# INTELLECTUAL DISABILITY GENE PANEL

| Gene symbol | Depth (reads) | Coverage (avg %) | OMIM disease | Description |
|---|---|---|---|---|
| ABCC9 | 115 | 96 | 614050 | Atrial fibrillation familial 12 |
| ABCD1 | 59 | 84 | 300100 | Adrenoleukodystrophy |
| ABCD4 | 121 | 88 | 614857 | Methylmalonic aciduria and homocystinuria cblJ type |
| ABHD5 | 125 | 91 | 275630 | Chanarin-Dorfman syndrome |
| ACAD9 | 105 | 89 | 611126 | ACAD9 deficiency |
| ACO2 | 76 | 67 | 614559 | Infantile cerebellar-retinal degeneration |
| ACOX1 | 93 | 87 | 264470 | Peroxisomal acyl-CoA oxidase deficiency |
| ACSF3 | 79 | 89 | 614265 | Combined malonic and methylmalonic aciduria |
| ACSL4 | 90 | 96 | 300387 | Mental retardation X-linked 63 |
| ACTB | 101 | 17 | 243310 | Baraitser-Winter syndrome 1 |
| ACTG1 | 113 | 16 | 614583 | Baraitser-Winter syndrome 2 |
| ACVR1 | 115 | 84 | 135100 | Fibrodysplasia ossificans progressiva |
| ADAR | 147 | 93 | 615010 | Aicardi-Goutieres syndrome 6 |
| ADCK3 | 97 | 87 | 200 | - |
| ADCK3 | 97 | 87 | 200 | - |
| ADSL | 124 | 83 | 103050 | Adenylosuccinase deficiency |
| AFF2 | 120 | 92 | 309548 | Mental retardation X-linked FRAXE type |
| AGA | 121 | 94 | 208400 | Aspartylglucosaminuria |
| AGPAT2 | 57 | 98 | 608594 | Lipodystrophy congenital generalized type 1 |
| AGTR2 | 143 | 100 | 300852 | Mental retardation X-linked 88 |
| AHCY | 93 | 77 | 613752 | Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase |
| AHI1 | 117 | 97 | 608629 | Joubert syndrome-3 |
| AIFM1 | 106 | 79 | 300816 | Combined oxidative phosphorylation deficiency 6 |
| AIMP1 | 123 | 84 | 260600 | Leukodystrophy hypomyelinating 3 |
| AK1 | 84 | 91 | 612631 | Hemolytic anemia due to adenylate kinase deficiency |
| AKT3 | 99 | 95 | 603387 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome |
| ALDH18A1 | 103 | 82 | 219150 | Cutis laxa autosomal recessive type IIIA |
| ALDH3A2 | 113 | 41 | 270200 | Sjogren-Larsson syndrome |

| | | | | |
|---|---|---|---|---|
| ALDH5A1 | 72 | 88 | 271980 | Succinic semialdehyde dehydrogenase deficiency |
| ALG1 | 104 | 87 | 608540 | Congenital disorder of glycosylation type Ik |
| ALG12 | 105 | 88 | 607143 | Congenital disorder of glycosylation type Ig |
| ALG2 | 109 | 97 | 607906 | Congenital disorder of glycosylation type Ii |
| ALG3 | 118 | 87 | 601110 | Congenital disorder of glycosylation type Id |
| ALG6 | 87 | 100 | 603147 | Congenital disorder of glycosylation type Ic |
| ALG9 | 107 | 87 | 608776 | Congenital disorder of glycosylation type Il |
| AMT | 135 | 94 | 605899 | Glycine encephalopathy |
| ANKH | 117 | 88 | 118600 | Chondrocalcinosis 2 |
| ANKRD11 | 80 | 48 | 148050 | KBG syndrome |
| ANO10 | 118 | 94 | 613728 | Spinocerebellar ataxia autosomal recessive 10 |
| AP1S2 | 68 | 66 | 300630 | Mental retardation X-linked syndromic Fried type |
| AP3B1 | 109 | 95 | 608233 | Hermansky-Pudlak syndrome 2 |
| AP4B1 | 106 | 96 | 614066 | Spastic paraplegia 47 autosomal recessive |
| AP4E1 | 130 | 95 | 613744 | Spastic paraplegia 51 autosomal recessive |
| AP4S1 | 93 | 94 | 614067 | Spastic paraplegia 52 autosomal recessive |
| APTX | 143 | 96 | 208920 | Ataxia early-onset with oculomotor apraxia and hypoalbuminemia |
| ARFGEF2 | 127 | 87 | 608097 | Periventricular heterotopia with microcephaly |
| ARHGEF6 | 88 | 94 | 300436 | Mental retardation X-linked 46 |
| ARHGEF9 | 91 | 80 | 300607 | Epileptic encephalopathy early infantile 8 |
| ARID1A | 112 | 91 | 614607 | Mental retardation autosomal dominant 14 |
| ARID1B | 118 | 91 | 614562 | Mental retardation autosomal dominant 12 |
| ARL13B | 113 | 94 | 612291 | Joubert syndrome 8 |
| ARL6 | 142 | 99 | 209900 | Bardet-Biedl syndrome 3 |
| ARX | 37 | 94 | 308350 | Epileptic encephalopathy early infantile 1 |
| ASL | 79 | 88 | 207900 | Argininosuccinic aciduria |
| ASPA | 118 | 96 | 271900 | Canavan disease |
| ASPM | 129 | 98 | 608716 | Microcephaly 5 primary autosomal recessive |
| ASXL1 | 157 | 95 | 605039 | Bohring-Opitz syndrome |
| ATP1A2 | 106 | 82 | 104290 | Alternating hemiplegia of childhood |
| ATP2A2 | 120 | 93 | 101900 | Acrokeratosis verruciformis |
| ATP6AP2 | 58 | 69 | 300423 | Mental retardation X-linked with epilepsy |
| ATP6V0A2 | 110 | 93 | 219200 | Cutis laxa autosomal recessive type IIA |

| | | | | |
|---|---|---|---|---|
| ATP7A | 101 | 96 | 309400 | Menkes disease |
| ATR | 118 | 94 | 614564 | Cutaneous telangiectasia and cancer syndrome familial |
| ATRX | 110 | 97 | 300448 | Alpha-thalassemia myelodysplasia syndrome somatic |
| AUH | 111 | 87 | 250950 | 3-methylglutaconic aciduria type I |
| B3GALTL | 109 | 92 | 261540 | Peters-plus syndrome |
| B4GALT1 | 86 | 95 | 607091 | Congenital disorder of glycosylation type IId |
| B4GALT7 | 74 | 95 | 130070 | Ehlers-Danlos syndrome progeroid type 1 |
| BBS1 | 126 | 89 | 209900 | Bardet-Biedl syndrome 1 |
| BBS10 | 128 | 100 | 209900 | Bardet-Biedl syndrome 10 |
| BBS12 | 163 | 100 | 209900 | Bardet-Biedl syndrome 12 |
| BBS2 | 130 | 91 | 209900 | Bardet-Biedl syndrome 2 |
| BBS4 | 104 | 87 | 209900 | Bardet-Biedl syndrome 4 |
| BBS5 | 114 | 96 | 209900 | Bardet-Biedl syndrome 5 |
| BBS7 | 121 | 98 | 209900 | Bardet-Biedl syndrome 7 |
| BBS9 | 117 | 95 | 209900 | Bardet-Biedl syndrome 9 |
| BCKDHA | 109 | 92 | 248600 | Maple syrup urine disease type Ia |
| BCKDHB | 89 | 98 | 248600 | Maple syrup urine disease type Ib |
| BCOR | 97 | 95 | 300166 | Microphthalmia syndromic 2 |
| BCS1L | 133 | 86 | 262000 | Bjornstad syndrome |
| BIVM-ERCC5 | 129 | 95 | 200 | - |
| BLM | 128 | 89 | 210900 | Bloom syndrome |
| BRAF | 82 | 81 | 211980 | Adenocarcinoma of lung somatic |
| BRWD3 | 90 | 98 | 300659 | Mental retardation X-linked 93 |
| BSCL2 | 119 | 85 | 269700 | Lipodystrophy congenital generalized type 2 |
| BUB1B | 127 | 94 | 114500 | Colorectal cancer somatic |
| C5orf42 | 128 | 96 | 614615 | Joubert syndrome 17 |
| CA2 | 157 | 87 | 259730 | Osteopetrosis autosomal recessive 3 with renal tubular acidosis |
| CACNA1C | 111 | 84 | 611875 | Brugada syndrome 3 |
| CACNA1C | 111 | 84 | 611875 | Brugada syndrome 3 |
| CACNA1C | 111 | 84 | 611875 | Brugada syndrome 3 |
| CACNG2 | 89 | 100 | 614256 | Mental retardation autosomal dominant 10 |
| CASK | 78 | 93 | 300422 | FG syndrome 4 |

| | | | | |
|---|---|---|---|---|
| CBS | 77 | 86 | 236200 | Homocystinuria B6-responsive and nonresponsive types |
| CC2D1A | 100 | 90 | 608443 | Mental retardation autosomal recessive 3 |
| CC2D2A | 101 | 90 | 216360 | COACH syndrome |
| CCBE1 | 97 | 91 | 235510 | Hennekam lymphangiectasia-lymphedema syndrome |
| CCDC78 | 84 | 95 | 614807 | Myopathy centronuclear 4 |
| CDH15 | 75 | 89 | 612580 | Mental retardation autosomal dominant 3 |
| CDK5RAP2 | 122 | 88 | 604804 | Microcephaly 3 primary autosomal recessive |
| CDKL5 | 118 | 92 | 105830 | Angelman syndrome-like |
| CDON | 132 | 83 | 614226 | Holoprosencephaly 11 |
| CENPJ | 131 | 96 | 608393 | Microcephaly 6 primary autosomal recessive |
| CEP135 | 116 | 93 | 614673 | Microcephaly 8 primary autosomal recessive |
| CEP152 | 138 | 93 | 614852 | Microcephaly 9 primary autosomal recessive |
| CEP290 | 96 | 92 | 209900 | Bardet-Biedl syndrome 14 |
| CEP41 | 88 | 93 | 614464 | Joubert syndrome 15 |
| CHD7 | 129 | 92 | 214800 | CHARGE syndrome |
| CHKB-CPT1B | 98 | 82 | 200 | - |
| CNTNAP2 | 115 | 86 | 610042 | Cortical dysplasia-focal epilepsy syndrome |
| COG1 | 125 | 90 | 611209 | Congenital disorder of glycosylation type IIg |
| COG7 | 99 | 76 | 608779 | Congenital disorder of glycosylation type IIe |
| COG8 | 119 | 79 | 611182 | Congenital disorder of glycosylation type IIh |
| COL4A1 | 109 | 77 | 611773 | Angiopathy hereditary with nephropathy aneurysms and muscle |
| COL4A2 | 92 | 84 | 614483 | Porencephaly 2 |
| COLEC11 | 109 | 97 | 265050 | 3MC syndrome 2 |
| COQ2 | 79 | 93 | 607426 | Coenzyme Q10 deficiency primary 1 |
| COX15 | 90 | 89 | 615119 | Cardioencephalomyopathy fatal infantile due to cytochrome c oxidase deficiency 2 |
| CRBN | 135 | 98 | 607417 | Mental retardation autosomal recessive 2 |
| CREBBP | 90 | 92 | 180849 | Rubinstein-Taybi syndrome |
| CTDP1 | 80 | 88 | 604168 | Congenital cataracts facial dysmorphism and neuropathy |
| CTNNB1 | 140 | 95 | 114500 | Colorectal cancer somatic |
| CUL4B | 89 | 95 | 300354 | Mental retardation X-linked syndromic 15 (Cabezas type) |
| CYB5R3 | 81 | 92 | 250800 | Methemoglobinemia type I |
| D2HGDH | 58 | 84 | 600721 | D-2-hydroxyglutaric aciduria |

| | | | | |
|---|---|---|---|---|
| DARS2 | 112 | 97 | 611105 | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation |
| DBT | 100 | 93 | 248600 | Maple syrup urine disease type II |
| DCX | 109 | 94 | 300067 | Lissencephaly X-linked |
| DDHD2 | 112 | 91 | 615033 | Spastic paraplegia 54 autosomal recessive |
| DHCR24 | 95 | 79 | 602398 | Desmosterolosis |
| DHCR7 | 118 | 88 | 270400 | Smith-Lemli-Opitz syndrome |
| DHFR | 57 | 32 | 613839 | Megaloblastic anemia due to dihydrofolate reductase deficiency |
| DHTKD1 | 114 | 86 | 204750 | 2-aminoadipic 2-oxoadipic aciduria |
| DIP2B | 110 | 87 | 136630 | Mental retardation FRA12A type |
| DKC1 | 83 | 94 | 305000 | Dyskeratosis congenita X-linked |
| DLD | 141 | 94 | 246900 | Dihydrolipoamide dehydrogenase deficiency |
| DLG3 | 64 | 87 | 300850 | Mental retardation X-linked 90 |
| DMD | 91 | 91 | 300376 | Becker muscular dystrophy |
| DMPK | 102 | 91 | 160900 | Myotonic dystrophy 1 |
| DNAJC19 | 101 | 83 | 610198 | 3-methylglutaconic aciduria type V |
| DNMT3B | 94 | 83 | 242860 | Immunodeficiency-centromeric instability-facial anomalies syndrome 1 |
| DOCK8 | 97 | 84 | 243700 | Hyper-IgE recurrent infection syndrome autosomal recessive |
| DPAGT1 | 115 | 89 | 608093 | Congenital disorder of glycosylation type Ij |
| DPM1 | 145 | 100 | 608799 | Congenital disorder of glycosylation type Ie |
| DPYD | 122 | 88 | 274270 | 5-fluorouracil toxicity |
| DYM | 98 | 94 | 223800 | Dyggve-Melchior-Clausen disease |
| DYNC1H1 | 133 | 88 | 614228 | Charcot-Marie-Tooth disease axonal type 20 |
| DYRK1A | 151 | 89 | 614104 | Mental retardation autosomal dominant 7 |
| EFTUD2 | 109 | 87 | 610536 | Mandibulofacial dysostosis Guion-Almeida type |
| EHMT1 | 105 | 85 | 610253 | Kleefstra syndrome |
| EIF2AK3 | 117 | 94 | 226980 | Wolcott-Rallison syndrome |
| ELOVL4 | 111 | 92 | 614457 | Ichthyosis spastic quadriplegia and mental retardation |
| EMX2 | 106 | 99 | 269160 | Schizencephaly |
| EP300 | 141 | 93 | 114500 | Colorectal cancer somatic |
| EPB41L1 | 91 | 85 | 614257 | Mental retardation autosomal dominant 11 |
| ERCC2 | 88 | 89 | 610756 | Cerebrooculofacioskeletal syndrome 2 |
| ERCC3 | 134 | 93 | 601675 | Trichothiodystrophy |
| ERCC6 | 157 | 92 | 214150 | Cerebrooculofacioskeletal syndrome 1 |

| | | | | |
|---|---|---|---|---|
| ERCC8 | 86 | 95 | 216400 | Cockayne syndrome type A |
| ERLIN2 | 125 | 94 | 611225 | Spastic paraplegia 18 autosomal recessive |
| ESCO2 | 91 | 97 | 268300 | Roberts syndrome |
| ETHE1 | 65 | 93 | 602473 | Ethylmalonic encephalopathy |
| FANCD2 | 118 | 86 | 227646 | Fanconi anemia complementation group D2 |
| FBN1 | 110 | 88 | 102370 | Acromicric dysplasia |
| FGD1 | 77 | 91 | 305400 | Aarskog-Scott syndrome |
| FGFR2 | 132 | 80 | 207410 | Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis |
| FGFR3 | 52 | 93 | 100800 | Achondroplasia |
| FH | 88 | 80 | 606812 | Fumarase deficiency |
| FKRP | 67 | 98 | 613153 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 5 |
| FKTN | 112 | 91 | 611615 | Cardiomyopathy dilated 1X |
| FLNA | 83 | 89 | 314400 | Cardiac valvular dysplasia X-linked |
| FMR1 | 84 | 93 | 300624 | Fragile X syndrome |
| FOXG1 | 97 | 100 | 613454 | Rett syndrome congenital variant |
| FOXP1 | 112 | 82 | 613670 | Mental retardation with language impairment and autistic features |
| FRAS1 | 116 | 82 | 219000 | Fraser syndrome |
| FTO | 124 | 87 | 612938 | Growth retardation developmental delay coarse facies and early death |
| FTSJ1 | 84 | 86 | 309549 | Mental retardation X-linked 9 |
| FUCA1 | 81 | 88 | 230000 | Fucosidosis |
| GAD1 | 112 | 87 | 603513 | Cerebral palsy spastic quadriplegic 1 |
| GALE | 107 | 97 | 230350 | Galactose epimerase deficiency |
| GALT | 128 | 90 | 230400 | Galactosemia |
| GAMT | 89 | 93 | 612736 | Cerebral creatine deficiency syndrome 2 |
| GATAD2B | 133 | 88 | 615074 | Mental retardation autosomal dominant 18 |
| GATM | 91 | 87 | 612718 | Cerebral creatine deficiency syndrome 3 |
| GCH1 | 85 | 98 | 128230 | Dystonia DOPA-responsive with or without hyperphenylalaninemia |
| GCSH | 50 | 31 | 605899 | Glycine encephalopathy |
| GDI1 | 100 | 87 | 300849 | Mental retardation X-linked 41 |
| GFAP | 83 | 89 | 203450 | Alexander disease |
| GJC2 | 34 | 99 | 608804 | Leukodystrophy hypomyelinating 2 |
| GK | 58 | 29 | 307030 | Glycerol kinase deficiency |
| GLDC | 74 | 67 | 605899 | Glycine encephalopathy |

| | | | | |
|---|---|---|---|---|
| GLI2 | 111 | 89 | 610829 | Holoprosencephaly-9 |
| GLI3 | 113 | 90 | 175700 | Greig cephalopolysyndactyly syndrome |
| GNAS | 120 | 56 | 102200 | Acromegaly |
| GNPAT | 139 | 83 | 222765 | Chondrodysplasia punctata rhizomelic type 2 |
| GNS | 86 | 85 | 252940 | Mucopolysaccharidosis type IIID |
| GPC3 | 85 | 94 | 312870 | Simpson-Golabi-Behmel syndrome type 1 |
| GPHN | 120 | 95 | 252150 | Molybdenum cofactor deficiency type C |
| GPR56 | 91 | 95 | 606854 | Polymicrogyria bilateral frontoparietal |
| GRIA3 | 97 | 80 | 300699 | Mental retardation X-linked 94 |
| GRIK2 | 119 | 90 | 611092 | Mental retardation autosomal recessive 6 |
| GRIN1 | 68 | 94 | 614254 | Mental retardation autosomal dominant 8 |
| GRIN2A | 144 | 96 | 613971 | Epilepsy with neurodevelopmental defects |
| GRIN2B | 147 | 92 | 613970 | Mental retardation autosomal dominant 6 |
| GSS | 104 | 77 | 266130 | Glutathione synthetase deficiency |
| GTF2H5 | 101 | 37 | 601675 | Trichothiodystrophy complementation group A |
| GUSB | 79 | 70 | 253220 | Mucopolysaccharidosis VII |
| HAX1 | 149 | 99 | 610738 | Neutropenia severe congenital 3 autosomal recessive |
| HCCS | 100 | 88 | 309801 | Microphthalmia syndromic 7 |
| HCFC1 | 56 | 90 | 309541 | Mental retardation X-linked 3 |
| HDAC4 | 77 | 82 | 600430 | Brachydactyly-mental retardation syndrome |
| HDAC8 | 89 | 86 | 300882 | Cornelia de Lange syndrome 5 |
| HESX1 | 86 | 87 | 182230 | Growth hormone deficiency with pituitary anomalies |
| HLCS | 131 | 89 | 253270 | Holocarboxylase synthetase deficiency |
| HOXA1 | 118 | 94 | 601536 | Athabaskan brainstem dysgenesis syndrome |
| HPD | 94 | 97 | 140350 | Hawkinsinuria |
| HPRT1 | 75 | 80 | 300323 | HPRT-related gout |
| HRAS | 80 | 92 | 218040 | Congenital myopathy with excess of muscle spindles |
| HSD17B10 | 88 | 86 | 300438 | 17-beta-hydroxysteroid dehydrogenase X deficiency |
| IDS | 92 | 89 | 309900 | Mucopolysaccharidosis II |
| IDUA | 83 | 88 | 607014 | Mucopolysaccharidosis Ih |
| IER3IP1 | 92 | 49 | 614231 | Microcephaly epilepsy and diabetes syndrome |
| IGF1 | 151 | 78 | 608747 | Growth retardation with deafness and mental retardation due to IGF1 deficiency |
| IKBKG | 84 | 79 | 300291 | Ectodermal dysplasia hypohidrotic with immune deficiency |

| | | | | |
|---|---|---|---|---|
| IL1RAPL1 | 121 | 94 | 300143 | Mental retardation X-linked 21/34 |
| INPP5E | 67 | 95 | 213300 | Joubert syndrome 1 |
| IQSEC2 | 59 | 92 | 309530 | Mental retardation X-linked 1 |
| ISPD | 97 | 87 | 614643 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 7 |
| JAM3 | 89 | 91 | 613730 | Hemorrhagic destruction of the brain subependymal calcification and cataracts |
| KANSL1 | 81 | 86 | 610443 | Koolen-De Vries syndrome |
| KAT6B | 154 | 65 | 606170 | Genitopatellar syndrome |
| KCNJ11 | 107 | 100 | 606176 | Diabetes mellitus permanent neonatal with neurologic features |
| KCNK9 | 119 | 97 | 612292 | Birk-Barel mental retardation dysmorphism syndrome |
| KCNQ2 | 72 | 96 | 613720 | Epileptic encephalopathy early infantile 7 |
| KCNT1 | 67 | 94 | 615005 | Epilepsy nocturnal frontal lobe 5 |
| KCTD7 | 109 | 87 | 611726 | Epilepsy progressive myoclonic 3 with or without intracellular inclusions |
| KDM5C | 97 | 87 | 300534 | Mental retardation X-linked syndromic Claes-Jensen type |
| KDM6A | 95 | 84 | 300867 | Kabuki syndrome 2 |
| KIAA1279 | 120 | 97 | 609460 | Goldberg-Shprintzen megacolon syndrome |
| KIF11 | 104 | 93 | 152950 | Microcephaly with or without chorioretinopathy lymphedema or mental retardation |
| KIF7 | 67 | 89 | 200990 | Acrocallosal syndrome |
| KIRREL3 | 78 | 84 | 612581 | Mental retardation autosomal dominant 4 |
| KMT2D | 111 | 96 | 147920 | Kabuki syndrome 1 |
| KRAS | 61 | 10 | 109800 | Bladder cancer somatic |
| KRBOX4 | 113 | 54 | 200 | - |
| L1CAM | 95 | 93 | 304100 | Corpus callosum partial agenesis of |
| L2HGDH | 81 | 97 | 236792 | L-2-hydroxyglutaric aciduria |
| LAMA2 | 112 | 86 | 607855 | Muscular dystrophy congenital merosin-deficient |
| LAMC3 | 99 | 85 | 614115 | Cortical malformations occipital |
| LAMP2 | 90 | 87 | 300257 | Danon disease |
| LARGE | 116 | 82 | 613154 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 6 |
| LARP7 | 107 | 80 | 615071 | Alazami syndrome |
| LIG4 | 186 | 100 | 606593 | LIG4 syndrome |
| LRP2 | 124 | 87 | 222448 | Donnai-Barrow syndrome |
| LRPPRC | 98 | 90 | 220111 | Leigh syndrome French-Canadian type |
| MAGT1 | 86 | 94 | 300853 | Immunodeficiency X-linked with magnesium defect Epstein-Barr virus infection and neoplasia |
| MAN1B1 | 97 | 85 | 614202 | Mental retardation autosomal recessive 15 |

| | | | | |
|---|---|---|---|---|
| MAN2B1 | 89 | 85 | 248500 | Mannosidosis alpha- types I and II |
| MANBA | 98 | 92 | 248510 | Mannosidosis beta |
| MAOA | 81 | 90 | 300615 | Brunner syndrome |
| MAP2K1 | 104 | 73 | 615279 | Cardiofaciocutaneous syndrome 3 |
| MAP2K2 | 99 | 73 | 615280 | Cardiofaciocutaneous syndrome 4 |
| MAT1A | 105 | 87 | 250850 | Hypermethioninemia persistent autosomal dominant due to methionine adenosyltransferase I/III deficiency |
| MBD5 | 161 | 97 | 156200 | Mental retardation autosomal dominant 1 |
| MCCC1 | 117 | 91 | 210200 | 3-Methylcrotonyl-CoA carboxylase 1 deficiency |
| MCCC2 | 118 | 91 | 210210 | 3-Methylcrotonyl-CoA carboxylase 2 deficiency |
| MCOLN1 | 97 | 90 | 252650 | Mucolipidosis IV |
| MCPH1 | 121 | 97 | 251200 | Microcephaly 1 primary autosomal recessive |
| MECP2 | 111 | 100 | 105830 | Angelman syndrome |
| MED12 | 115 | 88 | 309520 | Lujan-Fryns syndrome |
| MED17 | 142 | 89 | 613668 | Microcephaly postnatal progressive with seizures and brain atrophy |
| MED23 | 119 | 94 | 614249 | Mental retardation autosomal recessive 18 |
| MEF2C | 119 | 87 | 613443 | Mental retardation stereotypic movements epilepsy and/or cerebral malformations |
| MGAT2 | 207 | 100 | 212066 | Congenital disorder of glycosylation type IIa |
| MID1 | 115 | 91 | 300000 | Opitz GBBB syndrome type I |
| MKKS | 147 | 97 | 209900 | Bardet-Biedl syndrome 6 |
| MLYCD | 90 | 86 | 248360 | Malonyl-CoA decarboxylase deficiency |
| MMAA | 172 | 98 | 251100 | Methylmalonic aciduria vitamin B12-responsive |
| MMACHC | 166 | 89 | 277400 | Methylmalonic aciduria and homocystinuria cblC type |
| MMADHC | 79 | 80 | 200 | - |
| MMADHC | 79 | 80 | 200 | - |
| MOCS1 | 125 | 20 | 252150 | Molybdenum cofactor deficiency type A |
| MOCS2 | 85 | 70 | 252150 | Molybdenum cofactor deficiency type B |
| MPDU1 | 134 | 96 | 609180 | Congenital disorder of glycosylation type If |
| MPLKIP | 76 | 100 | 234050 | Trichothiodystrophy nonphotosensitive 1 |
| MRPS22 | 104 | 77 | 611719 | Combined oxidative phosphorylation deficiency 5 |
| MTR | 119 | 86 | 250940 | Homocystinuria-megaloblastic anemia cblG complementation type |
| MTRR | 114 | 93 | 236270 | Homocystinuria-megaloblastic anemia cbl E type |
| MUT | 125 | 94 | 251000 | Methylmalonic aciduria mut(0) type |
| MVK | 101 | 78 | 260920 | Hyper-IgD syndrome |

| | | | | |
|---|---|---|---|---|
| MYCN | 78 | 98 | 164280 | Feingold syndrome |
| MYO5A | 101 | 86 | 214450 | Griscelli syndrome type 1 |
| NAA10 | 72 | 95 | 300855 | N-terminal acetyltransferase deficiency |
| NAGA | 93 | 84 | 609242 | Kanzaki disease |
| NAGLU | 62 | 92 | 252920 | Mucopolysaccharidosis type IIIB (Sanfilippo B) |
| NBN | 127 | 94 | 613065 | Leukemia, acute lymphoblastic |
| NBN | 127 | 94 | 251260 | Nijmegen breakage syndrome |
| NBN | 127 | 94 | 609135 | Aplastic anemia |
| NDE1 | 94 | 61 | 614019 | Lissencephaly 4 (with microcephaly) |
| NDP | 112 | 86 | 305390 | Exudative vitreoretinopathy X-linked |
| NDUFA1 | 128 | 100 | 252010 | Mitochondrial complex I deficiency |
| NDUFA11 | 59 | 90 | 252010 | Mitochondrial complex I deficiency |
| NDUFA12 | 102 | 87 | 256000 | Leigh syndrome due to mitochondrial complex 1 deficiency |
| NDUFS1 | 87 | 93 | 252010 | Mitochondrial complex I deficiency |
| NDUFS2 | 161 | 83 | 252010 | Mitochondrial complex I deficiency |
| NDUFS3 | 180 | 89 | 256000 | Leigh syndrome due to mitochondrial complex I deficiency |
| NDUFS4 | 120 | 94 | 256000 | Leigh syndrome |
| NDUFS7 | 103 | 91 | 256000 | Leigh syndrome |
| NDUFS8 | 93 | 86 | 256000 | Leigh syndrome due to mitochondrial complex I deficiency |
| NDUFV1 | 70 | 85 | 252010 | Mitochondrial complex I deficiency |
| NEU1 | 16 | 87 | 256550 | Sialidosis type I |
| NF1 | 109 | 85 | 607785 | Leukemia juvenile myelomonocytic |
| NHS | 109 | 94 | 302200 | Cataract 40 X-linked |
| NIPBL | 126 | 97 | 122470 | Cornelia de Lange syndrome 1 |
| NKX2-1 | 88 | 93 | 118700 | Chorea hereditary benign |
| NLGN4X | 94 | 24 | 200 | - |
| NLRP3 | 128 | 96 | 607115 | CINCA syndrome |
| NPHP1 | 117 | 92 | 609583 | Joubert syndrome 4 |
| NRXN1 | 130 | 92 | 614325 | Pitt-Hopkins-like syndrome 2 |
| NSD1 | 141 | 93 | 130650 | Beckwith-Wiedemann syndrome |
| NSDHL | 77 | 91 | 308050 | CHILD syndrome |
| NSUN2 | 129 | 87 | 611091 | Mental retardation autosomal recessive 5 |
| NTRK1 | 70 | 89 | 256800 | Insensitivity to pain congenital with anhidrosis |

| | | | | |
|---|---|---|---|---|
| OCLN | 144 | 89 | 251290 | Band-like calcification with simplified gyration and polymicrogyria |
| OCRL | 107 | 89 | 300555 | Dent disease 2 |
| OFD1 | 61 | 54 | 300804 | Joubert syndrome 10 |
| OPHN1 | 95 | 84 | 300486 | Mental retardation X-linked with cerebellar hypoplasia and distinctive facial appearance |
| ORC1 | 120 | 84 | 224690 | Meier-Gorlin syndrome 1 |
| PACS1 | 112 | 92 | 615009 | Mental retardation autosomal dominant 17 |
| PAFAH1B1 | 87 | 63 | 607432 | Lissencephaly 1 |
| PAK3 | 86 | 92 | 300558 | Mental retardation X-linked 30/47 |
| PANK2 | 113 | 97 | 607236 | HARP syndrome |
| PAX6 | 103 | 82 | 106210 | Aniridia |
| PC | 98 | 91 | 266150 | Pyruvate carboxylase deficiency |
| PCDH19 | 107 | 95 | 300088 | Epileptic encephalopathy early infantile 9 |
| PCNT | 106 | 81 | 210720 | Microcephalic osteodysplastic primordial dwarfism type II |
| PDHA1 | 105 | 85 | 308930 | Leigh syndrome X-linked |
| PDSS1 | 109 | 86 | 614651 | Coenzyme Q10 deficiency primary 2 |
| PDSS2 | 94 | 92 | 614652 | Coenzyme Q10 deficiency primary 3 |
| PEPD | 78 | 89 | 170100 | Prolidase deficiency |
| PEX1 | 122 | 93 | 214100 | Peroxisome biogenesis disorder 1A (Zellweger) |
| PEX10 | 77 | 90 | 614870 | Peroxisome biogenesis disorder 6A (Zellweger) |
| PEX11B | 184 | 88 | 614920 | Peroxisome biogenesis disorder 14B |
| PEX13 | 131 | 95 | 614883 | Peroxisome biogenesis disorder 11A (Zellweger) |
| PEX26 | 122 | 92 | 614872 | Peroxisome biogenesis disorder 7A (Zellweger) |
| PEX5 | 102 | 76 | 214110 | Peroxisome biogenesis disorder 2A (Zellweger) |
| PEX7 | 112 | 91 | 614879 | Peroxisome biogenesis disorder 9B |
| PGK1 | 82 | 43 | 300653 | Phosphoglycerate kinase 1 deficiency |
| PHF6 | 87 | 94 | 301900 | Borjeson-Forssman-Lehmann syndrome |
| PHF8 | 93 | 83 | 300263 | Mental retardation syndrome X-linked Siderius type |
| PHGDH | 103 | 86 | 601815 | Phosphoglycerate dehydrogenase deficiency |
| PIGN | 101 | 97 | 614080 | Multiple congenital anomalies-hypotonia-seizures syndrome 1 |
| PIGO | 114 | 93 | 614749 | Hyperphosphatasia with mental retardation syndrome 2 |
| PIGV | 191 | 97 | 239300 | Hyperphosphatasia with mental retardation syndrome 1 |
| PIK3R2 | 84 | 89 | 603387 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome |
| PLCB1 | 117 | 93 | 613722 | Epileptic encephalopathy early infantile 12 |

| | | | | |
|---|---|---|---|---|
| PLP1 | 79 | 79 | 312080 | Pelizaeus-Merzbacher disease |
| PMM2 | 118 | 89 | 212065 | Congenital disorder of glycosylation type Ia |
| PNKP | 77 | 92 | 613402 | Epileptic encephalopathy early infantile 10 |
| PNP | 138 | 97 | 613179 | Immunodeficiency due to purine nucleoside phosphorylase deficiency |
| POC1A | 96 | 76 | 614813 | Short stature onychodysplasia facial dysmorphism and hypotrichosis |
| POLR3A | 107 | 83 | 607694 | Leukodystrophy hypomyelinating 7 with or without oligodontia and/or hypogonadotropic hypogonadism |
| POLR3B | 112 | 89 | 614381 | Leukodystrophy hypomyelinating 8 with or without oligodontia and/or hypogonadotropic hypogonadism |
| POMGNT1 | 106 | 90 | 253280 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 3 |
| POMT1 | 118 | 87 | 236670 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 1 |
| POMT2 | 84 | 90 | 613150 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 2 |
| PORCN | 88 | 84 | 305600 | Focal dermal hypoplasia |
| PPOX | 107 | 93 | 176200 | Porphyria variegata |
| PQBP1 | 105 | 80 | 309500 | Renpenning syndrome |
| PRODH | 63 | 81 | 239500 | Hyperprolinemia type I |
| PRPS1 | 116 | 73 | 301835 | Arts syndrome |
| PRSS12 | 113 | 87 | 249500 | Mental retardation autosomal recessive 1 |
| PTCH1 | 85 | 48 | 605462 | Basal cell carcinoma somatic |
| PTCHD1 | 138 | 99 | 200 | - |
| PTEN | 130 | 65 | 153480 | Bannayan-Riley-Ruvalcaba syndrome |
| PTPN11 | 90 | 25 | 151100 | LEOPARD syndrome 1 |
| PUS1 | 58 | 95 | 200 | Mitochondrial myopathy and sideroblastic anemia 1 |
| PVRL1 | 72 | 97 | 200 | - |
| PYCR1 | 78 | 86 | 612940 | Cutis laxa autosomal recessive type IIB |
| RAB18 | 109 | 90 | 614222 | Warburg micro syndrome 3 |
| RAB27A | 137 | 91 | 607624 | Griscelli syndrome type 2 |
| RAB39B | 116 | 96 | 300271 | Mental retardation X-linked 72 |
| RAB3GAP1 | 121 | 95 | 600118 | Warburg micro syndrome 1 |
| RAB3GAP2 | 111 | 93 | 212720 | Martsolf syndrome |
| RAB40AL | 71 | 9 | 300519 | Mental retardation X-linked syndromic Martin-Probst type |
| RAD21 | 96 | 79 | 614701 | Cornelia de Lange syndrome 4 |
| RAF1 | 115 | 82 | 611554 | LEOPARD syndrome 2 |
| RAI1 | 134 | 100 | 182290 | Smith-Magenis syndrome |
| RARS2 | 96 | 96 | 611523 | Pontocerebellar hypoplasia type 6 |

| | | | | |
|---|---|---|---|---|
| RBM28 | 106 | 86 | 612079 | Alopecia neurologic defects and endocrinopathy syndrome |
| RELN | 119 | 87 | 257320 | Lissencephaly 2 (Norman-Roberts type) |
| RFT1 | 91 | 84 | 612015 | Congenital disorder of glycosylation type In |
| RMND1 | 66 | 54 | 614922 | Combined oxidative phosphorylation deficiency 11 |
| RNASEH2A | 103 | 87 | 610333 | Aicardi-Goutieres syndrome 4 |
| RNASEH2B | 98 | 94 | 610181 | Aicardi-Goutieres syndrome 2 |
| RNASEH2C | 134 | 100 | 610329 | Aicardi-Goutieres syndrome 3 |
| ROGDI | 83 | 96 | 226750 | Kohlschutter-Tonz syndrome |
| RPGRIP1L | 100 | 92 | 216360 | COACH syndrome |
| RPS6KA3 | 76 | 98 | 303600 | Coffin-Lowry syndrome |
| SALL1 | 135 | 52 | 107480 | Townes-Brocks branchiootorenal-like syndrome |
| SATB2 | 124 | 86 | 119540 | Cleft palate and mental retardation |
| SC5D | 145 | 90 | 200 | - |
| SC5D | 145 | 90 | 200 | - |
| SCN1A | 116 | 54 | 607208 | Dravet syndrome |
| SCN2A | 149 | 56 | 613721 | Epileptic encephalopathy early infantile 11 |
| SCN8A | 147 | 89 | 614306 | Cognitive impairment with or without cerebellar ataxia |
| SCO2 | 88 | 100 | 604377 | Cardioencephalomyopathy fatal infantile due to cytochrome c oxidase deficiency 1 |
| SDHA | 39 | 18 | 613642 | Cardiomyopathy dilated 1GG |
| SERAC1 | 95 | 95 | 614739 | 3-methylglutaconic aciduria with deafness encephalopathy and Leigh-like syndrome |
| SETBP1 | 164 | 97 | 269150 | Schinzel-Giedion midface retraction syndrome |
| SHH | 81 | 100 | 142945 | Holoprosencephaly-3 |
| SHOC2 | 127 | 96 | 607721 | Noonan-like syndrome with loose anagen hair |
| SHROOM4 | 102 | 79 | 300434 | Stocco dos Santos X-linked mental retardation syndrome |
| SIL1 | 111 | 82 | 248800 | Marinesco-Sjogren syndrome |
| SIX3 | 101 | 96 | 157170 | Holoprosencephaly-2 |
| SKI | 66 | 89 | 182212 | Shprintzen-Goldberg syndrome |
| SLC12A6 | 106 | 83 | 218000 | Agenesis of the corpus callosum with peripheral neuropathy |
| SLC16A2 | 89 | 90 | 300523 | Allan-Herndon-Dudley syndrome |
| SLC17A5 | 107 | 94 | 604369 | Salla disease |
| SLC25A15 | 146 | 88 | 238970 | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome |
| SLC25A22 | 64 | 94 | 609304 | Epileptic encephalopathy early infantile 3 |
| SLC2A1 | 90 | 94 | 601042 | Dystonia 9 |

| | | | | |
|---|---|---|---|---|
| SLC33A1 | 93 | 96 | 614482 | Congenital cataracts hearing loss and neurodegeneration |
| SLC35C1 | 83 | 100 | 266265 | Congenital disorder of glycosylation type IIc |
| SLC4A4 | 112 | 89 | 604278 | Renal tubular acidosis proximal with ocular abnormalities |
| SLC6A8 | 15 | 27 | 300352 | Cerebral creatine deficiency syndrome 1 |
| SLC9A6 | 93 | 92 | 300243 | Mental retardation X-linked syndromic Christianson type |
| SMAD4 | 123 | 93 | 200 | - |
| SMAD4 | 123 | 93 | 200 | - |
| SMARCA2 | 101 | 85 | 601358 | Nicolaides-Baraitser syndrome |
| SMARCA4 | 87 | 86 | 614609 | Mental retardation autosomal dominant 16 |
| SMARCB1 | 150 | 85 | 614608 | Mental retardation autosomal dominant 15 |
| SMC1A | 106 | 93 | 200 | - |
| SMC3 | 117 | 83 | 200 | - |
| SMOC1 | 98 | 82 | 206920 | Microphthalmia with limb anomalies |
| SMPD1 | 95 | 94 | 257200 | Niemann-Pick disease type A |
| SMS | 49 | 23 | 309583 | Mental retardation X-linked Snyder-Robinson type |
| SNAP29 | 109 | 93 | 609528 | Cerebral dysgenesis neuropathy ichthyosis and palmoplantar keratoderma syndrome |
| SOBP | 111 | 92 | 613671 | Mental retardation anterior maxillary protrusion and strabismus |
| SOS1 | 112 | 94 | 135300 | Fibromatosis gingival |
| SOX10 | 55 | 89 | 609136 | PCWH syndrome |
| SOX2-OT | ? | ? | 200 | - |
| SOX3 | 42 | 48 | 300123 | Mental retardation X-linked with isolated growth hormone deficiency |
| SPRED1 | 135 | 95 | 611431 | Legius syndrome |
| SRCAP | 143 | 92 | 136140 | Floating-Harbor syndrome |
| SRD5A3 | 107 | 84 | 612379 | Congenital disorder of glycosylation type Iq |
| SRPX2 | 87 | 77 | 300643 | Rolandic epilepsy mental retardation and speech dyspraxia |
| ST3GAL3 | 128 | 92 | 615006 | Epileptic encephalopathy early infantile 15 |
| ST3GAL3 | 128 | 92 | 615006 | Epileptic encephalopathy early infantile 15 |
| ST3GAL3 | 128 | 92 | 615006 | Epileptic encephalopathy early infantile 15 |
| STIL | 146 | 98 | 612703 | Microcephaly 7 primary autosomal recessive |
| STRA6 | 77 | 84 | 601186 | Microphthalmia isolated with coloboma 8 |
| STXBP1 | 94 | 46 | 612164 | Epileptic encephalopathy early infantile 4 |
| SUOX | 174 | 99 | 272300 | Sulfite oxidase deficiency |
| SURF1 | 90 | 79 | 256000 | Leigh syndrome due to COX deficiency |

| | | | | |
|---|---|---|---|---|
| SYN1 | 55 | 88 | 300491 | Epilepsy X-linked with variable learning disabilities and behavior disorders |
| SYNGAP1 | 56 | 88 | 612621 | Mental retardation autosomal dominant 5 |
| SYP | 68 | 98 | 300802 | Mental retardation X-linked 96 |
| SYT14 | 136 | 80 | 614229 | Spinocerebellar ataxia autosomal recessive 11 |
| TAT | 122 | 85 | 276600 | Tyrosinemia type II |
| TBC1D24 | 99 | 99 | 615338 | Epileptic encephalopathy early infantile 16 |
| TBCE | 131 | 91 | 241410 | Hypoparathyroidism-retardation-dysmorphism syndrome |
| TCF4 | 117 | 81 | 610954 | Pitt-Hopkins syndrome |
| TECR | 77 | 77 | 614020 | Mental retardation autosomal recessive 14 |
| TGFBR1 | 125 | 93 | 609192 | Loeys-Dietz syndrome type 1A |
| TGFBR2 | 101 | 87 | 614331 | Colorectal cancer hereditary nonpolyposis type 6 |
| TGIF1 | 160 | 94 | 200 | - |
| THRB | 129 | 82 | 188570 | Thyroid hormone resistance |
| TIMM8A | 56 | 60 | 200 | Deafness X-linked 1 |
| TMCO1 | 83 | 78 | 614132 | Craniofacial dysmorphism skeletal anomalies and mental retardation syndrome |
| TMEM165 | 83 | 91 | 614727 | Congenital disorder of glycosylation type IIk |
| TMEM231 | 87 | 66 | 614970 | Joubert syndrome 20 |
| TMEM237 | 99 | 91 | 614424 | Joubert syndrome 14 |
| TMEM67 | 107 | 96 | 216360 | COACH syndrome |
| TRAPPC9 | 86 | 85 | 613192 | Mental retardation autosomal recessive 13 |
| TREX1 | 122 | 100 | 225750 | Aicardi-Goutieres syndrome 1 dominant and recessive |
| TSC1 | 106 | 87 | 607341 | Focal cortical dysplasia Taylor balloon cell type |
| TSC2 | 83 | 92 | 606690 | Lymphangioleiomyomatosis somatic |
| TSPAN7 | 77 | 87 | 300210 | Mental retardation X-linked 58 |
| TTC8 | 107 | 91 | 209900 | Bardet-Biedl syndrome 8 |
| TUBA1A | 58 | 9 | 611603 | Lissencephaly 3 |
| TUBB2B | 129 | 13 | 610031 | Polymicrogyria symmetric or asymmetric |
| TUSC3 | 73 | 33 | 611093 | Mental retardation autosomal recessive 7 |
| UBE2A | 74 | 93 | 300860 | Mental retardation X-linked syndromic Nascimento-type |
| UBE3A | 100 | 31 | 105830 | Angelman syndrome |
| UBR1 | 105 | 97 | 243800 | Johanson-Blizzard syndrome |
| UPB1 | 124 | 81 | 613161 | Beta-ureidopropionase deficiency |
| UPF3B | 86 | 87 | 300676 | Mental retardation X-linked syndromic 14 |

| | | | | |
|---|---|---|---|---|
| VLDLR | 131 | 89 | 224050 | Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1 |
| VPS13B | 117 | 92 | 216550 | Cohen syndrome |
| WDR62 | 103 | 89 | 604317 | Microcephaly 2 primary autosomal recessive with or without cortical malformations |
| XPA | 79 | 89 | 278700 | Xeroderma pigmentosum group A |
| ZDHHC9 | 86 | 83 | 300799 | Mental retardation X-linked syndromic Raymond type |
| ZEB2 | 166 | 92 | 235730 | Mowat-Wilson syndrome |
| ZIC2 | 40 | 95 | 609637 | Holoprosencephaly-5 |
| ZNF41 | 107 | 51 | 300848 | Mental retardation X-linked 89 |
| ZNF592 | 128 | 86 | 606937 | Spinocerebellar ataxia autosomal recessive 5 |
| ZNF674 | 106 | 38 | 300851 | Mental retardation X-linked 92 |
| ZNF711 | 107 | 98 | 300803 | Mental retardation X-linked 97 |
| ZNF81 | 84 | 99 | 300498 | Mental retardation X-linked 45 |

*Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013*
*Depth describes the average number of reads seen across 50 exomes*
*Coverage describes the average coverage of a gene across 50 exomes in percentiles*
*OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013*

*Ad 1. OMIM identifier 200 signifies a gene without a current OMIM association*

*Ad 2. OMIM phenotype descriptions between {} signify risk factors*