developmental and epileptic encephalopathy 21, 615833
NEDD4L	100,0%	100,0%	Periventricular nodular heterotopia 7, 617201
NEU1	100,0%	100,0%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEXMIF	100,0%	100,0%	Intellectual developmental disorder, X-linked 98, 300912
NGLY1	100,0%	100,0%	Congenital disorder of deglycosylation 1, 615273
NHLRC1	100,0%	100,0%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NPRL2	100,0%	100,0%	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	100,0%	100,0%	Epilepsy, familial focal, with variable foci 3, 617118
NR2F1	100,0%	99,8%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRXN1	100,0%	100,0%	Pitt-Hopkins-like syndrome 2, 614325
NUBPL	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUS1	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
OCLN	100,0%	100,0%	Pseudo-TORCH syndrome 1, 251290
OFD1	100,0%	100,0%	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
OPHN1	100,0%	99,5%	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486
PACS1	100,0%	100,0%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	100,0%	100,0%	Developmental and epileptic encephalopathy 66, 618067
PAFAH1B1	100,0%	100,0%	Subcortical laminar heterotopia, 607432 Lissencephaly 1, 607432
PAK3	100,0%	100,0%	Intellectual developmental disorder, X-linked 30, 300558
PC	100,0%	100,0%	Pyruvate carboxylase deficiency, 266150
PCDH19	100,0%	100,0%	Developmental and epileptic encephalopathy 9, 300088

PDHA1	100,0%	100,0%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	100,0%	100,0%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	100,0%	100,0%	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	100,0%	100,0%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDX1	100,0%	100,0%	Pancreatic agenesis 1, 260370 MODY, type IV, 606392
PET100	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	100,0%	100,0%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100,0%	100,0%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX12	100,0%	100,0%	Peroxisome biogenesis disorder 3B, 266510 Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	100,0%	100,0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	100,0%	100,0%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	100,0%	100,0%	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	100,0%	100,0%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX26	100,0%	100,0%	Peroxisome biogenesis disorder 7B, 614873 Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	100,0%	100,0%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	100,0%	100,0%	Peroxisome biogenesis disorder 2B, 202370 Peroxisome biogenesis disorder 2A (Zellweger), 214110 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100,0%	100,0%	Peroxisome biogenesis disorder 4B, 614863 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Heimler syndrome 2, 616617
PGAP3	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PHF6	100,0%	100,0%	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	100,0%	100,0%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	100,0%	100,0%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072

PIGN	98,8%	98,8%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	100,0%	100,0%	Developmental and epileptic encephalopathy 55, 617599
PIGT	100,0%	100,0%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PLA2G6	92,3%	92,3%	Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600
PLCB1	100,0%	100,0%	Developmental and epileptic encephalopathy 12, 613722
PLP1	100,0%	100,0%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	100,0%	100,0%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMM2	100,0%	100,0%	Congenital disorder of glycosylation, type Ia, 212065
PNKP	100,0%	100,0%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPO	100,0%	100,0%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLG	100,0%	100,0%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
PPP2R1A	93,6%	93,6%	Intellectual developmental disorder, autosomal dominant 36, 616362
PPP2R5D	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 35, 616355
PPT1	82,5%	82,5%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	100,0%	100,0%	Renpenning syndrome, 309500
PRF1	100,0%	100,0%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PRICKLE1	100,0%	100,0%	Epilepsy, progressive myoclonic 1B, 612437
PRRT2	100,0%	100,0%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Seizures, benign familial infantile, 2, 605751 Episodic kinesigenic dyskinesia 1, 128200
PSAP	100,0%	100,0%	Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539
PTRH2	100,0%	100,0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263

PTS	100,0%	100,0%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUM1	100,0%	100,0%	Spinocerebellar ataxia 47, 617931
PURA	100,0%	100,0%	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158
PYCR2	100,0%	100,0%	Leukodystrophy, hypomyelinating, 10, 616420
QARS1	100,0%	100,0%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	100,0%	100,0%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB39B	100,0%	100,0%	Intellectual developmental disorder, X-linked 72, 300271 Waisman syndrome, 311510
RARS2	100,0%	100,0%	Pontocerebellar hypoplasia, type 6, 611523
RNASEH2A	100,0%	100,0%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91,0%	91,0%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100,0%	100,0%	Aicardi-Goutieres syndrome 3, 610329
ROGDI	100,0%	100,0%	Kohlschutter-Tonz syndrome, 226750
RPS6KA3	100,0%	99,8%	Intellectual developmental disorder, X-linked 19, 300844 Coffin-Lowry syndrome, 303600
RRM2B	100,0%	100,0%	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
SAMHD1	100,0%	100,0%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SCARB2	100,0%	100,0%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN1A	100,0%	100,0%	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317 Migraine, familial hemiplegic, 3, 609634 Dravet syndrome, 607208 Febrile seizures, familial, 3A, 604403 Generalized epilepsy with febrile seizures plus, type 2, 604403
SCN1B	100,0%	100,0%	Generalized epilepsy with febrile seizures plus, type 1, 604233 Developmental and epileptic encephalopathy 52, 617350 Cardiac conduction defect, nonspecific, 612838 Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838
SCN2A	100,0%	100,0%	Seizures, benign familial infantile, 3, 607745 Developmental and epileptic encephalopathy 11, 613721 Episodic ataxia, type 9, 618924
SCN3A	100,0%	100,0%	Epilepsy, familial focal, with variable foci 4, 617935 Developmental and epileptic encephalopathy 62, 617938

SCN8A	100,0%	100,0%	?Myoclonus, familial, 2, 618364 Seizures, benign familial infantile, 5, 617080 Cognitive impairment with or without cerebellar ataxia, 614306 Developmental and epileptic encephalopathy 13, 614558
SEMA6B	100,0%	100,0%	Epilepsy, progressive myoclonic, 11, 618876
SEPSECS	100,0%	100,0%	Pontocerebellar hypoplasia type 2D, 613811
SERPINI1	100,0%	100,0%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SHANK3	98,3%	97,3%	Phelan-McDermid syndrome, 606232
SIK1	100,0%	100,0%	Developmental and epileptic encephalopathy 30, 616341
SLC12A5	97,4%	97,4%	Developmental and epileptic encephalopathy 34, 616645
SLC13A5	100,0%	100,0%	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905
SLC16A1	100,0%	100,0%	Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340 Monocarboxylate transporter 1 deficiency, 616095
SLC19A3	98,7%	98,7%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A2	100,0%	100,0%	Developmental and epileptic encephalopathy 41, 617105
SLC25A1	100,0%	100,0%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A15	100,0%	100,0%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	100,0%	100,0%	Developmental and epileptic encephalopathy 3, 609304
SLC2A1	100,0%	100,0%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC35A2	100,0%	100,0%	Congenital disorder of glycosylation, type IIm, 300896
SLC6A1	100,0%	100,0%	Myoclonic-atonic epilepsy, 616421
SLC6A8	100,0%	100,0%	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	100,0%	99,7%	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243
SMARCA2	98,4%	98,2%	Nicolaides-Baraitser syndrome, 601358 Blepharophimosis-impaired intellectual development syndrome, 619293
SMC1A	100,0%	100,0%	Cornelia de Lange syndrome 2, 300590 Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMPD4	100,0%	100,0%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMS	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNAP25	100,0%	100,0%	?Myasthenic syndrome, congenital, 18, 616330
SPATA5	100,0%	100,0%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
SPTAN1	100,0%	100,0%	Developmental and epileptic encephalopathy 5, 613477

ST3GAL3	95,8%	95,2%	Developmental and epileptic encephalopathy 15, 615006 Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	98,7%	98,7%	Salt and pepper developmental regression syndrome, 609056
STRADA	100,0%	100,0%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STX1B	100,0%	100,0%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	100,0%	100,0%	Developmental and epileptic encephalopathy 4, 612164
SUOX	100,0%	100,0%	Sulfite oxidase deficiency, 272300
SYN1	100,0%	100,0%	Intellectual developmental disorder, X-linked 50, 300115 Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 5, 612621
SYNJ1	100,0%	100,0%	Parkinson disease 20, early-onset, 615530 Developmental and epileptic encephalopathy 53, 617389
SYP	100,0%	100,0%	Intellectual developmental disorder, X-linked 96, 300802
SZT2	100,0%	100,0%	Developmental and epileptic encephalopathy 18, 615476
TANGO2	100,0%	100,0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TBC1D23	100,0%	100,0%	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	100,0%	100,0%	Deafness, autosomal recessive 86, 614617 Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105 Myoclonic epilepsy, infantile, familial, 605021 Deafness, autosomal dominant 65, 616044 Developmental and epileptic encephalopathy 16, 615338 DOORS syndrome, 220500
TBCD	100,0%	100,0%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	100,0%	100,0%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TCF4	100,0%	100,0%	Pitt-Hopkins syndrome, 610954 Corneal dystrophy, Fuchs endothelial, 3, 613267
TDP2	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TOE1	100,0%	100,0%	Pontocerebellar hypoplasia, type 7, 614969
TPP1	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TREX1	100,0%	100,0%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRPM3	100,0%	100,0%	No OMIM Disease ID
TRPM6	100,0%	100,0%	Hypomagnesemia 1, intestinal, 602014

TSC1	100,0%	100,0%	Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690
TSC2	100,0%	100,0%	Lymphangioleiomyomatosis, somatic, 606690 ?Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-2, 613254
TSEN15	100,0%	100,0%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	100,0%	100,0%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	100,0%	100,0%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TUBA1A	100,0%	100,0%	Lissencephaly 3, 611603
TUBB2A	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB4A	99,5%	97,4%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 4, 615412
UBA5	100,0%	100,0%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Developmental and epileptic encephalopathy 44, 617132
UBE3A	100,0%	100,0%	Angelman syndrome, 105830
UBTF	100,0%	100,0%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UGP2	96,6%	96,3%	Developmental and epileptic encephalopathy 83, 618744
VPS11	100,0%	100,0%	?Dystonia 32, 619637 Leukodystrophy, hypomyelinating, 12, 616683
VPS53	100,0%	99,8%	Pontocerebellar hypoplasia, type 2E, 615851
WDR26	97,0%	94,3%	Skraban-Deardorff syndrome, 617616
WDR45	100,0%	100,0%	Neurodegeneration with brain iron accumulation 5, 300894
WFS1	100,0%	100,0%	Deafness, autosomal dominant 6/14/38, 600965 ?Cataract 41, 116400 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WWOX	100,0%	100,0%	Esophageal squamous cell carcinoma, somatic, 133239 Developmental and epileptic encephalopathy 28, 616211 Spinocerebellar ataxia, autosomal recessive 12, 614322
XK	100,0%	100,0%	McLeod syndrome with or without chronic granulomatous disease, 300842
YWHAG	100,0%	100,0%	Developmental and epileptic encephalopathy 56, 617665
ZEB2	97,4%	97,4%	Mowat-Wilson syndrome, 235730

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
TWIST is the chemistry used for WES analysis.
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.
Genes with coverage denoting NC are non-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors