INTELLECTUAL DISABILITY GENE PANEL DG 2.3.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM ID*
A2ML1	100,9	99,90%	99,10%	Noonan-like syndrome (Vissers et al. 2015)
ABCC9	105,1	99,90%	96,10%	Cardiomyopathy, dilated, 1O, 608569 Atrial fibrillation, familial, 12, 614050 Hypertrichotic osteochondrodysplasia, 239850
ABCD1	30,2	72,00%	59,70%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD4	99,8	100,00%	98,20%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD5	122,6	100,00%	100,00%	Chanarin-Dorfman syndrome, 275630
ACAD9	84,8	100,00%	99,20%	ACAD9 deficiency, 611126
ACO2	72,5	92,60%	83,00%	Infantile cerebellar-retinal degeneration, 614559
ACOX1	75,1	98,20%	91,80%	Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	87,2	99,70%	98,20%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	67,2	100,00%	95,80%	Mental retardation, X-linked 63, 300387
ACTB	56,1	99,40%	93,00%	Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	60,1	97,60%	94,20%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ACVR1	90,3	100,00%	96,60%	Fibrodysplasia ossificans progressiva, 135100
ACY1	79,8	99,70%	95,40%	Aminoacylase 1 deficiency, 609924
ADAR	130,6	99,60%	99,40%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
ADAT3	51,2	100,00%	98,70%	Mental retardation, autosomal recessive 36, 615286
ADCK3	98,1	99,80%	97,50%	Coenzyme Q10 deficiency, primary, 4, 612016
ADK	114,5	94,10%	94,10%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	182	100,00%	100,00%	Mental retardation, autosomal dominant, 28, 615873
ADSL	121,7	99,90%	99,00%	ade(-)I bifunctional Adenylosuccinase deficiency, 103050
AFF2	65,1	97,80%	93,50%	Mental retardation, X-linked, FRAXE type, 309548
AGA	111,2	100,00%	97,90%	Aspartylglucosaminuria, 208400

AGPAT2	55,1	91,50%	89,80%	Lipodystrophy, congenital generalized, type 1, 608594
AGTR2	96,6	100,00%	100,00%	Mental retardation, X-linked 88, 300852
AHCY	71,5	90,40%	70,80%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHI1	105,6	100,00%	98,60%	Joubert syndrome-3, 608629
AIFM1	58,2	98,50%	87,20%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490
AIMP1	123,5	100,00%	100,00%	Leukodystrophy, hypomyelinating, 3, 260600
AK1	90	100,00%	100,00%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AKT3	106,5	100,00%	100,00%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
ALDH18A1	92,1	98,20%	93,10%	Cutis laxa, autosomal recessive, type IIIA, 219150
ALDH3A2	101,9	100,00%	100,00%	Sjogren-Larsson syndrome, 270200
ALDH4A1	63,5	91,40%	85,50%	Hyperprolinemia, type II, 239510
ALDH5A1	63,1	96,70%	93,90%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALG1	46,4	45,10%	45,10%	ngenital disorder of glycosylation, type Ik, 608540
ALG12	91,6	100,00%	100,00%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	59,7	92,10%	84,50%	Congenital disorder of glycosylation, type Is, 300884
ALG2	105,6	100,00%	97,60%	Congenital disorder of glycosylation, type Ii, 607906
ALG3	83	99,80%	92,60%	Congenital disorder of glycosylation, type Id, 601110
ALG6	107,5	100,00%	99,20%	Congenital disorder, type Ic, 603147
ALG9	81,9	99,10%	96,90%	Congenital disorder of glycosylation, type Il, 608776
ALX1	161,5	100,00%	100,00%	Frontonasal dysplasia 3, 613456
ALX4	69,8	100,00%	100,00%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMPD2	97	98,80%	94,60%	Pontocerebellar hypoplasia, type 9, 615809 ?Spastic paraplegia 63, 615686
AMT	117	100,00%	100,00%	Glycine encephalopathy, 605899
ANK3	145,5	100,00%	99,70%	?Mental retardation, autosomal recessive, 37
ANKH	107,6	100,00%	100,00%	Craniometaphyseal dysplasia, 123000 Chondrocalcinosis 2, 118600
ANKRD11	108	91,00%	87,20%	KBG syndrome, 148050
ANO10	98,1	100,00%	97,20%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANTXR1	76,2	95,40%	88,30%	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089
AP1S2	73	76,70%	76,60%	Mental retardation, X-linked syndromic, Fried type, 300630

AP3B1	109,5	100,00%	100,00%	Hermansky-Pudlak syndrome 2, 608233
AP4B1	95,5	100,00%	99,90%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	132,4	100,00%	100,00%	Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	96,7	100,00%	99,50%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	69,9	91,90%	86,00%	Spastic paraplegia 52, autosomal recessive, 614067
APTX	121	99,00%	94,50%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARFGEF2	105	100,00%	99,30%	Periventricular heterotopia with microcephaly, 608097
ARG1	136,7	95,60%	89,80%	Argininemia, 207800
ARHGEF6	54,6	92,70%	89,80%	Mental retardation, X-linked 46, 300436
ARHGEF9	45,6	94,60%	83,80%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1A	97,8	99,00%	96,90%	Mental retardation, autosomal dominant 14, 614607
ARID1B	104,1	99,50%	96,00%	Mental retardation, autosomal dominant 12, 614562
ARL13B	121,8	100,00%	95,50%	Joubert syndrome 8, 612291
ARL6	155,1	100,00%	100,00%	Bardet-Biedl syndrome 3, 209900 {Bardet-Biedl syndrome 1, modifier of}, 209900 Retinitis pigmentosa 55, 613575
ARSE	42,7	89,60%	72,40%	Chondrodysplasia punctata, X-linked recessive, 302950
ARX	29,5	82,60%	62,90%	Epileptic encephalopathy, early infantile, 1, 308350 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Proud syndrome, 300004 Partington syndrome, 309510 Hydranencephaly with abnormal genitalia, 300215
ASL	81,8	100,00%	91,90%	Argininosuccinic aciduria, 207900
ASNS	61,2	93,60%	80,30%	Asparagine synthetase deficiency, 615574
ASPA	110,9	100,00%	100,00%	Canavan disease, 271900
ASPM	139	100,00%	99,60%	Microcephaly 5, primary, autosomal recessive, 608716
ASXL1	140,3	98,50%	96,70%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	146,6	99,90%	99,20%	Bainbridge-Ropers syndrome, 615485
ATIC	105,9	100,00%	98,80%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP1A2	96,2	100,00%	97,90%	Migraine, familial hemiplegic, 2, 602481 Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481

ATP2A2	113,1	100,00%	99,50%	Darier disease, 124200 Acrokeratosis verruciformis, 101900
ATP6AP2	27,5	89,70%	69,10%	Mental retardation, X-linked, with epilepsy, 300423
ATP6V0A2	96,9	100,00%	98,40%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP7A	61,8	100,00%	95,30%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATP8A2	94,5	99,80%	97,10%	?Cerebellar ataxia, mental retardation and dysequilibrium syndrome 4, 615268
ATR	119,6	100,00%	99,80%	Seckel syndrome 1, 210600 Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	70	99,80%	97,60%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUH	95,1	93,00%	90,00%	3-methylglutaconic aciduria, type I, 250950
AUTS2	112,6	100,00%	99,50%	Mental Retardation, autosomal dominant 26, 615834
B3GALTL	106,8	100,00%	95,30%	Peters-plus syndrome, 261540
B4GALT1	83,4	97,60%	97,60%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	80	99,60%	93,20%	Ehlers-Danlos syndrome, progeroid type, 1, 130070
BBS1	116,1	99,80%	98,40%	Bardet-Biedl syndrome 1, 209900
BBS10	115,8	100,00%	100,00%	Bardet-Biedl syndrome 10, 209900
BBS12	132,3	100,00%	100,00%	Bardet-Biedl syndrome 12, 209900
BBS2	114,4	100,00%	100,00%	Bardet-Biedl syndrome 2, 209900
BBS4	98,5	96,10%	93,40%	Bardet-Biedl syndrome 4, 209900
BBS5	137,8	100,00%	100,00%	Bardet-Biedl syndrome 5, 209900
BBS7	119,1	100,00%	98,90%	Bardet-Biedl syndrome 7, 209900
BBS9	122,1	100,00%	98,60%	Bardet-Biedl syndrome 9, 209900
BCKDHA	101,6	99,50%	97,00%	Maple syrup urine disease, type Ia, 248600
BCKDHB	89,7	100,00%	98,50%	Maple syrup urine disease, type Ib, 248600
BCOR	59,3	99,60%	95,00%	Microphthalmia, syndromic 2, 300166
BCS1L	133,7	100,00%	100,00%	Mitochondrial complex III deficiency, nuclear type 1, 124000 Leigh syndrome, 256000 Bjornstad syndrome, 262000 GRACILE syndrome, 603358

BLM	113,4	99,90%	98,90%	Bloom syndrome, 210900
BRAF	73,3	100,00%	93,80%	Melanoma, malignant, somatic Colorectal cancer, somatic Adenocarcinoma of lung, somatic, 211980 Nonsmall cell lung cancer, somatic Cardiofaciocutaneous syndrome, 115150 Noonan syndrome 7, 613706 LEOPARD syndrome 3, 613707
BRWD3	57,3	98,50%	95,10%	Mental retardation, X-linked 93, 300659
BSCL2	101,5	100,00%	100,00%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type V, 600794
BTD	144	100,00%	100,00%	Biotinidase deficiency, 253260
BUB1B	113,6	100,00%	98,90%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300 [Premature chromatid separation trait], 176430
C12orf57	65,9	100,00%	94,10%	Temtamy syndrome, 218340
C12orf65	163,2	100,00%	100,00%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55,autosomal recessive, 615035
C5orf42	120,5	100,00%	100,00%	Joubert syndrome 17, 614615
CA2	147,9	100,00%	100,00%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA8	80,1	100,00%	94,90%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNG2	99,9	100,00%	100,00%	Mental retardation, autosomal dominant 10, 614256
CAMTA1	113,7	95,70%	95,40%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CASK	47,2	99,20%	90,00%	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422 Mental retardation, with or without nystagmus, 300422
CBL	122,9	100,00%	100,00%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CBS	74,6	97,70%	87,30%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CC2D1A	93,7	99,90%	97,50%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	94,8	98,20%	97,70%	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360

CCBE1	88,4	97,10%	87,30%	Hennekam lymphangiectasia-lymphedema syndrome, 235510
CCDC78	88,3	100,00%	100,00%	Myopathy, centronuclear, 4, 614807
CDH15	77,3	100,00%	96,10%	Mental retardation, autosomal dominant 3, 612580
CDK5RAP2	97,2	99,50%	95,60%	Microcephaly 3, primary, autosomal recessive, 604804
CDKL5	65,1	98,50%	94,50%	Epileptic encephalopathy, early infantile, 2, 300672 Angelman syndrome-like, 105830
CDON	111,9	100,00%	98,00%	Holoprosencephaly 11, 614226
CENPJ	130,6	100,00%	100,00%	Microcephaly 6, primary, autosomal recessive, 608393 Seckel syndrome 4, 613676
CEP135	122,9	100,00%	98,10%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	131,8	99,70%	98,90%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP290	95,3	100,00%	98,90%	Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Bardet-Biedl syndrome 14, 209900
CEP41	89,6	100,00%	100,00%	Joubert syndrome 15, 614464
CHD2	120	99,60%	97,60%	Epileptic encephalopathy, childhood-onset, 615369
CHD7	115,7	100,00%	98,90%	CHARGE syndrome, 214800 {Scoliosis, idiopathic 3}, 608765 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
CHD8	113,1	100,00%	99,80%	{Autism, susceptibility to, 18}, 615032
CHKB	81,8	93,70%	91,40%	Muscular dystrophy, congenital, megaconial type, 602541
CLCNKB	77,3	88,10%	84,60%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLIC2	39,1	94,20%	63,00%	Mental retardation, X-linked, syndromic 32, 300886
CLN3	76	97,40%	96,80%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	143	95,80%	91,40%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	67,5	100,00%	81,70%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	142,7	100,00%	100,00%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003

CNTNAP2	102,1	100,00%	99,50%	Cortical dysplasia-focal epilepsy syndrome, 610042 {Autism susceptibility 15}, 612100 Pitt-Hopkins like syndrome 1, 610042
COG1	119,7	99,10%	98,10%	Congenital disorder of glycosylation, type IIg, 611209
COG6	98,9	98,70%	94,40%	Congenital disorder of glycosylation, type 2l, 614576 Shaheen syndrome, 615328
COG7	78,4	100,00%	95,70%	Congenital disorder of glycosylation, type IIe, 608779
COG8	108,6	100,00%	99,10%	Congenital disorder of glycosylation, type IIh, 611182
COL4A1	79,8	98,40%	94,20%	Porencephaly 1, 175780 Brain small vessel disease with hemorrhage, 607595 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle, 611773 Brain small vessel disease with Axenfeld-Rieger anomaly, 607595 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A2	81	100,00%	95,10%	Porencephaly 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3BP	103,4	100,00%	100,00%	Mental retardation, autosomal dominant 34, 616351
COLEC11	103,7	100,00%	100,00%	3MC syndrome 2, 265050
COQ2	69,8	94,90%	81,90%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COX10	124,5	99,50%	94,70%	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110
COX15	80,9	100,00%	95,90%	Leigh syndrome due to cytochrome c oxidase deficiency, 256000 Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119
CPS1	104,9	100,00%	100,00%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CRADD	98	79,80%	76,70%	Mental retardation, autosomal recessive 34, 614499
CRBN	134,3	100,00%	100,00%	Mental retardation, autosomal recessive 2, 607417
CREBBP	82,3	99,60%	97,90%	Rubinstein-Taybi syndrome, 180849
CTCF	111,4	100,00%	99,30%	Mental retardation, autosomal dominant 21, 615502
CTDP1	75,3	89,10%	87,30%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168

CTNNB1	119,9	99,50%	97,30%	Mental retardation, autosomal dominant 19, 615075 Colorectal cancer, somatic, 114500 Hepatocellular carcinoma, somatic, 114550 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600
CTSA	95,2	100,00%	100,00%	Galactosialidosis, 256540
CTSD	87,5	100,00%	95,30%	Ceroid lipofuscinosis, neuronal, 10, 610127
CUBN	83,8	98,60%	96,00%	Megaloblastic anemia-1, Finnish type, 261100
CUL4B	59	98,40%	95,80%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CYB5R3	92,1	97,50%	93,40%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
D2HGDH	63	94,20%	85,40%	D-2-hydroxyglutaric aciduria, 600721
DARS2	108,6	100,00%	99,40%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	108,4	100,00%	98,90%	Maple syrup urine disease, type II, 248600
DCAF17	92,3	100,00%	96,40%	Woodhouse-Sakati syndrome, 241080
DCX	57,3	98,70%	96,90%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DDHD2	98,7	100,00%	99,40%	Spastic paraplegia 54, autosomal recessive, 615033
DDX11	9,4	19,80%	13,30%	Warsaw breakage syndrome, 613398
DEAF1	68,1	82,60%	79,40%	Mental retardation, autosomal dominant 24, 615828
DHCR24	87	99,60%	95,40%	Desmosterolosis, 602398
DHCR7	107,5	100,00%	95,90%	Smith-Lemli-Opitz syndrome, 270400
DHFR	46	81,70%	58,60%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHTKD1	106,3	100,00%	99,50%	2-aminoadipic 2-oxoadipic aciduria, 204750 Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DIP2B	99,6	100,00%	97,90%	Mental retardation, FRA12A type, 136630
DKC1	46	99,90%	91,00%	Dyskeratosis congenita, X-linked, 305000
DLD	136,8	100,00%	100,00%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	38	91,20%	72,50%	Mental retardation, X-linked 90, 300850
DMD	55	99,50%	93,10%	Duchenne muscular dystrophy, 310200 Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045
DMPK	97,8	98,60%	94,50%	Myotonic dystrophy 1, 160900
DNAJC19	60,5	79,70%	78,90%	3-methylglutaconic aciduria, type V, 610198

DNMT3A	79,9	100,00%	98,70%	Tatton-Brown-Rahman syndrome, 615879
DNMT3B	93,2	100,00%	98,40%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK8	83,8	100,00%	96,30%	Mental retardation, autosomal dominant 2, 614113 Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DPAGT1	92,3	100,00%	96,60%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, with tubular aggregates 2, 614750
DPM1	139	90,70%	90,60%	Congenital disorder of glycosylation, type Ie, 608799
DPP6	101,4	94,40%	90,00%	Mental retardation, autosomal dominant 33, 616311 {Ventricular fibrillation, paroxysmal familial, 2}
DPYD	114,5	99,30%	97,20%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DST	145,3	99,60%	99,30%	Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, sutosomal recessive 2, 615425
DYM	96,3	97,40%	97,10%	Dyggve-Melchior-Clausen disease, 223800 Smith-McCort dysplasia, 607326 Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388
DYNC1H1	110,7	98,90%	95,60%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant, AD, 158600
DYRK1A	132,8	99,60%	98,80%	Mental retardation, autosomal dominant 7, 614104
EBP	45,6	94,50%	76,90%	Chondrodysplasia punctata, X-linked dominant, 302960
EFTUD2	82,3	99,80%	97,50%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EHMT1	92,5	98,80%	91,80%	Kleefstra syndrome, 610253
EIF2AK3	104,7	94,80%	91,70%	Wolcott-Rallison syndrome, 226980
EIF4G1	102,5	100,00%	99,20%	Parkinsons disease 18, 614251
ELOVL4	95,8	100,00%	100,00%	Stargardt disease 3, 600110 Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
EMX2	96,8	100,00%	100,00%	Schizencephaly, 269160
EP300	129,8	99,20%	98,10%	Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPB41L1	89,8	99,40%	95,50%	Mental retardation, autosomal dominant 11, 614257

ERCC2	85,7	100,00%	94,50%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy, 601675 Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	117,1	100,00%	99,20%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy, 601675
ERCC5	115,4	98,60%	96,90%	Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	147,6	98,50%	97,10%	Cockayne syndrome, type B, 133540 Cerebrooculofacioskeletal syndrome 1, 214150 De Sanctis-Cacchione syndrome, 278800 {Macular degeneration, age-related, susceptibility to 5}, 613761 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980
ERCC8	87,3	100,00%	99,10%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERLIN2	105,5	99,40%	96,50%	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	84,1	100,00%	98,30%	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
ETFB	101,6	100,00%	100,00%	Glutaric acidemia 2B, 231680
ETHE1	55,1	95,10%	90,40%	Ethylmalonic encephalopathy, 602473
EXOSC3	47,5	89,80%	66,50%	Pontocerebellar hypoplasia, type 1B, 614678
EZH2	77,4	98,60%	94,00%	Weaver syndrome, 277590
FAM126A	128,1	100,00%	100,00%	Leukodystrophy, hypomyelinating, 5, 610532
FBN1	94,9	99,80%	97,90%	Marfan syndrome, 154700 Ectopia lentis, familial, 129600 MASS syndrome, 604308 Weill-Marchesani syndrome 2, dominant, 608328 Aortic aneurysm, ascending, and dissection Stiff skin syndrome, 184900 Acromicric dysplasia, 102370 Geleophysic dysplasia 2, 614185
FBXO31	56	97,80%	92,10%	Mental retardation, autosomal recessive 45, 615979
FGD1	44,9	93,60%	81,20%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400

FGFR1	111,5	100,00%	95,60%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Osteoglophonic dysplasia, 166250 Trigonocephaly 1, 190440 Hartsfield syndrome, 615465
FGFR2	112,7	97,80%	97,80%	Crouzon syndrome, 123500 Jackson-Weiss syndrome, 123150 Beare-Stevenson cutis gyrata syndrome, 123790 Pfeiffer syndrome, 101600 Apert syndrome, 101200 Saethre-Chotzen syndrome, 101400 Craniosynostosis, nonspecific Gastric cancer, somatic, 613659 Craniofacial-skeletal-dermatologic dysplasia, 101600 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Scaphocephaly and Axenfeld-Rieger anomaly LADD syndrome, 149730 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592
FGFR3	71,8	96,00%	88,60%	Achondroplasia, 100800 Hypochondroplasia, 146000 Thanatophoric dysplasia, type I, 187600 Crouzon syndrome with acanthosis nigricans, 612247 Muenke syndrome, 602849 Bladder cancer, somatic, 109800 Colorectal cancer, somatic, 114500 Cervical cancer, somatic, 603956 LADD syndrome, 149730 CATSHL syndrome, 610474 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Spermatocytic seminoma, somatic, 273300
FH	86,7	99,30%	91,20%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800

FKRP	90	100,00%	100,00%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FKTN	115,6	100,00%	100,00%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588
FLNA	60	98,30%	92,10%	Heterotopia, periventricular, 300049 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, ED variant, 300537 FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048
FLVCR1	92,4	100,00%	100,00%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FMN2	103,9	88,90%	85,30%	Mental retardation, autosomal recessive 47, 616193
FMR1	55,2	100,00%	93,30%	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360
FOXG1	73,4	89,40%	76,00%	Rett syndrome, congenital variant, 613454
FOXP1	98,3	100,00%	99,50%	Mental retardation with language impairment and autistic features, 613670
FOXP2	106,1	100,00%	100,00%	Speech-language disorder-1, 602081
FRAS1	96,4	98,80%	95,90%	Fraser syndrome, 219000
FTO	108,6	99,10%	97,20%	Growth retardation, developmental delay, coarse facies, and early death, 612938
FTSJ1	50,3	87,70%	79,20%	Mental retardation, X-linked 9, 309549
FUCA1	74,9	100,00%	96,40%	Fucosidosis, 230000
GABRA1	121,5	100,00%	96,20%	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4} {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GAD1	95,4	100,00%	98,30%	Cerebral palsy, spastic quadriplegic, 1, 603513

GALE	109,2	100,00%	100,00%	Galactose epimerase deficiency, 230350
GALT	104,1	100,00%	97,90%	Galactosemia, 230400
GAMT	98,9	100,00%	92,40%	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	117,7	99,30%	96,90%	Mental retardation, autosomal dominant 18, 615074
GATM	88,4	100,00%	92,70%	Cerebral creatine deficiency syndrome 3, 612718
GCH1	98,2	100,00%	100,00%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCSH	14,7	51,70%	37,20%	Glycine encephalopathy, 605899
GDI1	59,8	100,00%	92,20%	Mental retardation, X-linked 41, 300849
GFAP	73,7	100,00%	97,60%	Alexander disease, 203450
GJB1	77,6	100,00%	100,00%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJC2	54,1	95,80%	81,80%	Leukodystrophy, hypomyelinating, 2, 608804 Spastic paraplegia 44, autosomal recessive, 613206 Lymphedema, hereditary, IC, 613480
GK	24,6	76,30%	53,80%	Glycerol kinase deficiency, 307030
GLB1	75,6	100,00%	95,90%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLDC	55,8	95,90%	86,20%	Glycine encephalopathy, 605899
GLI2	99,2	99,90%	95,80%	Holoprosencephaly-9, 610829
GLI3	117,2	100,00%	99,80%	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700 Polydactyly, postaxial, types A1 and B, 174200 {Hypothalamic hamartomas, somatic}, 241800
GM2A	96,7	100,00%	98,00%	GM2-gangliosidosis, AB variant, 272750
GMPPA	119,7	100,00%	100,00%	Alacrima, achalasia and mental retardation syndrome, 615510
GMPPB	108,2	100,00%	100,00%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A,14, 6135350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GNAO1	105,6	100,00%	99,00%	Epileptic encephalopathy, early infantile, 17, 615473

GNAS	117,6	99,90%	97,10%	Pseudohypoparathyroidism Ia, 103580 McCune-Albright syndrome, 174800 Pseudohypoparathyroidism Ic, 612462 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ib, 603233 Prolonged bleeding time, brachydactyly and mental retardation Acromegaly, 102200 Pseudopseudohypoparathyroidism, 612463 Prolonged bleeding time, brachydactyly, and mental retardation ACTH-independent macronodular adrenal hyperplasia, 219080
GNPAT	119,7	99,20%	97,80%	Chondrodysplasia punctata, rhizomelic, type 2, 222765
GNS	78,7	93,50%	82,90%	Mucopolysaccharidosis type IIID, 252940
GPC3	45,5	100,00%	96,10%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	111,3	100,00%	100,00%	Molybdenum cofactor deficiency, type C, 252150
GPR56	84,3	100,00%	98,80%	Polymicrogyria, bilateral frontoparietal, 606854
GPT2	83,9	100,00%	92,30%	?Mental retardation, autosomal recessive 49, 616281
GRIA3	54,9	97,90%	89,10%	Mental retardation, X-linked 94, 300699
GRID2	130,6	100,00%	98,80%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	109,3	96,30%	96,30%	Mental retardation, autosomal recessive, 6, 611092
GRIN1	81,4	99,90%	94,10%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	128,4	99,70%	98,00%	Epilepsy with neurodevelopmental defects, 613971
GRIN2B	134,3	99,30%	98,50%	Mental retardation, autosomal dominant 6, 613970
GRM1	138,1	99,90%	98,00%	Spinocerebellar ataxia, autosomal recessive 13, 614831
GSS	84,5	100,00%	100,00%	Hemolytic anemia due to glutathione synthetase deficiency, 231900
GTF2H5	88,2	100,00%	100,00%	Trichothiodystrophy, complementation group A, 601675
GUSB	64,6	88,20%	80,10%	Mucopolysaccharidosis VII, 253220
HAX1	131,1	100,00%	100,00%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	53,1	98,30%	88,10%	Microphthalmia, syndromic 7, 309801
HCFC1	37,1	93,60%	80,50%	Mental retardation, X-linked 3, 309541
HCN1	101,1	100,00%	100,00%	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	64,2	95,30%	89,30%	Brachydactyly-mental retardation syndrome, 600430
HDAC6	56,2	94,60%	83,00%	Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863

HDAC8	51,5	99,90%	87,00%	Wilson-Turner syndrome, 309585 Cornelia de Lange syndrome 5, 300882
HERC2	61	63,20%	59,30%	Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
HESX1	99	100,00%	99,90%	Septooptic dysplasia, 182230 Pituitary hormone deficiency, combined, 5, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	90,8	100,00%	99,40%	Tay-Sachs disease, 272800 GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800
HEXB	115,4	100,00%	100,00%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HLCS	131,3	100,00%	100,00%	Holocarboxylase synthetase deficiency, 253270
HOXA1	121,7	100,00%	100,00%	Bosley-Salih-Alorainy syndrome, 601536 Athabaskan brainstem dysgenesis syndrome, 601536
HPD	94,6	99,80%	98,10%	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPRT1	50	97,10%	81,10%	Lesch-Nyhan syndrome, 300322
HRAS	93,6	100,00%	94,70%	{Bladder cancer, somatic}, 109800 Costello syndrome, 218040 {Thyroid carcinoma, follicular, somatic}, 188470 Congenital myopathy with excess of muscle spindles, 218040 {Nevus sebaceous, somatic}, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
HSD17B10	56,9	96,30%	89,10%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked syndromic 10, 300220 Mental retardation, X-linked 17/31, microduplication, 300705
HSPD1	14,8	60,90%	35,20%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HUWE1	47	97,10%	86,50%	Mental retardation, X-linked syndromic, Turner type, 300706
IDS	47,9	83,90%	75,00%	Mucopolysaccharidosis II, 309900
IDUA	79,9	98,20%	89,30%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015

IER3IP1	66,3	100,00%	99,80%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFT172	92,7	99,90%	96,60%	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IGBP1	43,6	83,50%	70,40%	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472
IGF1	135,8	100,00%	100,00%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IKBKG	13,4	24,90%	22,40%	Incontinentia pigmenti, type II, 308300 Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency, isolated, 300584 {Atypical mycobacteriosis, familial}, 300636 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IL1RAPL1	63,7	100,00%	97,90%	Mental retardation, X-linked 21/34, 300143
INPP5E	75,8	99,80%	96,60%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
IQSEC2	38	86,20%	65,40%	Mental retardation, X-linked 1, 309530
ISPD	80,8	96,90%	91,20%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
ITPR1	105,2	100,00%	98,30%	Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
JAG1	101,6	97,60%	95,70%	Alagille syndrome, 118450 Deafness, congenital heart defects and posterior embryotoxon Tetralogy of Fallot, 187500
JAM3	70,9	95,00%	90,70%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANK1	131,2	100,00%	99,90%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	74	93,20%	85,90%	Koolen-De Vries syndrome, 610443
KAT6A	120	99,70%	97,50%	Mental retardation, autosomal dominant 32, 616268
KAT6B	137	100,00%	100,00%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KCNH1	109	100,00%	99,20%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	158,3	100,00%	100,00%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791

KCNJ11	120,5	100,00%	100,00%	Hyperinsulinemic hypoglycemia, familial, 2, 601820 Diabetes, permanent neonatal, 606176 Diabetes mellitus, permanent neonatal, with neurologic features, 606176 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes mellitus, transient neonatal, 3, 610582
KCNK9	125	100,00%	100,00%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KCNQ2	69,5	100,00%	95,10%	Seizures, benign neonatal, 1, 121200 Myokymia, 121200 Epileptic encephalopathy, early infantile, 7, 613720
KCNT1	74,5	95,90%	92,20%	Epileptic encephalopathy, early infantile, 14, 614959 Epilepsy, nocturnal frontal lobe, 5, 615005
KCTD7	80,3	71,40%	68,80%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	56,9	99,00%	93,60%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6A	61,2	97,60%	90,00%	Kabuki syndrome 2, 300867
KIAA0226	90,4	100,00%	99,40%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
KIAA1033	106,1	99,50%	97,90%	?Mental retardation, autosomal recessive 43, 615817
KIAA1279	99,9	97,70%	95,40%	Goldberg-Shprintzen megacolon syndrome, 609460
KIAA2022	78	100,00%	99,10%	Mental retardation, X-linked 98, 300912
KIF11	94,6	100,00%	98,30%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF1A	68,2	98,40%	90,50%	Spastic paraplegia 30, autosomal recessive, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255
KIF5C	76,6	97,60%	94,00%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	68,8	89,20%	84,80%	Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
KIRREL3	75,2	99,10%	94,60%	Mental retardation, autosomal dominant 4, 612581
KMT2A	133,3	99,20%	98,50%	Leukemia, myeloid/lymphoid or mixed-lineage, 159555 Wiedemann-Steiner syndrome, 605130
KMT2D	97,3	99,30%	98,00%	Kabuki syndrome 1, 147920
KPTN	76,8	100,00%	97,40%	Mental retardation, autosomal recessive 41, 615637

KRAS	73,4	97,00%	91,20%	Noonan syndrome 3, 609942 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Lung cancer, somatic, 211980 Pancreatic carcinoma, somatic, 260350 SFM syndrome, somatic mosaic, 163200
KRBOX4	64,5	100,00%	96,60%	No OMIM phenotype nonsyndromic X-linked mental retardation (Lugtenberg et al. 2006)
L1CAM	61,8	99,00%	91,90%	Hydrocephalus due to aqueductal stenosis, 307000 MASA syndrome, 303350 CRASH syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100
L2HGDH	73,9	94,60%	92,30%	L-2-hydroxyglutaric aciduria, 236792
LAMA1	91,2	99,50%	95,70%	Poretti-Boltshauser syndrome, 615960
LAMA2	101,1	100,00%	99,20%	Muscular dystrophy, congenital merosin-deficient, 607855 Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855
LAMC3	90,8	99,20%	95,50%	Cortical malformations, occipital, 614115
LAMP2	54,9	92,70%	87,90%	Danon disease, 300257
LARGE	98,3	97,90%	90,70%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840
LARP7	95,7	100,00%	99,30%	Alazami syndrome, 615071
LIG4	188,1	100,00%	100,00%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500 Severe combined immunodeficiency with sensitivity to ionizing radiation, 602450
LINS	122,6	100,00%	100,00%	Mental retardation, autosomal recessive 27, 614340
LRP2	104,2	99,90%	98,80%	Donnai-Barrow syndrome, 222448
LRPPRC	96,2	98,70%	96,10%	Leigh syndrome, French-Canadian type, 220111
MAGEL2	121,2	100,00%	100,00%	Prader-Willi-like syndrome, 615547
MAGT1	54,7	98,70%	97,30%	Mental retardation, X-linked 95, 300716 Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853

MAN1B1	90,1	99,40%	97,60%	Mental retardation, autosomal recessive 15, 614202
MAN2B1	78,9	99,30%	91,40%	Mannosidosis, alpha-, types I and II, 248500
MANBA	87,5	99,80%	98,20%	Mannosidosis, beta, 248510
MAOA	52,2	98,90%	90,60%	Brunner syndrome, 300615
MAP2K1	92,2	93,80%	80,30%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	98,4	100,00%	96,30%	Cardiofaciocutaneous syndrome 4, 615280
MAT1A	86	99,50%	95,60%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MBD5	142,3	100,00%	99,80%	Mental retardation, autosomal dominant 1, 156200
MBTPS2	68,8	100,00%	98,70%	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800
MCCC1	90,5	99,20%	97,30%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	114,6	95,80%	90,80%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCOLN1	96,7	95,50%	89,60%	Mucolipidosis IV, 252650
MCPH1	121,4	100,00%	100,00%	Microcephaly 1, primary, autosomal recessive, 251200
MECP2	85,3	98,20%	87,00%	Rett syndrome, 312750 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, preserved speech variant, 312750 Encephalopathy, neonatal severe, 300673 {Autism susceptibility, X-linked 3}, 300496 Angelman syndrome, 105830 Mental retardation, X-linked syndromic, Lubs type, 300260
MED12	60,1	94,50%	86,40%	Opitz-Kaveggia syndrome, 305450 Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895
MED13L	114	100,00%	99,00%	Transposition of the great arteries, dextro-looped 1, 608808
MED17	140,1	99,80%	97,70%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	114,3	99,70%	99,40%	Mental retardation, autosomal recessive 18, 614249
MEF2C	108,6	100,00%	98,40%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 Chromosome 5q14.3 deletion syndrome, 613443
METTL23	162,5	100,00%	100,00%	Mental retardation, autosomal recessive 44, 615942
MGAT2	196,6	100,00%	100,00%	Congenital disorder of glycosylation, type IIa, 212066
MID1	70,6	98,60%	93,70%	Opitz GBBB syndrome, type I, 300000

MKKS	128,9	89,90%	89,90%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 209900
MLYCD	76,5	92,80%	84,60%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	176	100,00%	99,40%	Methylmalonic aciduria, vitamin B12-responsive, 251100
MMACHC	166	100,00%	100,00%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	65,3	89,70%	89,20%	Homocystinuria, cbID type, 277410
MOCS1	79,2	98,90%	93,20%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	117	99,60%	99,60%	Molybdenum cofactor deficiency, type B, 252150
MOGS	122,1	100,00%	100,00%	Congenital disorder of gycosylation, type 2b, 606056
MPDU1	111	100,00%	100,00%	Congenital disorder of glycosylation, type If, 609180
MPDZ	99,3	98,90%	96,80%	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219
MPLKIP	71,1	100,00%	100,00%	Trichothiodystrophy, nonphotosensitive 1, 234050
MRPS22	96,1	100,00%	100,00%	Combined oxidative phosphorylation deficiency 5, 611719
MTHFR	92	99,20%	97,10%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}
MTR	104,5	99,20%	97,90%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTRR	106,8	100,00%	99,90%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MUT	116,3	100,00%	100,00%	Methylmalonic aciduria, mut(0) type, 251000
MVK	92,2	100,00%	99,20%	Mevalonic aciduria, 610377 Hyper-IgD syndrome, 260920 Porokeratosis 3, disseminated superficial actinic, 175900
MYCN	100,4	98,80%	91,40%	Feingold syndrome, 164280
MYH9	85,7	99,20%	97,50%	Deafness, autosomal dominant 17, 603622 Epstein syndrome, 153650 Fechtner syndrome, 153640 Macrothrombocytopenia and progressive sensorineural deafness, 600208 May-Hegglin anomaly, 155100 Sebastian syndrome, 605249
MYO5A	89,9	99,00%	95,90%	Griscelli syndrome, type 1, 214450
MYT1L	106,6	100,00%	98,60%	No OMIM phenotype

NAA10	53,9	97,30%	92,30%	N-terminal acetyltransferase deficiency, 300855
NAGA	79,5	100,00%	99,30%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241
NAGLU	67,6	92,80%	91,50%	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NALCN	100,2	99,10%	96,10%	?Neuroaxonal neurodegeneration, infantile, with facial dysmophism, 615419
NBN	119,3	97,80%	95,60%	Aplastic anemia, 609135 Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260
NDE1	93,8	98,90%	97,90%	Lissencephaly 4 (with microcephaly), 614019
NDP	36,7	85,00%	63,70%	Norrie disease, 310600 Exudative vitreoretinopathy, X-linked, 305390
NDST1	103,2	100,00%	99,60%	Mental retardation, autosomal recessive 46, 616116
NDUFA1	109,9	100,00%	100,00%	Mitochondrial complex I deficiency, 252010
NDUFA11	102,5	94,90%	81,10%	Mitochondrial complex I deficiency, 252010
NDUFA12	86	100,00%	100,00%	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000
NDUFS1	80,8	100,00%	96,90%	Mitochondrial complex I deficiency, 252010
NDUFS2	122,6	100,00%	96,70%	Mitochondrial complex I deficiency, 252010
NDUFS3	134,8	92,00%	90,70%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	126,1	100,00%	100,00%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS7	99,7	100,00%	99,50%	Leigh syndrome, 256000
NDUFS8	105,4	100,00%	99,90%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
NDUFV1	62,5	99,80%	92,20%	Mitochondrial complex I deficiency, 252010
NEDD4L	112,9	100,00%	99,50%	No OMIM phenotype
NEU1	13,8	58,70%	26,40%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NF1	79,9	83,40%	80,00%	Neurofibromatosis, type 1, 162200 Leukemia, juvenile myelomonocytic, 607785 Melanoma, desmoplastic neurotrophic (2) Neurofibromatosis, familial spinal, 162210 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520

NFIA	110,1	100,00%	98,50%	No OMIM phenotype
NFIX	131	98,30%	96,50%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NHS	63,7	93,80%	85,70%	Nance-Horan syndrome, 302350 Cataract 40, X-linked, 302200
NIPBL	115,1	99,10%	98,70%	Cornelia de Lange syndrome 1, 122470
NKX2-1	86,5	100,00%	100,00%	Goiter, familial, due to TTF-1 defect (1) Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NLGN3	58,1	98,40%	93,10%	{Asperger syndrome susceptibility, X-linked 1}, 300494 {Autism susceptibility, X-linked 1}, 300425
NLGN4X	30,4	67,10%	53,70%	Mental retardation, X-linked, 300495 {Asperger syndrome susceptibility, X-linked 2}, 300497
NLRP3	112,6	99,90%	99,20%	Cold-induced autoinflammatory syndrome, familial, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115
NPHP1	118,9	100,00%	99,50%	hronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NR2F1	160	100,00%	98,40%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NRAS	125	100,00%	100,00%	Autoimmune lymphoproliferative syndrome type IV, 614470 Noonan syndrome 6, 613224 Epidermal nevus, somatic, 162900 Thyroid carcinoma, follicular, somatic, 188470 Colorectal cancer, somatic, 114500
NRXN1	114,6	99,10%	96,20%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSD1	117,6	100,00%	99,60%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 (1) Beckwith-Wiedemann syndrome, 130650
NSDHL	45,5	99,50%	97,50%	CHILD syndrome, 308050 CK syndrome, 300831
NSUN2	119,6	99,60%	92,70%	Mental retardation, autosomal recessive 5, 611091

NTRK1	66,4	99,30%	88,80%	Insensitivity to pain, congenital, with anhidrosis, 256800 Medullary thyroid carcinoma, familial, 155240
OCLN	99,3	72,60%	72,60%	Band-like calcification with simplified gyration and polymicrogyria, 251290
OCRL	59,7	98,60%	96,10%	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	36,6	89,40%	76,60%	Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804
OPHN1	48,3	97,20%	85,60%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
ORC1	105,1	100,00%	97,40%	Meier-Gorlin syndrome 1, 224690
OTC	52,3	100,00%	92,10%	CGD Ornithine transcarbamylase deficiency, 311250
PACS1	98,8	99,00%	96,80%	Mental retardation, autosomal dominant 17, 615009
PAFAH1B1	69,1	85,80%	77,50%	Lissencephaly, 607432 Subcortical laminar heterotopia, 607432
PAH	76,3	96,80%	95,40%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAK3	52,4	100,00%	99,20%	Mental retardation, X-linked 30/47, 300558
PANK2	106,5	100,00%	94,00%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PAX1	81,9	87,80%	82,50%	?Orofaciocervical syndrome 2, 615560
PAX6	85,7	100,00%	100,00%	Aniridia, 106210 Peters anomaly, 604229 Cataract with late-onset corneal dystrophy, 106210 Keratitis, 148190 Foveal hyperplasia, 136520 Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550 Coloboma, ocular, 120200 Coloboma of optic nerve, 120430 Gillespie syndrome, 206700
PAX8	61,7	100,00%	86,20%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PC	97,1	94,60%	91,90%	Pyruvate carboxylase deficiency, 266150
PCDH19	71,1	100,00%	98,20%	Epileptic encephalopathy, early infantile, 9, 300088
PCNT	95,1	99,00%	92,20%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720

PDE4D	104,5	97,20%	90,60%	Acrocydostosis 2 with or without hormone resistance, 614613 {Stroke, susceptibility to, 1}, 606799
PDHA1	61,1	100,00%	88,80%	Pyruvate dehydrogenase E1-alpha deficiency, 312170 Leigh syndrome, X-linked, 308930
PDSS1	86,7	89,50%	87,10%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	92,8	100,00%	100,00%	Coenzyme Q10 deficiency, primary, 3, 614652
PEPD	68	95,40%	91,20%	Prolidase deficiency, 170100
PEX1	114,8	100,00%	100,00%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	65,6	93,20%	87,60%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	144,7	100,00%	100,00%	Peroxisome biogenesis disorder 14B, 614920
PEX12	125	100,00%	100,00%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	137,8	96,60%	94,40%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX16	87,3	97,10%	86,50%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	107,2	100,00%	100,00%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	142,7	100,00%	100,00%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	103,9	100,00%	100,00%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	133,5	100,00%	100,00%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	84,6	98,40%	96,60%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370
PEX6	87,8	96,20%	84,50%	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PEX7	96,3	89,10%	83,60%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PGAP1	114,3	100,00%	99,30%	?Mental retardation, autosomal recessive 42, 615802
PGAP2	111	98,50%	97,20%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	65,1	98,80%	86,80%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	41,3	76,20%	70,50%	Phosphoglycerate kinase 1 deficiency, 300653

PHF6	64,1	98,80%	95,70%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	51,1	98,80%	87,70%	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	83,7	100,00%	98,60%	Phosphoglycerate dehydrogenase deficiency, 601815
PIGL	90,9	100,00%	100,00%	CHIME syndrome, 280000
PIGN	96,3	100,00%	100,00%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	107,9	100,00%	99,80%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	129,9	100,00%	99,80%	?Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PIGV	169,8	100,00%	100,00%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3R2	81	88,70%	84,10%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387
PLA2G6	75,5	98,40%	90,50%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953
PLCB1	110,1	100,00%	94,00%	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	42,1	94,10%	71,80%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PMM2	92,6	100,00%	100,00%	Congenital disorder of glycosylation, type Ia, 212065
PNKP	68,3	100,00%	98,60%	Epileptic encephalopathy, early infantile, 10, 613402
PNP	117,7	100,00%	98,80%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
POC1A	96	98,50%	96,00%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POGZ	120,1	99,90%	98,40%	Autism (Neale (2012) Nature 485, 242) Intellectual disability (Gilissen (2014) Nature 511, 344) Schizophrenia (Fromer (2014) Nature 506, 179)
POLG	83,7	98,80%	94,60%	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome, 607459 Progressive external ophthalmoplegia, autosomal dominant, 157640 Progressive external ophthalmoplegia, autosomal recessive, 258450
POLR3A	85	99,60%	94,80%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	106,4	100,00%	98,10%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381

POMGNT1	93,6	100,00%	99,30%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157
POMT1	97,7	100,00%	98,70%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308
POMT2	73,7	97,60%	88,30%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158
PORCN	53,4	89,80%	79,40%	Focal dermal hypoplasia, 305600
POU1F1	115,5	100,00%	100,00%	Pituitary hormone deficiency, combined, 1, 613038
PPOX	93,7	98,90%	96,30%	Porphyria variegata, 176200
PPP2R1A	83,1	93,10%	88,60%	Mental retardation, autosomal dominant 36, 616362
PPP2R5D	125,4	98,60%	97,00%	Mental retardation, autosomal dominant 35, 616355
PPT1	66,8	100,00%	95,90%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	65,2	100,00%	93,90%	Renpenning syndrome, 309500
PRODH	46,6	83,60%	62,40%	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PRPS1	63,7	100,00%	95,70%	Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Arts syndrome, 301835 Deafness, X-linked 1, 304500
PRSS12	99,2	100,00%	97,00%	Mental retardation, autosomal recessive 1, 249500
PSAP	80	100,00%	97,00%	Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539 Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722
PSEN1	96,9	99,40%	94,40%	Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Cardiomyopathy, dilated, 1U, 613694 Dementia, frontotemporal, 600274 Pick disease, 172700

PTCH1	83,8	99,40%	94,70%	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828
PTCHD1	87,1	100,00%	99,20%	No OMIM phenotype
PTDSS1	120,5	100,00%	100,00%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	121,7	100,00%	97,60%	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Bannayan-Riley-Ruvalcaba syndrome, 153480 {Meningioma}, 607174 {Glioma susceptibility 2}, 613028 Macrocephaly/autism syndrome, 605309 PTEN hamartoma tumor syndrome VATER association with macrocephaly and ventriculomegaly, 276950 {Prostate cancer, somatic}, 176807 Thyroid carcinoma, follicular, somatic, 188470 Malignant melanoma, somatic, 155600 Endometrial carcinoma, somatic, 608089 Squamous cell carcinoma, head and neck, somatic, 275355
PTPN11	46,8	87,30%	67,80%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, 607785 Metachondromatosis, 156250
PUF60	122,2	98,30%	93,50%	Verheij syndrome, 615583
PURA	109,3	99,10%	89,00%	Mental retardation, autosomal dominant 31, 616158
PUS1	65,2	99,80%	96,40%	Mitochondrial myopathy and sideroblastic anemia 1, 600462
PYCR1	84,5	100,00%	99,40%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
RAB18	122,7	100,00%	100,00%	Warburg micro syndrome 3, 614222
RAB27A	110	100,00%	100,00%	Griscelli syndrome, type 2, 607624
RAB39B	73,7	100,00%	98,80%	Mental retardation, X-linked 72, 300271
RAB3GAP1	120,8	98,50%	97,30%	Warburg micro syndrome 1, 600118
RAB3GAP2	108,1	100,00%	98,40%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAB40AL	13,3	59,10%	39,80%	Mental retardation, X-linked, syndromic, Martin-Probst type, 300519

RAD21	88,8	98,40%	90,10%	Cornelia de Lange syndrome 4, 614701
RAF1	89,9	100,00%	98,90%	Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RAI1	127,8	99,80%	99,70%	Immunodeficiency 9, 612782 Smith-Magenis syndrome, 182290
RARS2	83,6	99,80%	98,20%	Pontocerebellar hypoplasia, type 6, 611523
RBM10	52,2	95,70%	84,10%	TARP syndrome, 311900
RBM28	94,7	100,00%	97,10%	Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RELN	104,8	99,20%	97,10%	Lissencephaly 2 (Norman-Roberts type), 257320
RFT1	67,5	99,60%	96,20%	Congenital disorder of glycosylation, type In, 612015
RIT1	150,2	100,00%	100,00%	Noonan syndrome 8, 615355
RMND1	78	92,40%	87,70%	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH2A	90,9	100,00%	99,90%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	108,4	100,00%	99,00%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	134,2	100,00%	100,00%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	91,1	100,00%	97,40%	Leukoencephalopathy, cystic, without megalencephaly, 612951
ROGDI	100,3	95,20%	95,10%	Kohlschutter-Tonz syndrome, 226750
RPGRIP1L	99,8	96,70%	96,70%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360
RPL10	42,1	99,20%	76,60%	{Autism, susceptibility to, X-linked 5}, 300847
RPS6KA3	48,3	99,30%	95,20%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RTEL1	76,5	98,40%	88,50%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
SALL1	133,7	99,10%	98,10%	Townes-Brocks syndrome, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SATB2	109	100,00%	97,20%	Cleft palate and mental retardation, 119540
SC5D	155,6	100,00%	100,00%	Lathosterolosis, 607330
SCN1A	116,9	99,20%	97,40%	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Dravet syndrome, 607208 Migraine, familial hemiplegic, 3, 609634 Febrile seizures, familial, 3A, 604403

SCN2A	123,1	100,00%	99,80%	Seizures, benign familial infantile, 3, 607745 Epileptic encephalopathy, early infantile, 11, 613721
SCN8A	136,5	100,00%	99,10%	Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558
SCO2	78,6	100,00%	100,00%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 Myopia 6, 608908
SDHA	10,9	29,90%	17,50%	Leigh syndrome, 256000 Mitochondrial respiratory chain complex II deficiency, 252011 Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165
SERAC1	86,2	100,00%	100,00%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETBP1	138,1	97,20%	95,50%	Schinzel-Giedion midface retraction syndrome, 269150
SETD5	145,3	99,70%	98,20%	No OMIM phenotype Autism (Neale (2012) Nature 485, 242) Intellectual disability (Grozeva (2014) Am J Hum Genet 94, 618)
SGSH	74,8	94,30%	94,30%	Mucopolysaccharidisis type 3A (Sanfilippo A), 252900
SHANK2	93,7	99,50%	96,70%	{Autism susceptibility 17}, 613436
SHANK3	64,8	91,10%	79,40%	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SHH	97,5	98,70%	90,40%	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160
SHOC2	112,8	100,00%	98,40%	Noonan-like syndrome with loose anagen hair, 607721
SHROOM4	60,6	99,30%	93,20%	Stocco dos Santos X-linked mental retardation syndrome, 300434
SIL1	95,6	100,00%	99,80%	Marinesco-Sjogren syndrome, 248800
SIX3	114,9	100,00%	98,80%	Holoprosencephaly-2, 157170 Schizensephaly, 269160
SKI	60,8	90,40%	80,20%	Shprintzen-Goldberg syndrome, 182212
SLC12A6	90,6	100,00%	99,50%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC16A2	44,7	94,60%	77,60%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	93,2	100,00%	99,20%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920

SLC1A1	112	100,00%	99,90%	Dicarboxylic aminoaciduria, 222730 {?Schizophrenia susceptibility 18}, 615232
SLC25A15	99,4	86,40%	85,30%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	68	100,00%	92,30%	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	90	100,00%	100,00%	GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 {Epilepsy, idiopathic generalized, suscpetibility to, 12}, 614847 Dystonia 9, 601042
SLC33A1	90,9	100,00%	100,00%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC35A2	44,6	100,00%	95,90%	Congenital disorder of glycosylation, type 2m, 300896
SLC35C1	97,9	100,00%	100,00%	Congenital disorder of glycosylation, type IIc, 266265
SLC4A4	107,3	100,00%	100,00%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A17	84,6	97,10%	87,60%	Mental retardation, autosomal recessive 48, 616269
SLC6A3	73,9	100,00%	99,90%	Parkinsonism -dystonia, infantile, 613135 {Nicotine dependence, protection against}, 188890
SLC6A8	5,1	11,70%	9,70%	Cerebral creatine deficiency syndrome 1, 300352
SLC7A7	88	100,00%	100,00%	Lysinuric protein intolerance, 222700
SLC9A6	61,7	95,90%	87,60%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMAD4	126,2	100,00%	93,60%	Pancreatic cancer Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210
SMARCA2	87,6	97,70%	94,80%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	81,5	97,90%	91,40%	Rhabdoid tumor predisposition syndrome 2, 613325 Mental retardation, autosomal dominant 16, 614609
SMARCB1	118,9	100,00%	100,00%	Rhabdoid tumors, somatic, 609322 Rhabdoid predisposition syndrome 1, 609322 Mental retardation, autosomal dominant 15, 614608
SMARCE1	66,6	80,20%	66,20%	{Meningioma, familial, susceptibility to}, 607174
SMC1A	66,9	95,70%	90,00%	Cornelia de Lange syndrome 2, 300590
SMC3	108,5	99,60%	96,30%	Cornelia de Lange syndrome 3, 610759
SMOC1	78,6	99,90%	91,60%	Microphthalmia with limb anomalies, 206920

SMPD1	100,5	96,90%	90,40%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMS	15,2	56,20%	26,20%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAP29	116,7	100,00%	100,00%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNIP1	146	100,00%	97,70%	Psychomotor retardation, epilepsy and craniofacial dysmorphism, 614501
SOBP	122,8	100,00%	92,30%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOS1	118,3	100,00%	99,70%	Fibromatosis, gingival, 135300 Noonan syndrome 4, 610733
SOX10	65,1	100,00%	97,40%	Waardenburg syndrome, type 4C, 613266 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136
SOX11	98,4	100,00%	100,00%	Mental retardation, autosomal dominant, 27, 615866
SOX2	130,7	100,00%	100,00%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	36,6	95,60%	86,90%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SPG11	105,9	100,00%	98,10%	Spastic paraplegia 11, autosomal recessive, 604360
SPRED1	117,8	100,00%	100,00%	Legius syndrome, 611431
SPTAN1	92,8	100,00%	97,90%	Epileptic encephalopathy, early infantile, 5
SRCAP	129,4	100,00%	99,10%	Floating-Harbor syndrome, 136140
SRD5A3	116,3	100,00%	100,00%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
SRPX2	41,9	95,40%	80,20%	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
ST3GAL3	109,3	100,00%	100,00%	Mental retardation, autosomal recessive 12, 611090 Epileptic encephalopathy, early infantile, 15, 615006
ST3GAL5	112,3	94,90%	94,50%	Amish infantile epilepsy syndrome, 609056
STIL	148,1	100,00%	100,00%	Microcephaly 7, primary, autosomal recessive, 612703
STRA6	71,8	100,00%	96,90%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STT3A	115,1	100,00%	98,80%	?Congenital disorder of glycosylation, type Iw, 615596
STT3B	106,8	99,00%	98,20%	?Congenital disorder of glycosylation, type Ix, 615597
STXBP1	88,6	100,00%	97,90%	Epileptic encephalopathy, early infantile, 4, 612164
SUCLA2	74,5	94,90%	87,80%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073

SUOX	163,4	100,00%	100,00%	Sulfite oxidase deficiency, 272300
SURF1	82,5	88,30%	88,30%	Leigh syndrome, due to COX deficiency, 256000
SYN1	31,6	84,40%	48,10%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNE1	105,7	99,20%	97,40%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNGAP1	50,3	91,20%	77,80%	Mental retardation, autosomal dominant 5, 612621
SYP	46	98,50%	81,30%	Mental retardation, X-linked 96, 300802
SYT14	116,7	93,50%	92,00%	Spinocerebellar ataxia, autosomal recessive 11, 614229
TAF2	108,6	100,00%	100,00%	Mental retardation, autosomal recessive 40, 615599
TAT	91,6	100,00%	100,00%	Tyrosinemia, type II, 276600
TBC1D24	101,4	100,00%	98,80%	Myoclonic epilepsy, infantile, familial, 605021 Epileptic encephalopathy, early infantile, 16, 615338
TBC1D7	121,7	100,00%	100,00%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCE	112,5	100,00%	97,80%	Kenny-Caffey syndrome-1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410
TBR1	85,4	100,00%	94,50%	No OMIM phenotype
TCF4	96	97,50%	97,40%	Pitt-Hopkins syndrome, 610954
TECR	79,3	99,20%	95,00%	Mental retardation, autosomal recessive 14, 614020
TEX28	0,1	0,00%	0,00%	No OMIM phenotype
TFAP2A	68,3	98,30%	87,40%	Branchiooculofacial syndrome, 113620
TGFBR1	126,6	93,70%	93,60%	Loeys-Dietz syndrome, type 1A, 609192 Loeys-Dietz syndrome, type 2A, 608967 {Multiple self-healing squamous epithelioma, susceptiblity to}, 132800
TGFBR2	86,3	100,00%	97,70%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome, type 1B, 610168 Loeys-Dietz syndrome, type 2B, 610380
TGIF1	161,9	100,00%	100,00%	Holoprosencephaly-4, 142946
THOC6	151,2	100,00%	98,50%	Beaulieu-Boycott-Innes syndrome, 613680
THRB	114,5	100,00%	99,90%	Thyroid hormone resistance, 188570 Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, selective pituitary, 145650

TIMM8A	31	90,80%	73,00%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TMCO1	80,2	100,00%	100,00%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 614132
TMEM165	89,4	100,00%	100,00%	Congenital disorder of glycosylation, type IIk, 614727
TMEM231	59,6	97,00%	87,60%	Joubert syndrome 20, 614970 Meckel syndrome, type 11, 615397
TMEM237	87,8	100,00%	92,50%	Joubert syndrome 14, 614424
TMEM67	115,7	100,00%	99,80%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TMLHE	33,1	70,50%	67,90%	Epsilon-trimethyllysine hydrolylase deficiency, 300872
TPP1	123,4	100,00%	100,00%	Ceroid lipofuscinosis, neuronal, 2, 204500
TRAPPC11	110,3	100,00%	100,00%	Muscular dystrophy, limb-girdle, type 2S
TRAPPC9	67,5	96,30%	90,40%	Mental retardation, autosomal recessive 13, 613192
TREX1	129,5	100,00%	100,00%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRIM32	106,2	100,00%	100,00%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110
TRMT10A	129,5	100,00%	100,00%	Microcephaly, short stature and impaired glucose metabolism, 616033
TSC1	86,3	99,10%	96,30%	Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690 Focal cortical dysplasia, Taylor balloon cell type, 607341
TSC2	80,3	99,20%	95,00%	Tuberous sclerosis-2, 613254 Lymphangioleiomyomatosis, somatic, 606690
TSEN54	102,6	96,50%	93,40%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TSPAN7	49,5	98,60%	78,40%	Mental retardation, X-linked 58, 300210
TTC8	103,9	100,00%	100,00%	Bardet-Biedl syndrome 8, 209900 Retinitis pigmentosa 51, 613464

TTI2	97,8	100,00%	98,70%	Mental retardation, autosomal recessive 39, 615541
TUBA1A	24,2	95,50%	51,10%	Lissencephaly 3, 611603
TUBA8	84,8	99,00%	98,10%	Polymicrogyria with optic nerve hypoplasia, 613180
TUBB2B	48	100,00%	88,60%	Polymicrogyria, symmetric or asymmetric, 610031
TUBGCP6	115,4	99,60%	97,40%	Microcephaly and chorioretinopathy, autosomal recessive 1, 251270
TUSC3	118,8	100,00%	98,60%	Mental retardation, autosomal recessive 7, 611093
TWIST1	120,3	100,00%	99,90%	Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400
UBE2A	69,7	100,00%	100,00%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBE3A	99,7	100,00%	100,00%	Angelman syndrome, 105830
UBE3B	97,5	95,40%	92,60%	Blepharophimosis-ptosis-intellectual disability syndrome, 615057
UBR1	100,3	100,00%	100,00%	Johanson-Blizzard syndrome, 243800
UPB1	134,8	100,00%	99,00%	Beta-ureidopropionase deficiency, 613161
UPF3B	56,2	99,30%	90,10%	Mental retardation, X-linked, syndromic 14, 300676
USP9X	66,1	99,50%	95,40%	Mental retardation, X-linked 99, 300919
VLDLR	109,1	100,00%	99,40%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13B	108	99,10%	98,30%	Cohen syndrome, 216550
VRK1	136,9	100,00%	100,00%	Pontocerebellar hypoplasia type 1A, 607596
WDR19	122,7	100,00%	100,00%	Nephronophtisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR45	38,3	91,20%	85,60%	Neurodegeneration with brain iron acculation 5, 300894
WDR62	99,9	98,80%	92,60%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR81	105,4	99,60%	98,00%	Cerebellar ataxia, mental retardation and dysequilibrium syndrome 2, 610185
WWOX	98,3	97,90%	97,50%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive, 12, 614322
XPA	81,5	100,00%	90,20%	Xeroderma pigmentosum, group A, 278700
XPNPEP3	115,8	97,30%	94,90%	Nephronophthisis-like nephropathy 1, 613159
XYLT1	107,3	96,40%	87,10%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800

YAP1	68	95,80%	86,60%	Coloboma, ocular with or without hearing impairment, cleft lip/palate and mental retardation, 120433
YWHAE	37,6	77,40%	60,60%	No OMIM phenotype
ZBTB16	112,7	100,00%	99,00%	Leukemia, acute promyelocytic, PL2F/RARA type Skeletal defects, genital hypoplasia, and mental retardation, 612447
ZBTB18	147,8	100,00%	98,40%	?Mental retardation, autosomal dominant 22, 612337
ZDHHC15	52,1	100,00%	97,40%	?Mental retardation, X-linked 91, 300577
ZDHHC9	43,5	98,40%	88,90%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB2	153,3	100,00%	100,00%	Mowat-Wilson syndrome, 235730
ZFYVE26	86,9	97,50%	93,20%	Spastic paraplegia 15, autosomal recessive, 270700
ZIC2	60	94,10%	85,20%	Holoprosencephaly-5, 609637
ZMYND11	118,7	100,00%	100,00%	Mental retardation, autosomal dominant 30, 616083
ZNF41	61,7	100,00%	96,30%	Mental retardation, X-linked 89, 300848
ZNF592	103,2	93,40%	90,50%	Spinocerebellar ataxia, autosomal recessive 5, 606937
ZNF674	60,9	97,60%	88,40%	Mental retardation, X-linked 92, 300851
ZNF711	65,5	100,00%	99,20%	Mental retardation, X-linked 97, 300803
ZNF81	50,9	97,90%	97,30%	Mental retardation, X-linked 45, 300498

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
Median Coverage describes the average number of reads seen across 50 exomes
% 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
OMIM release used for OMIM disease identifiers and descriptions : June 30th, 2015
This list is accurate for all panel versions starting with DG 2.3. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors