EPILEPSY GENE PANEL DG 3.1.0 (367 genes)

Releasedate: 23-03-2021

Gene	Agilent V5 covered >10x	Agilent V5 covered >20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
AARS1	100	99,9	100	100	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
ABAT	100	99,4	100	100	GABA-transaminase deficiency, 613163
ABCC8	100	99,8	100	100	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ACTB	99,7	96,1	100	100	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTL6B	100	99,8	100	100	Developmental and epileptic encephalopathy 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACY1	100	98,8	100	100	Aminoacylase 1 deficiency, 609924
ADSL	99,2	98,7	100	100	Adenylosuccinase deficiency, 103050
AGA	100	100	100	100	Aspartylglucosaminuria, 208400
ALDH5A1	91	81,5	100	100	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH7A1	94,4	88,8	100	100	Epilepsy, pyridoxine-dependent, 266100
ALG1	53	45,8	100	100	Congenital disorder of glycosylation, type Ik, 608540
ALG11	96,8	96,8	96,8	96,8	Congenital disorder of glycosylation, type Ip, 613661
ALG13	98,4	92,6	100	99,6	Developmental and epileptic encephalopathy 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884
ALG3	100	99,7	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	98,6	94,8	100	100	Congenital disorder of glycosylation, type Ic, 603147
AMACR	100	100	100	100	Bile acid synthesis defect, congenital, 4, 214950 Alpha-methylacyl-CoA racemase deficiency, 614307
AMPD2	99,8	98,9	100	100	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	100	100	100	100	Glycine encephalopathy, 605899
ANKRD11	96,1	93,5	100	100	KBG syndrome, 148050

AP3B2	93,3	89,5	99,8	98,6	Developmental and epileptic encephalopathy 48, 617276
ARHGEF9	76,5	74,1	97,2	97,1	Developmental and epileptic encephalopathy 8, 300607
ARID1B	96,2	95,2	97,9	96,7	Coffin-Siris syndrome 1, 135900
ARX	81	64	91,5	85,7	Lissencephaly, X-linked 2, 300215 Proud syndrome, 300004 Partington syndrome, 309510 Developmental and epileptic encephalopathy 1, 308350 Mental retardation, X-linked 29 and others, 300419 Hydranencephaly with abnormal genitalia, 300215
ASAH1	99,7	98,6	100	100	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	100	99,6	100	100	Argininosuccinic aciduria, 207900
ASNS	99,4	95,2	100	100	Asparagine synthetase deficiency, 615574
ASXL3	99,9	99,7	100	100	Bainbridge-Ropers syndrome, 615485
ATP1A2	100	100	100	100	Migraine, familial hemiplegic, 2, 602481 Migraine, familial basilar, 602481 Alternating hemiplegia of childhood 1, 104290
ATP1A3	100	99,9	100	100	CAPOS syndrome, 601338 Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235
ATP6AP2	94,1	76,6	100	100	Congenital disorder of glycosylation, type IIr, 301045 Mental retardation, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911
ATP7A	99	96,9	100	100	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
ATRX	99,4	96,3	100	100	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUTS2	98,2	95,8	100	100	Mental retardation, autosomal dominant 26, 615834
BOLA3	99,4	90,2	100	100	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BRAT1	99,7	98,2	100	100	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056
BTD	83,1	83	83,1	83,1	Biotinidase deficiency, 253260
CACNA1A	93,2	90	100	99,9	Spinocerebellar ataxia 6, 183086 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500

					Developemental and epileptic encephalopathy 42, 617106 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500
CACNA1E	100	99,9	100	100	Developmental and epileptic encephalopathy 69, 618285
CACNA2D2	94	93,2	99,2	97,6	Cerebellar atrophy with seizures and variable developmental delay, 618501
CACNB4	95,5	94,3	100	100	{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 Episodic ataxia, type 5, 613855
CAD	100	99,2	100	100	Developmental and epileptic encephalopathy 50, 616457
CASK	97,3	94,2	100	100	Mental retardation, with or without nystagmus, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422
CASQ2	100	100	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CDKL5	91,7	90,2	92,3	91,7	Developmental and epileptic encephalopathy 2, 300672
CERT1	90,2	87,3	100	100	Mental retardation, autosomal dominant 34, 616351
CHD2	99,4	99,2	100	100	Epileptic encephalopathy, childhood-onset, 615369
CHRNA2	100	100	100	100	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	98,3	96,2	100	100	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNB2	99,3	96	100	100	Epilepsy, nocturnal frontal lobe, 3, 605375
CIC	63,3	63,3	100	99,9	Mental retardation, autosomal dominant 45, 617600
CILK1	99,9	98,7	100	100	Endocrine-cerebroosteodysplasia, 612651 {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924
CLCN4	99,9	98,9	100	100	Raynaud-Claes syndrome, 300114
CLDN16	100	100	100	100	Hypomagnesemia 3, renal, 248250
CLDN19	98,5	93,1	100	100	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	92,5	91,8	92,5	92,5	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	69,3	66,3	72,1	71,6	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	99,9	97,1	100	100	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	83,5	83,5	100	100	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CNNM2	100	100	100	100	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNTN2	92,7	92,7	100	100	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP2	100	99,8	100	100	{Autism susceptibility 15}, 612100 Pitt-Hopkins like syndrome 1, 610042 Cortical dysplasia-focal epilepsy syndrome, 610042

COA8	81,9	80,7	93,5	93,4	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COL4A1	98,7	97,4	100	100	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 175780 {Hemorrhage, intracerebral, susceptibility to}, 614519 ?Retinal arteries, tortuosity of, 180000 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564
COLGALT1	93,3	89	98,6	97	Brain small vessel disease 3, 618360
COQ2	98	95,3	97,2	97,2	{Multiple system atrophy, susceptibility to}, 146500 Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	90,9	89,3	100	100	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	100	99,5	100	100	Coenzyme Q10 deficiency, primary, 4, 612016
CPA6	99,6	97,5	100	100	Febrile seizures, familial, 11, 614418 Epilepsy, familial temporal lobe, 5, 614417
CPS1	100	99,9	100	100	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371
CPT2	98,2	97,8	100	100	CPT II deficiency, myopathic, stress-induced, 255110 CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CSNK2B	100	100	100	100	Poirier-Bienvenu neurodevelopmental syndrome, 618732
CSTB	99,6	89,8	100	100	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	98,4	95	100	100	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	84	79,3	100	99,9	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CUL4B	98	90,8	99,9	99,2	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUX2	99,9	99,1	100	100	Developmental and epileptic encephalopathy 67, 618141
D2HGDH	99,2	97,2	100	100	D-2-hydroxyglutaric aciduria, 600721
DARS1	100	99,3	100	100	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	94,9	94,3	100	100	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DCX	100	99,9	100	100	Subcortical laminal heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067
DDX3X	81,2	78,9	98	96,1	Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958
DENND5A	100	99,4	100	100	Developmental and epileptic encephalopathy 49, 617281
DEPDC5	100	99,8	100	100	Epilepsy, familial focal, with variable foci 1, 604364
DHDDS	99	95	95,2	95,2	Retinitis pigmentosa 59, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861
DLAT	100	99,7	100	100	Pyruvate dehydrogenase E2 deficiency, 245348

DNAJC5	100	100	100	100	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNM1	92,6	89,1	97,4	97,4	Developmental and epileptic encephalopathy 31, 616346
DNM1L	99,9	98,5	100	100	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DOCK7	99,8	98,2	100	99,9	Developmental and epileptic encephalopathy 23, 615859
DPAGT1	100	100	100	100	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	98,2	91,3	99,7	97,1	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100	98,7	100	100	Congenital disorder of glycosylation, type Iu, 615042
DPYD	99,7	97,7	100	100	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	100	99,9	100	100	Dihydropyrimidinuria, 222748
DYNC1H1	99,9	99,4	100	100	Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Charcot-Marie-Tooth disease, axonal, type 20, 614228
DYRK1A	100	100	100	100	Mental retardation, autosomal dominant 7, 614104
EBP	99,7	95,8	100	100	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
EEF1A2	100	100	99,9	99,1	Developmental and epileptic encephalopathy 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFHC1	93,1	91,6	98	98	{Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770 {Epilepsy, juvenile absence, susceptibility to, 1}, 607631
EGF	99,9	99,7	100	100	Hypomagnesemia 4, renal, 611718
EHMT1	94,5	93,7	99,6	99,5	Kleefstra syndrome 1, 610253
EIF2B1	100	100	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	99,9	99,5	100	100	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
EIF2B3	100	100	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	100	99,9	100	100	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	100	99,1	100	100	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EPM2A	94,2	91,5	100	97,7	Epilepsy, progressive myoclonic 2A (Lafora), 254780
ETHE1	99,9	97,4	100	100	Ethylmalonic encephalopathy, 602473
EXOSC3	99,5	94,9	100	100	Pontocerebellar hypoplasia, type 1B, 614678
FA2H	92	83,1	100	100	Spastic paraplegia 35, autosomal recessive, 612319

FARS2	100	100	100	100	Spastic paraplegia 77, autosomal recessive, 617046 Combined oxidative phosphorylation deficiency 14, 614946
FGD1	97,3	92,8	100	100	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF12	99,9	98,1	100	100	Developmental and epileptic encephalopathy 47, 617166
FLNA	100	99,9	100	100	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620
FOLR1	100	100	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	88,6	82,1	99,2	96,4	Rett syndrome, congenital variant, 613454
FOXRED1	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRMPD4	97,5	96,5	98,3	98,3	Mental retardation, X-linked 104, 300983
FRRS1L	79,7	69,1	99,2	95,8	Developmental and epileptic encephalopathy 37, 616981
FXYD2	100	100	100	100	Hypomagnesemia 2, renal, 154020
GABRA1	100	100	100	100	{Epilepsy, childhood absence, susceptibility to, 4}, 611136 Developmental and epileptic encephalopathy 19, 615744 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRB3	99,6	98,2	100	100	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 Developmental and epileptic encephalopathy 43, 617113
GABRG2	90,8	90,2	93	93	Febrile seizures, familial, 8, 607681 Developmental and epileptic encephalopathy 74, 618396 Epilepsy, generalized, with febrile seizures plus, type 3, 607681
GAMT	93,1	82,7	100	100	Cerebral creatine deficiency syndrome 2, 612736
GCK	95,4	95,4	92,5	92,2	Diabetes mellitus, noninsulin-dependent, late onset, 125853 MODY, type II, 125851 Diabetes mellitus, permanent neonatal 1, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485
GCSH	75,7	68,9	100	100	?Glycine encephalopathy, 605899
GLDC	89,9	82	100	99,9	Glycine encephalopathy, 605899
GLRA1	100	99,8	100	100	Hyperekplexia 1, 149400
GLRB	99,2	95,1	100	100	Hyperekplexia 2, 614619

GLUD1	94,2	82,9	100	100	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	93,8	93,8	100	100	Developmental and epileptic encephalopathy 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GOSR2	95,9	94,6	100	100	Epilepsy, progressive myoclonic 6, 614018
GPC3	99,1	94,7	100	100	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	100	99,5	100	100	Molybdenum cofactor deficiency C, 615501
GRIA3	99,7	96,1	100	99,6	Intellectual developmental disorder, X-linked, syndromic, Wu type, 300699
GRIN1	100	100	100	100	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GRIN2A	100	100	100	100	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	99,8	99,2	100	100	Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 Developmental and epileptic encephalopathy 27, 616139
GRIN2D	79,8	65,4	93,9	88,7	Developmental and epileptic encephalopathy 46, 617162
GRN	100	100	100	100	Ceroid lipofuscinosis, neuronal, 11, 614706 Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
HACE1	100	99,3	100	100	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	99	97,5	100	100	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HCFC1	98,3	93,6	100	100	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	98,5	98,2	98,5	98,5	Generalized epilepsy with febrile seizures plus, type 10, 618482 Developmental and epileptic encephalopathy 24, 615871
HECW2	100	99,1	100	100	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HLCS	100	100	100	100	Holocarboxylase synthetase deficiency, 253270
HNRNPU	99,9	98,9	100	100	Developmental and epileptic encephalopathy 54, 617391
HSD17B10	100	99,1	100	100	HSD10 mitochondrial disease, 300438
HSD17B4	95,4	93,1	96,6	96,6	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
IDH2	99,7	97,4	100	99,8	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	91,9	82,6	100	100	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	99,7	98,4	100	100	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IQSEC2	96,8	88,6	99,4	98,4	Mental retardation, X-linked 1/78, 309530

IRF2BPL	99,5	95	99,9	99,2	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
ITPA	100	100	100	100	Developmental and epileptic encephalopathy 35, 616647 [Inosine triphosphatase deficiency], 613850
JAM3	100	99,9	100	100	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANSL1	99,9	99,2	100	100	Koolen-De Vries syndrome, 610443
KATNB1	100	99,9	100	100	Lissencephaly 6, with microcephaly, 616212
KCNA1	100	99,9	100	100	Episodic ataxia/myokymia syndrome, 160120
KCNA2	100	99,6	100	100	Developmental and epileptic encephalopathy 32, 616366
KCNB1	100	99,6	100	100	Developmental and epileptic encephalopathy 26, 616056
KCNC1	100	100	100	100	Epilepsy, progressive myoclonic 7, 616187
KCNH1	98,7	98,7	98,7	98,7	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	89,3	89	100	100	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	100	100	100	100	Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes, permanent neonatal 2, with or without neurologic features, 618856 Diabetes mellitus, transient neonatal 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNMA1	94,4	93,6	100	100	Liang-Wang syndrome, 618729 {Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446
KCNQ2	91,3	89,8	100	100	Seizures, benign neonatal, 1, 121200 Developmental and epileptic encephalopathy 7, 613720 Myokymia, 121200
KCNQ3	100	99,4	99,8	99,1	Seizures, benign neonatal, 2, 121201
KCNT1	96	95,2	98,6	97,3	Epilepsy nocturnal frontal lobe, 5, 615005 Developmental and epileptic encephalopathy 14, 614959
KCNT2	99,4	97,1	100	100	Developmental and epileptic encephalopathy 57, 617771
KCTD7	95	95	100	100	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	99,8	97,9	100	100	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6B	98,8	97,9	100	100	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505
KIF5A	100	99,9	100	100	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187 {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921

KMT5B	99,9	99,1	100	100	Mental retardation, autosomal dominant 51, 617788
KPTN	100	100	100	100	Mental retardation, autosomal recessive 41, 615637
LAMB1	100	99,9	100	100	Lissencephaly 5, 615191
LGI1	98,5	97,5	100	100	Epilepsy, familial temporal lobe, 1, 600512
LIAS	100	99,1	100	100	Hyperglycinemia, lactic acidosis, and seizures, 614462
MAPK8IP3	99,4	99	100	100	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MBD5	99,9	99,9	100	100	Mental retardation, autosomal dominant 1, 156200
MECP2	100	98,7	100	99,9	Mental retardation, X-linked syndromic, Lubs type, 300260 Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, atypical, 312750 {Autism susceptibility, X-linked 3}, 300496 Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750
MED12	99,8	96,7	100	100	Ohdo syndrome, X-linked, 300895 Lujan-Fryns syndrome, 309520 Opitz-Kaveggia syndrome, 305450
MEF2C	99,9	96	100	100	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MFF	94,3	89,9	100	100	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD8	100	99,7	100	100	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MLC1	100	99	100	100	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MOCS1	99,2	95,1	100	100	Molybdenum cofactor deficiency A, 252150
MOCS2	99,6	99,5	100	100	Molybdenum cofactor deficiency B, 252160
MPDU1	100	100	100	100	Congenital disorder of glycosylation, type If, 609180
MPDZ	99,8	98,8	100	100	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MTFMT	100	99,8	100	100	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFR	97,3	96	100	100	{Schizophrenia, susceptibility to}, 181500 Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MTOR	100	99,5	100	100	Smith-Kingsmore syndrome, 616638 Focal cortical dysplasia, type II, somatic, 607341
MTRR	100	99,6	100	100	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 Homocystinuria-megaloblastic anemia, cbl E type, 236270

NACC1	100	99,8	100	100	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NANS	100	99,9	100	100	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	98,3	97,4	100	100	?Deafness, autosomal recessive 94, 618434 Combined oxidative phosphorylation deficiency 24, 616239
NBEA	92	90,6	100	100	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NDUFA1	99,9	99,3	100	100	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA11	100	100	100	99,8	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFAF1	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	95	83,4	100	99,9	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	99,8	98,2	100	100	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	100	99,5	100	100	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFB3	95,8	80,5	100	100	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB9	96,5	92,5	98,7	98,7	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFS1	100	99,5	100	100	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	90,7	90,6	91,9	90,7	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100	99,4	100	100	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFV1	98	96,1	100	100	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	86,9	76,9	100	100	Mitochondrial complex I deficiency, nuclear type 7, 618229
NECAP1	100	100	100	100	Developmental and epileptic encephalopathy 21, 615833
NEDD4L	72	71,5	100	100	Periventricular nodular heterotopia 7, 617201
NEU1	99,7	97,7	100	100	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEXMIF	100	99,5	100	100	Mental retardation, X-linked 98, 300912
NGLY1	100	99,8	100	100	Congenital disorder of deglycosylation, 615273
NHLRC1	100	98,7	100	100	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NPRL2	100	100	100	100	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	100	99,6	100	100	Epilepsy, familial focal, with variable foci 3, 617118
NRXN1	97,4	96,9	100	99,8	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NUBPL	99,7	98,4	100	100	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUS1	60	44,5	100	100	Mental retardation, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
OCLN	100	100	100	100	Pseudo-TORCH syndrome 1, 251290

OFD1	88	73,7	100	99,9	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OPHN1	99,5	97,6	99,9	98,8	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
PACS1	98,8	96,9	100	100	Schuurs-Hoeijmakers syndrome, 615009
PACS2	99,3	96,2	100	99,8	Developmental and epileptic encephalopathy 66, 618067
PAFAH1B1	94,1	87,1	100	100	Subcortical laminar heterotopia, 607432 Lissencephaly 1, 607432
PAK3	99,3	95,9	100	99,8	Mental retardation, X-linked 30/47, 300558
PC	99,8	97,3	100	100	Pyruvate carboxylase deficiency, 266150
PCDH19	100	98,9	100	100	Developmental and epileptic encephalopathy 9, 300088
PDHA1	99,4	97,1	100	100	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	99,1	97,5	100	100	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	99,9	99,4	100	100	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	100	100	100	100	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDX1	93	82,4	100	100	{Diabetes mellitus, type II, susceptibility to}, 125853 Pancreatic agenesis 1, 260370 MODY, type IV, 606392
PET100	100	99,6	100	100	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	99,9	99,4	100	100	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	96,8	89,7	100	99,9	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX12	100	100	100	100	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	100	100	100	100	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	96,7	90,8	100	100	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	97,9	94,2	100	100	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	99,9	98,5	100	100	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX26	100	100	100	100	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	100	99,3	100	100	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370

PEX5	99,9	99	100	100	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	94,5	86,7	100	100	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PGAP3	63,5	59,6	100	100	Hyperphosphatasia with mental retardation syndrome 4, 615716
PHF6	97,8	88,3	99,9	98,9	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	99,9	98,8	100	100	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	93,8	86,7	100	100	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	93,8	91,5	98,8	98,8	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	95,8	87,3	100	100	Developmental and epileptic encephalopathy 55, 617599
PIGT	98,1	98,1	100	100	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PLA2G6	92,2	90,7	92,3	92,3	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLCB1	100	99,8	100	100	Developmental and epileptic encephalopathy 12, 613722
PLP1	100	99,2	100	100	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	98,2	90,1	100	100	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMM2	100	100	100	100	Congenital disorder of glycosylation, type Ia, 212065
PNKP	100	100	100	100	Ataxia-oculomotor apraxia 4, 616267 ?Charcot-Marie-Tooth disease, type 2B2, 605589 Microcephaly, seizures, and developmental delay, 613402
PNPO	99,9	97,7	100	100	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLG	100	99,3	100	100	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
PPP2R1A	91,6	91,5	93,6	93,6	Mental retardation, autosomal dominant 36, 616362
PPP2R5D	100	100	100	100	Mental retardation, autosomal dominant 35, 616355
PPT1	90,3	90,3	82,5	82,5	Ceroid lipofuscinosis, neuronal, 1, 256730

PQBP1	100	100	100	100	Renpenning syndrome, 309500
PRF1	91,2	90,8	100	100	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PRICKLE1	100	100	100	100	Epilepsy, progressive myoclonic 1B, 612437
PRRT2	100	99,6	100	100	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066
PSAP	100	100	100	100	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Combined SAP deficiency, 611721 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PTRH2	100	100	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	99,9	99,1	100	100	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUM1	100	99,9	100	100	Spinocerebellar ataxia 47, 617931
PURA	99	95,2	100	99,8	Mental retardation, autosomal dominant 31, 616158
PYCR2	100	99,1	100	100	Leukodystrophy, hypomyelinating, 10, 616420
QARS1	100	100	100	100	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	100	99,7	100	100	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB39B	100	100	100	100	Waisman syndrome, 311510 Mental retardation, X-linked 72, 300271
RARS2	100	99,8	100	100	Pontocerebellar hypoplasia, type 6, 611523
RNASEH2A	100	100	100	100	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	80,6	78,1	91	90,9	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100	99,5	100	100	Aicardi-Goutieres syndrome 3, 610329
ROGDI	98,4	95,2	99,9	99,1	Kohlschutter-Tonz syndrome, 226750
RPS6KA3	98,4	94,5	100	98,9	Mental retardation, X-linked 19, 300844 Coffin-Lowry syndrome, 303600
RRM2B	100	99,7	100	100	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
SAMHD1	98,7	98,4	100	100	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SCARB2	100	99,8	100	100	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN1A	99,9	99,5	100	100	Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634

					Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403
SCN1B	98	96,4	99,8	99,3	Atrial fibrillation, familial, 13, 615377 Developmental and epileptic encephalopathy 52, 617350 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838
SCN2A	99,6	97,6	100	100	Episodic ataxia, type 9, 618924 Developmental and epileptic encephalopathy 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN3A	99,8	99,2	100	100	Epilepsy, familial focal, with variable foci 4, 617935 Developmental and epileptic encephalopathy 62, 617938
SCN8A	100	99,8	100	100	Seizures, benign familial infantile, 5, 617080 Developmental and epileptic encephalopathy 13, 614558 Cognitive impairment with or without cerebellar ataxia, 614306 ?Myoclonus, familial, 2, 618364
SEMA6B	80,6	73,6	100	100	Epilepsy, progressive myoclonic, 11, 618876
SEPSECS	100	100	100	100	Pontocerebellar hypoplasia type 2D, 613811
SERPINI1	99,9	99	100	100	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SHANK3	91,6	81,5	96	91,9	{Schizophrenia 15}, 613950 Phelan-McDermid syndrome, 606232
SIK1	98,7	94,4	100	100	Developmental and epileptic encephalopathy 30, 616341
SLC12A5	83,9	83,8	97,4	97,4	Developmental and epileptic encephalopathy 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC13A5	100	99,9	100	100	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905
SLC16A1	100	99,3	100	100	Monocarboxylate transporter 1 deficiency, 616095 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340
SLC19A3	97,8	97,6	98,7	98,7	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A2	96,1	95,4	100	100	Developmental and epileptic encephalopathy 41, 617105
SLC25A1	95,8	88,6	99,5	97,8	Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A15	99,8	98,1	100	100	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	98,6	95,8	100	100	Developmental and epileptic encephalopathy 3, 609304
SLC2A1	92,8	92,8	100	100	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 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC35A2	99,9	98,4	100	100	Congenital disorder of glycosylation, type IIm, 300896
SLC6A1	96,7	96,7	100	100	Myoclonic-atonic epilepsy, 616421
SLC6A8	93,5	81,6	100	99,8	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	95,2	91,6	100	98,4	Mental retardation, X-linked syndromic, Christianson type, 300243
SMARCA2	96,7	96,2	97,4	96,8	Nicolaides-Baraitser syndrome, 601358
SMC1A	100	98,7	100	99,8	Cornelia de Lange syndrome 2, 300590 Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMPD4	99,4	94,2	100	100	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMS	91,5	78,5	100	99,9	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAP25	100	99,9	100	100	?Myasthenic syndrome, congenital, 18, 616330
SPATA5	100	99,7	100	100	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPTAN1	99,1	98,6	100	100	Developmental and epileptic encephalopathy 5, 613477
ST3GAL3	68,8	68,6	95,3	95,2	Developmental and epileptic encephalopathy 15, 615006 Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	85	84,2	98,7	98,4	Salt and pepper developmental regression syndrome, 609056
STX1B	100	100	100	100	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	96,8	96,5	100	100	Developmental and epileptic encephalopathy 4, 612164
SUOX	100	100	100	100	Sulfite oxidase deficiency, 272300
SYN1	81,9	73,2	100	99,6	?Intellectual developmental disorder, X-linked 50, 300115 Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	99,4	98,1	100	100	Mental retardation, autosomal dominant 5, 612621
SYNJ1	99,9	99,4	100	100	Developmental and epileptic encephalopathy 53, 617389 Parkinson disease 20, early-onset, 615530
SYP	99,9	96,7	100	100	Mental retardation, X-linked 96, 300802
SZT2	99,6	99,5	100	99,9	Developmental and epileptic encephalopathy 18, 615476
TANGO2	100	99,3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TBC1D23	99,7	97,2	100	100	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	100	100	100	100	Developmental and epileptic encephalopathy 16, 615338 Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 DOORS syndrome, 220500 Deafness, autosomal dominant 65, 616044 Myoclonic epilepsy, infantile, familial, 605021 Deafness, autosomal recessive 86, 614617

TBCD	96,2	94,4	100	100	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	99,8	97,5	100	100	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TCF4	100	99,8	100	100	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TDP2	100	99,4	100	100	Spinocerebellar ataxia, autosomal recessive 23, 616949
TOE1	100	100	100	100	Pontocerebellar hypoplasia, type 7, 614969
TPP1	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
TREX1	100	100	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRPM3	100	99,5	100	100	No OMIM disease ID
TRPM6	99,9	99,5	100	100	Hypomagnesemia 1, intestinal, 602014
TSC1	99,8	98,8	100	100	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	100	99,6	100	100	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
TSEN15	79	77,2	100	100	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	100	99,6	100	100	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	96,3	94,3	99,9	98,9	Pontocerebellar hypoplasia type 4, 225753 Pontocerebellar hypoplasia type 2A, 277470 ?Pontocerebellar hypoplasia type 5, 610204
TUBA1A	99,9	97	100	100	Lissencephaly 3, 611603
TUBB2A	97	95,7	100	100	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100	99,5	100	100	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB4A	95,9	94	97,1	96	Leukodystrophy, hypomyelinating, 6, 612438 Dystonia 4, torsion, autosomal dominant, 128101
TUBG1	100	100	100	100	Cortical dysplasia, complex, with other brain malformations 4, 615412
UBA5	97,8	86,8	100	100	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Developmental and epileptic encephalopathy 44, 617132
UBE3A	99,1	94,8	100	100	Angelman syndrome, 105830
UBTF	100	99,4	100	100	Neurodegeneration, childhood-onset, with brain atrophy, 617672

UGP2	99	98,6	96,3	96,3	Developmental and epileptic encephalopathy 83, 618744
VPS11	94,9	93,6	100	100	Leukodystrophy, hypomyelinating, 12, 616683
VPS53	91,5	90,7	100	99,3	Pontocerebellar hypoplasia, type 2E, 615851
WDR26	88,7	83,9	94,2	91,7	Skraban-Deardorff syndrome, 617616
WDR45	98,1	92,4	100	100	Neurodegeneration with brain iron accumulation 5, 300894
WWOX	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 12, 614322 Esophageal squamous cell carcinoma, somatic, 133239 Developmental and epileptic encephalopathy 28, 616211
XK	99,8	98,1	100	100	McLeod syndrome with or without chronic granulomatous disease, 300842
YWHAG	100	100	100	100	Developmental and epileptic encephalopathy 56, 617665
ZEB2	99,9	99,1	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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID 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 : March 23rd , 2021.
This list is accurate for panel version DG 3.1.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors