# INTELLECTUAL DISABILITY GENE PANEL DG 2.4.x

| *Gene* | *Median coverage* | *% covered > 10x* | *% covered > 20x* | *Associated phenotype description and OMIM ID* |
|---|---|---|---|---|
| A2ML1 | 111.3 | 99% | 99% | Noonan-like syndrome (Vissers et al. 2015) |
| ABCC9 | 115.6 | 100% | 98% | Cardiomyopathy, dilated, 1O, 608569<br>Atrial fibrillation, familial, 12, 614050<br>Hypertrichotic osteochondrodysplasia, 239850 |
| ABCD1 | 77.5 | 79% | 74% | Adrenoleukodystrophy, 300100<br>Adrenomyeloneuropathy, adult, 300100 |
| ABCD4 | 116.1 | 100% | 98% | Methylmalonic aciduria and homocystinuria, cblJ type, 614857 |
| ABHD5 | 135.3 | 100% | 100% | Chanarin-Dorfman syndrome, 275630 |
| ACAD9 | 94.6 | 100% | 100% | ACAD9 deficiency, 611126 |
| ACO2 | 92.9 | 92% | 86% | Infantile cerebellar-retinal degeneration, 614559 |
| ACOX1 | 90.7 | 98% | 97% | Peroxisomal acyl-CoA oxidase deficiency, 264470 |
| ACSF3 | 93.9 | 100% | 100% | Combined malonic and methylmalonic aciduria, 614265 |
| ACSL4 | 135.2 | 100% | 100% | Mental retardation, X-linked 63, 300387 |
| ACTB | 72.5 | 100% | 95% | Dystonia, juvenile-onset, 607371<br>Baraitser-Winter syndrome 1, 243310 |
| ACTG1 | 65.8 | 100% | 91% | Deafness, autosomal dominant 20/26, 604717<br>Baraitser-Winter syndrome 2, 614583 |
| ACVR1 | 106.6 | 100% | 100% | Fibrodysplasia ossificans progressiva, 135100 |
| ACY1 | 98.7 | 100% | 99% | Aminoacylase 1 deficiency, 609924 |
| ADAR | 140.7 | 98% | 97% | Dyschromatosis symmetrica hereditaria, 127400<br>Aicardi-Goutieres syndrome 6, 615010 |
| ADAT3 | 80.5 | 100% | 100% | Mental retardation, autosomal recessive 36, 615286 |
| ADCK3 | 114.6 | 100% | 98% | Coenzyme Q10 deficiency, primary, 4, 612016 |
| ADK | 121.8 | 94% | 94% | Hypermethioninemia due to adenosine kinase deficiency, 614300 |
| ADNP | 212.3 | 100% | 100% | Mental retardation, autosomal dominant, 28, 615873 |
| ADSL | 140.6 | 100% | 100% | ade(-)I bifunctional Adenylosuccinase deficiency, 103050 |
| AFF2 | 154.4 | 99% | 98% | Mental retardation, X-linked, FRAXE type, 309548 |
| AGA | 126 | 100% | 99% | Aspartylglucosaminuria, 208400 |
| AGO2 | 97.1 | 97% | 93% | No OMIM phenotype |

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| AGPAT2 | 65.5 | 99% | 89% | Lipodystrophy, congenital generalized, type 1, 608594 |
| AGTR2 | 214.9 | 100% | 100% | Mental retardation, X-linked 88, 300852 |
| AHCY | 89.7 | 99% | 82% | Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 |
| AHDC1 | 121 | 99% | 98% | Xia-Gibbs syndrome,615829 |
| AHI1 | 120.1 | 100% | 99% | Joubert syndrome-3, 608629 |
| AIFM1 | 132.5 | 100% | 99% | Combined oxidative phosphorylation deficiency 6, 300816<br>Cowchock syndrome, 310490 |
| AIMP1 | 120.9 | 100% | 100% | Leukodystrophy, hypomyelinating, 3, 260600 |
| AK1 | 96.8 | 100% | 100% | Hemolytic anemia due to adenylate kinase deficiency, 612631 |
| AKT3 | 121.6 | 100% | 99% | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387 |
| ALDH18A1 | 105.7 | 99% | 94% | Cutis laxa, autosomal recessive, type IIIA, 219150 |
| ALDH3A2 | 104.8 | 100% | 100% | Sjogren-Larsson syndrome, 270200 |
| ALDH4A1 | 81.8 | 92% | 87% | Hyperprolinemia, type II, 239510 |
| ALDH5A1 | 80.6 | 97% | 97% | Succinic semialdehyde dehydrogenase deficiency, 271980 |
| ALG1 | 55.6 | 45% | 45% | ngenital disorder of glycosylation, type Ik, 608540 |
| ALG12 | 114.8 | 100% | 100% | Congenital disorder of glycosylation, type Ig, 607143 |
| ALG13 | 123.9 | 96% | 95% | Congenital disorder of glycosylation, type Is, 300884 |
| ALG2 | 118.3 | 100% | 97% | Congenital disorder of glycosylation, type Ii, 607906 |
| ALG3 | 101.7 | 98% | 96% | Congenital disorder of glycosylation, type Id, 601110 |
| ALG6 | 102.9 | 100% | 99% | Congenital disorder, type Ic, 603147 |
| ALG9 | 105 | 100% | 98% | Congenital disorder of glycosylation, type Il, 608776 |
| ALX1 | 173.4 | 100% | 100% | Frontonasal dysplasia 3, 613456 |
| ALX4 | 91.5 | 100% | 100% | Parietal foramina 2, 609597<br>Frontonasal dysplasia 2, 613451 |
| AMPD2 | 116.8 | 100% | 98% | Pontocerebellar hypoplasia, type 9, 615809<br>?Spastic paraplegia 63, 615686 |
| AMT | 142.3 | 100% | 100% | Glycine encephalopathy, 605899 |
| ANK3 | 164.5 | 100% | 100% | ?Mental retardation, autosomal recessive, 37 |
| ANKH | 124.9 | 100% | 100% | Craniometaphyseal dysplasia, 123000<br>Chondrocalcinosis 2, 118600 |
| ANKRD11 | 124.7 | 91% | 87% | KBG syndrome, 148050 |
| ANO10 | 117.8 | 100% | 100% | Spinocerebellar ataxia, autosomal recessive 10, 613728 |
| ANTXR1 | 91.9 | 95% | 89% | GAPO syndrome, 230740<br>{Hemangioma, capillary infantile, susceptibility to}, 602089 |

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| AP1S2 | 145.1 | 84% | 76% | Mental retardation, X-linked syndromic, Fried type, 300630 |
| AP3B1 | 118 | 100% | 100% | Hermansky-Pudlak syndrome 2, 608233 |
| AP4B1 | 108.8 | 100% | 99% | Spastic paraplegia 47, autosomal recessive, 614066 |
| AP4E1 | 146.4 | 100% | 100% | Spastic paraplegia 51, autosomal recessive, 613744 |
| AP4M1 | 111.9 | 100% | 99% | Spastic paraplegia 50, autosomal recessive, 612936 |
| AP4S1 | 71 | 88% | 88% | Spastic paraplegia 52, autosomal recessive, 614067 |
| APTX | 139.9 | 98% | 94% | Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 |
| ARFGEF2 | 116.3 | 100% | 99% | Periventricular heterotopia with microcephaly, 608097 |
| ARG1 | 143.7 | 100% | 99% | Argininemia, 207800 |
| ARHGEF6 | 112.1 | 96% | 95% | Mental retardation, X-linked 46, 300436 |
| ARHGEF9 | 103.2 | 100% | 98% | Epileptic encephalopathy, early infantile, 8, 300607 |
| ARID1A | 116 | 99% | 97% | Mental retardation, autosomal dominant 14, 614607 |
| ARID1B | 125.9 | 100% | 99% | Mental retardation, autosomal dominant 12, 614562 |
| ARID2 | 159.4 | 100% | 100% | No OMIM phenotype |
| ARL13B | 134 | 100% | 95% | Joubert syndrome 8, 612291 |
| ARL6 | 165 | 100% | 100% | Bardet-Biedl syndrome 3, 209900<br>{Bardet-Biedl syndrome 1, modifier of}, 209900<br>Retinitis pigmentosa 55, 613575 |
| ARSE | 96.7 | 98% | 92% | Chondrodysplasia punctata, X-linked recessive, 302950 |
| ARX | 74.3 | 84% | 77% | Epileptic encephalopathy, early infantile, 1, 308350<br>Lissencephaly, X-linked 2, 300215<br>Mental retardation, X-linked 29 and others, 300419<br>Proud syndrome, 300004<br>Partington syndrome, 309510 |
| ASL | 97 | 99% | 97% | Argininosuccinic aciduria, 207900 |
| ASNS | 68.5 | 92% | 86% | Asparagine synthetase deficiency, 615574 |
| ASPA | 119.3 | 100% | 100% | Canavan disease, 271900 |
| ASPM | 146.7 | 100% | 100% | Microcephaly 5, primary, autosomal recessive, 608716 |
| ASXL1 | 160.4 | 98% | 98% | Bohring-Opitz syndrome, 605039<br>Myelodysplastic syndrome, somatic, 614286 |
| ASXL3 | 165.7 | 100% | 99% | Bainbridge-Ropers syndrome, 615485 |
| ATIC | 120.3 | 100% | 99% | AICA-ribosiduria due to ATIC deficiency, 608688 |

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| ATP1A2 | 117.5 | 100% | 98% | Migraine, familial hemiplegic, 2, 602481<br>Alternating hemiplegia of childhood, 104290<br>Migraine, familial basilar, 602481 |
| ATP2A2 | 132.5 | 100% | 100% | Darier disease, 124200<br>Acrokeratosis verruciformis, 101900 |
| ATP6AP2 | 67.4 | 99% | 97% | Mental retardation, X-linked, with epilepsy, 300423 |
| ATP6V0A2 | 117 | 100% | 99% | Cutis laxa, autosomal recessive, type IIA, 219200<br>Wrinkly skin syndrome, 278250 |
| ATP7A | 131.9 | 100% | 100% | Menkes disease, 309400<br>Occipital horn syndrome, 304150<br>Spinal muscular atrophy, distal, X-linked 3, 300489 |
| ATP8A2 | 102.3 | 99% | 99% | ?Cerebellar ataxia, mental retardation and dysequilibrium syndrome 4, 615268 |
| ATR | 131.2 | 100% | 100% | Seckel syndrome 1, 210600<br>Cutaneous telangiectasia and cancer syndrome, familial, 614564 |
| ATRX | 152.6 | 100% | 100% | Alpha-thalassemia/mental retardation syndrome, 301040<br>Alpha-thalassemia myelodysplasia syndrome, somatic, 300448<br>Mental retardation-hypotonic facies syndrome, X-linked, 309580 |
| AUH | 109 | 98% | 93% | 3-methylglutaconic aciduria, type I, 250950 |
| AUTS2 | 133.1 | 100% | 99% | Mental Retardation, autosomal dominant 26, 615834 |
| B3GALTL | 113.6 | 98% | 94% | Peters-plus syndrome, 261540 |
| B4GALT1 | 92.4 | 97% | 97% | Congenital disorder of glycosylation, type IId, 607091 |
| B4GALT7 | 102.3 | 100% | 99% | Ehlers-Danlos syndrome, progeroid type, 1, 130070 |
| BBS1 | 130 | 99% | 99% | Bardet-Biedl syndrome 1, 209900 |
| BBS10 | 133.6 | 100% | 100% | Bardet-Biedl syndrome 10, 209900 |
| BBS12 | 166.4 | 100% | 100% | Bardet-Biedl syndrome 12, 209900 |
| BBS2 | 119.3 | 100% | 100% | Bardet-Biedl syndrome 2, 209900 |
| BBS4 | 109.1 | 100% | 99% | Bardet-Biedl syndrome 4, 209900 |
| BBS5 | 135.5 | 100% | 100% | Bardet-Biedl syndrome 5, 209900 |
| BBS7 | 136.7 | 100% | 100% | Bardet-Biedl syndrome 7, 209900 |
| BBS9 | 130.6 | 100% | 99% | Bardet-Biedl syndrome 9, 209900 |
| BCKDHA | 115.7 | 100% | 100% | Maple syrup urine disease, type Ia, 248600 |
| BCKDHB | 97.4 | 100% | 99% | Maple syrup urine disease, type Ib, 248600 |
| BCL11A | 158.4 | 100% | 100% | No OMIM phenotype |
| BCOR | 140.4 | 100% | 99% | Microphthalmia, syndromic 2, 300166 |

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| BCS1L | 152.6 | 100% | 100% | Mitochondrial complex III deficiency, nuclear type 1, 124000<br>Leigh syndrome, 256000<br>Bjornstad syndrome, 262000<br>GRACILE syndrome, 603358 |
| BLM | 128.3 | 100% | 99% | Bloom syndrome, 210900 |
| BRAF | 86.7 | 100% | 97% | Melanoma, malignant, somatic<br>Colorectal cancer, somatic<br>Adenocarcinoma of lung, somatic, 211980<br>Nonsmall cell lung cancer, somatic<br>Cardiofaciocutaneous syndrome, 115150<br>Noonan syndrome 7, 613706<br>LEOPARD syndrome 3, 613707 |
| BRWD3 | 132.2 | 100% | 99% | Mental retardation, X-linked 93, 300659 |
| BSCL2 | 126.2 | 100% | 100% | Lipodystrophy, congenital generalized, type 2, 269700<br>Silver spastic paraplegia syndrome, 270685<br>Neuropathy, distal hereditary motor, type V, 600794 |
| BTD | 166 | 100% | 100% | Biotinidase deficiency, 253260 |
| BUB1B | 134 | 100% | 99% | Colorectal cancer, somatic, 114500<br>Mosaic variegated aneuploidy syndrome 1, 257300<br>[Premature chromatid separation trait], 176430 |
| C12orf57 | 87.2 | 100% | 99% | Temtamy syndrome, 218340 |
| C12orf65 | 198.2 | 100% | 100% | Combined oxidative phosphorylation deficiency 7, 613559<br>Spastic paraplegia 55,autosomal recessive, 615035 |
| C5orf42 | 133.2 | 100% | 99% | Joubert syndrome 17, 614615 |
| CA2 | 158.3 | 100% | 100% | Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 |
| CA8 | 93.4 | 100% | 100% | Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227 |
| CACNG2 | 118.9 | 100% | 100% | Mental retardation, autosomal dominant 10, 614256 |
| CAMTA1 | 143.4 | 95% | 95% | Cerebellar ataxia, nonprogressive, with mental retardation, 614756 |
| CASK | 108 | 100% | 100% | Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749<br>FG syndrome 4, 300422<br>Mental retardation, with or without nystagmus, 300422 |
| CBL | 133.9 | 100% | 100% | Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 |
| CBS | 91.3 | 100% | 93% | Homocystinuria, B6-responsive and nonresponsive types, 236200<br>Thrombosis, hyperhomocysteinemic, 236200 |

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| CC2D1A | 107 | 100% | 99% | Mental retardation, autosomal recessive 3, 608443 |
| CC2D2A | 100.6 | 98% | 97% | Joubert syndrome 9, 612285<br>Meckel syndrome 6, 612284<br>COACH syndrome, 216360 |
| CCBE1 | 98.9 | 100% | 93% | Hennekam lymphangiectasia-lymphedema syndrome, 235510 |
| CCDC78 | 110.6 | 100% | 100% | Myopathy, centronuclear, 4, 614807 |
| CDH15 | 86 | 100% | 95% | Mental retardation, autosomal dominant 3, 612580 |
| CDK5RAP2 | 113.8 | 99% | 98% | Microcephaly 3, primary, autosomal recessive, 604804 |
| CDKL5 | 145.3 | 100% | 100% | Epileptic encephalopathy, early infantile, 2, 300672<br>Angelman syndrome-like, 105830 |
| CDON | 129.6 | 100% | 100% | Holoprosencephaly 11, 614226 |
| CENPJ | 139.7 | 100% | 100% | Microcephaly 6, primary, autosomal recessive, 608393<br>Seckel syndrome 4, 613676 |
| CEP135 | 136.2 | 99% | 98% | Microcephaly 8, primary, autosomal recessive, 614673 |
| CEP152 | 141.4 | 100% | 100% | Microcephaly 9, primary, autosomal recessive, 614852<br>Seckel syndrome 5, 613823 |
| CEP290 | 105 | 100% | 99% | Joubert syndrome 5, 610188<br>Senior-Loken syndrome 6, 610189<br>Leber congenital amaurosis 10, 611755<br>Meckel syndrome 4, 611134<br>Bardet-Biedl syndrome 14, 209900 |
| CEP41 | 97.8 | 100% | 98% | Joubert syndrome 15, 614464 |
| CHAMP1 | 223 | 100% | 100% | Mental retardation,autosomal dominant 40,616579 |
| CHD2 | 137.9 | 100% | 98% | Epileptic encephalopathy, childhood-onset, 615369 |
| CHD7 | 135.3 | 100% | 99% | CHARGE syndrome, 214800<br>{Scoliosis, idiopathic 3}, 608765<br>Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 |
| CHD8 | 130.6 | 100% | 100% | {Autism, susceptibility to, 18}, 615032 |
| CHKB | 94.6 | 94% | 88% | Muscular dystrophy, congenital, megaconial type, 602541 |
| CLCNKB | 93.2 | 92% | 84% | Bartter syndrome, type 3, 607364<br>Bartter syndrome, type 4b, digenic, 613090 |
| CLIC2 | 76.3 | 99% | 92% | Mental retardation, X-linked, syndromic 32, 300886 |
| CLN3 | 97 | 97% | 96% | Ceroid lipofuscinosis, neuronal, 3, 204200 |
| CLN5 | 142.1 | 98% | 95% | Ceroid lipofuscinosis, neuronal, 5, 256731 |

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| CLN6 | 85.2 | 99% | 91% | Ceroid lipofuscinosis, neuronal, 6, 601780<br>Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 |
| CLN8 | 163.2 | 100% | 100% | Ceroid lipofuscinosis, neuronal, 8, 600143<br>Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 |
| CNKSR2 | 132.4 | 99% | 98% | No OMIM phenotype |
| CNTNAP2 | 121.9 | 100% | 100% | Cortical dysplasia-focal epilepsy syndrome, 610042<br>{Autism susceptibility 15}, 612100<br>Pitt-Hopkins like syndrome 1, 610042 |
| COG1 | 145.5 | 100% | 99% | Congenital disorder of glycosylation, type IIg, 611209 |
| COG6 | 113.4 | 99% | 97% | Congenital disorder of glycosylation, type 2l, 614576<br>Shaheen syndrome, 615328 |
| COG7 | 86.8 | 100% | 98% | Congenital disorder of glycosylation, type IIe, 608779 |
| COG8 | 126.5 | 100% | 100% | Congenital disorder of glycosylation, type IIh, 611182 |
| COL4A1 | 96.1 | 98% | 97% | Porencephaly 1, 175780<br>Brain small vessel disease with hemorrhage, 607595<br>Angiopathy, hereditary, with nephropathy, aneurysms, and muscle, 611773<br>Brain small vessel disease with Axenfeld-Rieger anomaly, 607595 |
| COL4A2 | 90.2 | 100% | 99% | Porencephaly 2, 614483<br>{Hemorrhage, intracerebral, susceptibility to}, 614519 |
| COL4A3BP | 126.3 | 100% | 100% | Mental retardation, autosomal dominant 34, 616351 |
| COLEC11 | 121.2 | 100% | 100% | 3MC syndrome 2, 265050 |
| COQ2 | 79.2 | 97% | 93% | Coenzyme Q10 deficiency, primary, 1, 607426<br>{Multiple system atrophy, susceptibility to}, 146500 |
| COQ4 | 93.1 | 90% | 81% | Coenzyme Q10 deficiency,primary,7,616276 |
| COX10 | 137.6 | 100% | 94% | Leigh syndrome due to mitochondrial COX4 deficiency, 256000<br>Mitochondrial complex IV deficiency, 220110 |
| COX15 | 91.9 | 100% | 98% | Leigh syndrome due to cytochrome c oxidase deficiency, 256000<br>Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 |
| CPS1 | 116.2 | 100% | 99% | Carbamoylphosphate synthetase I deficiency, 237300<br>{Pulmonary hypertension, neonatal, susceptibility to}, 615371<br>{Venoocclusive disease after bone marrow transplantation} |
| CRADD | 122.5 | 82% | 79% | Mental retardation, autosomal recessive 34, 614499 |
| CRBN | 152.6 | 100% | 100% | Mental retardation, autosomal recessive 2, 607417 |
| CREBBP | 93.8 | 100% | 99% | Rubinstein-Taybi syndrome, 180849 |

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| CSNK2A1 | 131.1 | 92% | 83% | No OMIM phenotype |
| CTCF | 122.8 | 100% | 100% | Mental retardation, autosomal dominant 21, 615502 |
| CTDP1 | 92.9 | 89% | 88% | Congenital cataracts, facial dysmorphism, and neuropathy, 604168 |
| CTNNB1 | 135 | 100% | 99% | Mental retardation, autosomal dominant 19, 615075<br>Colorectal cancer, somatic, 114500<br>Hepatocellular carcinoma, somatic, 114550<br>Ovarian cancer, somatic, 167000<br>Pilomatricoma, somatic, 132600 |
| CTNND1 | 101.8 | 99% | 99% | No OMIM phenotype |
| CTSA | 106.6 | 100% | 99% | Galactosialidosis, 256540 |
| CTSD | 113 | 100% | 100% | Ceroid lipofuscinosis, neuronal, 10, 610127 |
| CTTNBP2 | 129.2 | 98% | 96% | No OMIM phenotype |
| CUBN | 96.5 | 99% | 97% | Megaloblastic anemia-1, Finnish type, 261100 |
| CUL4B | 135 | 100% | 99% | Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354 |
| CYB5R3 | 99.1 | 97% | 95% | Methemoglobinemia, type I, 250800<br>Methemoglobinemia, type II, 250800 |
| D2HGDH | 73.3 | 98% | 92% | D-2-hydroxyglutaric aciduria, 600721 |
| DARS2 | 122.4 | 100% | 100% | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 |
| DBT | 117.7 | 100% | 100% | Maple syrup urine disease, type II, 248600 |
| DCAF17 | 110.6 | 100% | 99% | Woodhouse-Sakati syndrome, 241080 |
| DCX | 123 | 100% | 100% | Lissencephaly, X-linked, 300067<br>Subcortical laminal heteropia, X-linked, 300067 |
| DDHD2 | 115 | 100% | 100% | Spastic paraplegia 54, autosomal recessive, 615033 |
| DDX11 | 11.5 | 22% | 14% | Warsaw breakage syndrome, 613398 |
| DDX3X | 130.9 | 100% | 99% | Mental retardation,X-linked 102,300958 |
| DEAF1 | 78.9 | 82% | 81% | Mental retardation, autosomal dominant 24, 615828 |
| DHCR24 | 102.7 | 100% | 99% | Desmosterolosis, 602398 |
| DHCR7 | 136 | 100% | 98% | Smith-Lemli-Opitz syndrome, 270400 |
| DHFR | 55.2 | 85% | 70% | Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 |
| DHTKD1 | 118.4 | 100% | 98% | 2-aminoadipic 2-oxoadipic aciduria, 204750<br>Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 |
| DIAPH1 | 86.8 | 99% | 90% | Deafness,autosomal dominant 1,124900<br>Seizures,cortical blindness,microcephaly syndrome,616632 |
| DIP2B | 108.6 | 100% | 99% | Mental retardation, FRA12A type, 136630 |

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| DKC1 | 104.2 | 100% | 100% | Dyskeratosis congenita, X-linked, 305000 |
| DLD | 150 | 100% | 100% | Dihydrolipoamide dehydrogenase deficiency, 246900 |
| DLG3 | 83.8 | 98% | 94% | Mental retardation, X-linked 90, 300850 |
| DLG4 | 134.2 | 99% | 97% | no OMIM phenotype |
| DMD | 123.2 | 100% | 99% | Duchenne muscular dystrophy, 310200<br>Becker muscular dystrophy, 300376<br>Cardiomyopathy, dilated, 3B, 302045 |
| DMPK | 115.8 | 100% | 99% | Myotonic dystrophy 1, 160900 |
| DNAJC19 | 62.7 | 79% | 79% | 3-methylglutaconic aciduria, type V, 610198 |
| DNM1 | 89.4 | 95% | 85% | Epileptic encephalopathy,early infantile,31,616346 |
| DNMT3A | 91.1 | 100% | 98% | Tatton-Brown-Rahman syndrome, 615879 |
| DNMT3B | 104.1 | 100% | 99% | Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 |
| DOCK8 | 95.7 | 100% | 99% | Mental retardation, autosomal dominant 2, 614113<br>Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 |
| DPAGT1 | 108.6 | 99% | 96% | Congenital disorder of glycosylation, type Ij, 608093<br>Myasthenic syndrome, congenital, with tubular aggregates 2, 614750 |
| DPM1 | 159.7 | 90% | 90% | Congenital disorder of glycosylation, type Ie, 608799 |
| DPP6 | 113 | 95% | 92% | Mental retardation, autosomal dominant 33, 616311<br>{Ventricular fibrillation, paroxysmal familial, 2} |
| DPYD | 129.2 | 100% | 99% | Dihydropyrimidine dehydrogenase deficiency, 274270<br>5-fluorouracil toxicity, 274270 |
| DST | 163.1 | 100% | 99% | Neuropathy, hereditary sensory and autonomic, type VI, 614653<br>Epidermolysis bullosa simplex, sutosomal recessive 2, 615425 |
| DYM | 108.5 | 97% | 97% | Dyggve-Melchior-Clausen disease, 223800<br>Smith-McCort dysplasia, 607326<br>Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388 |
| DYNC1H1 | 130.6 | 99% | 97% | Charcot-Marie-Tooth disease, axonal, type 20, 614228<br>Mental retardation, autosomal dominant 13, 614563<br>Spinal muscular atrophy, lower extremity-predominant, AD, 158600 |
| DYRK1A | 147.9 | 100% | 98% | Mental retardation, autosomal dominant 7, 614104 |
| EBP | 83.1 | 99% | 92% | Chondrodysplasia punctata, X-linked dominant, 302960 |
| EDC3 | 123.7 | 99% | 98% | ?Mental retardation, autosomal recessive 50,616460 |
| EEF1A2 | 118 | 100% | 98% | Epileptic encephalopathy, early infantile,33,616409<br>Mental retardation,autosomal dominant 38,616393 |

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| EFTUD2 | 99.6 | 100% | 98% | Mandibulofacial dysostosis, Guion-Almeida type, 610536 |
| EHMT1 | 111.5 | 98% | 96% | Kleefstra syndrome, 610253 |
| EIF2AK3 | 122.2 | 94% | 92% | Wolcott-Rallison syndrome, 226980 |
| EIF4G1 | 116.6 | 100% | 100% | Parkinsons disease 18, 614251 |
| ELOVL4 | 114.8 | 100% | 100% | Stargardt disease 3, 600110<br>Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110<br>Ichthyosis, spastic quadriplegia, and mental retardation, 614457 |
| EMX2 | 129.5 | 100% | 100% | Schizencephaly, 269160 |
| EP300 | 156.1 | 99% | 99% | Colorectal cancer, somatic, 114500<br>Rubinstein-Taybi syndrome 2, 613684 |
| EPB41L1 | 103.9 | 99% | 97% | Mental retardation, autosomal dominant 11, 614257 |
| ERCC2 | 97.5 | 98% | 95% | Xeroderma pigmentosum, group D, 278730<br>Trichothiodystrophy, 601675<br>Cerebrooculofacioskeletal syndrome 2, 610756 |
| ERCC3 | 136.8 | 100% | 100% | Xeroderma pigmentosum, group B, 610651<br>Trichothiodystrophy, 601675 |
| ERCC5 | 137.2 | 99% | 97% | Xeroderma pigmentosum, group G, 278780<br>Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 |
| ERCC6 | 164 | 100% | 98% | Cockayne syndrome, type B, 133540<br>Cerebrooculofacioskeletal syndrome 1, 214150<br>De Sanctis-Cacchione syndrome, 278800<br>{Macular degeneration, age-related, susceptibility to 5}, 613761<br>UV-sensitive syndrome 1, 600630 |
| ERCC8 | 96.9 | 100% | 100% | Cockayne syndrome, type A, 216400<br>UV-sensitive syndrome 2, 614621 |
| ERLIN2 | 128.9 | 100% | 100% | Spastic paraplegia 18, autosomal recessive, 611225 |
| ESCO2 | 91.2 | 100% | 99% | Roberts syndrome, 268300<br>SC phocomelia syndrome, 269000 |
| ETFB | 127.3 | 100% | 100% | Glutaric acidemia 2B, 231680 |
| ETHE1 | 68.4 | 97% | 93% | Ethylmalonic encephalopathy, 602473 |
| EXOSC3 | 54.4 | 89% | 69% | Pontocerebellar hypoplasia, type 1B, 614678 |
| EZH2 | 91.2 | 99% | 93% | Weaver syndrome, 277590 |
| FAM126A | 148.8 | 100% | 100% | Leukodystrophy, hypomyelinating, 5, 610532 |

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|---|---|---|---|---|
| FBN1 | 108.4 | 100% | 99% | Marfan syndrome, 154700<br>Ectopia lentis, familial, 129600<br>MASS syndrome, 604308<br>Weill-Marchesani syndrome 2, dominant, 608328<br>Aortic aneurysm, ascending, and dissection<br>Stiff skin syndrome, 184900<br>Acromicric dysplasia, 102370 |
| FBXO31 | 66.2 | 100% | 97% | Mental retardation, autosomal recessive 45, 615979 |
| FGD1 | 104.3 | 100% | 98% | Aarskog-Scott syndrome, 305400<br>Mental retardation, X-linked syndromic 16, 305400 |
| FGFR1 | 129.9 | 100% | 94% | Pfeiffer syndrome, 101600<br>Jackson-Weiss syndrome, 123150<br>Hypogonadotropic hypogonadism 2 with or without anosmia, 147950<br>Osteoglophonic dysplasia, 166250<br>Trigonocephaly 1, 190440<br>Hartsfield syndrome, 615465 |
| FGFR2 | 134.1 | 97% | 97% | Crouzon syndrome, 123500<br>Jackson-Weiss syndrome, 123150<br>Beare-Stevenson cutis gyrata syndrome, 123790<br>Pfeiffer syndrome, 101600<br>Apert syndrome, 101200<br>Saethre-Chotzen syndrome, 101400<br>Craniosynostosis, nonspecific |
| FGFR3 | 88 | 94% | 92% | Achondroplasia, 100800<br>Hypochondroplasia, 146000<br>Thanatophoric dysplasia, type I, 187600<br>Crouzon syndrome with acanthosis nigricans, 612247<br>Muenke syndrome, 602849<br>Bladder cancer, somatic, 109800<br>Colorectal cancer, somatic, 1 |
| FH | 102.8 | 99% | 92% | Fumarase deficiency, 606812<br>Leiomyomatosis and renal cell cancer, 150800 |
| FIGN | 160.6 | 100% | 100% | No OMIM phenotype |
| FKRP | 100.8 | 100% | 99% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153<br>Muscular dystrophy-dystroglycanopathy (congenital w/wo mental retardation), type B, 5, 606612 |

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| FKTN | 118.6 | 100% | 100% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800<br>Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152<br>Cardiomyopathy, dilated, 1X, 611615 |
| FLNA | 138 | 100% | 100% | Heterotopia, periventricular, 300049<br>Otopalatodigital syndrome, type I, 311300<br>Otopalatodigital syndrome, type II, 304120<br>Intestinal pseudoobstruction, neuronal, 300048<br>Melnick-Needles syndrome, 309350 |
| FLVCR1 | 105.5 | 100% | 100% | Ataxia, posterior column, with retinitis pigmentosa, 609033 |
| FMN2 | 124.1 | 91% | 86% | Mental retardation, autosomal recessive 47, 616193 |
| FMR1 | 115.6 | 100% | 99% | Fragile X syndrome, 300624<br>Fragile X tremor/ataxia syndrome, 300623<br>Premature ovarian failure 1, 311360 |
| FOXG1 | 94 | 86% | 80% | Rett syndrome, congenital variant, 613454 |
| FOXP1 | 119 | 100% | 100% | Mental retardation with language impairment and autistic features, 613670 |
| FOXP2 | 119.6 | 100% | 100% | Speech-language disorder-1, 602081 |
| FRAS1 | 107.7 | 98% | 96% | Fraser syndrome, 219000 |
| FTO | 134.7 | 100% | 98% | Growth retardation, developmental delay, coarse facies, and early death, 612938 |
| FTSJ1 | 103.6 | 97% | 91% | Mental retardation, X-linked 9, 309549 |
| FUCA1 | 89.4 | 100% | 97% | Fucosidosis, 230000 |
| GABRA1 | 129.3 | 100% | 95% | Epileptic encephalopathy, early infantile, 19, 615744<br>{Epilepsy, childhood absence, susceptibility to, 4}<br>{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 |
| GAD1 | 103.3 | 100% | 100% | Cerebral palsy, spastic quadriplegic, 1, 603513 |
| GALE | 122.2 | 100% | 100% | Galactose epimerase deficiency, 230350 |
| GALT | 123.5 | 100% | 100% | Galactosemia, 230400 |
| GAMT | 122.6 | 100% | 94% | Cerebral creatine deficiency syndrome 2, 612736 |
| GATAD2B | 125.7 | 100% | 99% | Mental retardation, autosomal dominant 18, 615074 |
| GATM | 94.5 | 98% | 95% | Cerebral creatine deficiency syndrome 3, 612718 |
| GCH1 | 121.5 | 100% | 100% | Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230<br>Hyperphenylalaninemia, BH4-deficient, B, 233910 |
| GCSH | 18.1 | 49% | 39% | Glycine encephalopathy, 605899 |
| GDI1 | 159.7 | 100% | 100% | Mental retardation, X-linked 41, 300849 |
| GFAP | 88.5 | 100% | 95% | Alexander disease, 203450 |

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| GJB1 | 182.5 | 100% | 100% | Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 |
| GJC2 | 63.9 | 97% | 87% | Leukodystrophy, hypomyelinating, 2, 608804<br>Spastic paraplegia 44, autosomal recessive, 613206<br>Lymphedema, hereditary, IC, 613480 |
| GK | 52.7 | 84% | 82% | Glycerol kinase deficiency, 307030 |
| GLB1 | 87.6 | 100% | 97% | GM1-gangliosidosis, type I, 230500<br>GM1-gangliosidosis, type II, 230600<br>GM1-gangliosidosis, type III, 230650<br>Mucopolysaccharidosis type IVB (Morquio), 253010 |
| GLDC | 63.9 | 98% | 83% | Glycine encephalopathy, 605899 |
| GLI2 | 127.3 | 100% | 98% | Holoprosencephaly-9, 610829 |
| GLI3 | 131.8 | 100% | 99% | Greig cephalopolysyndactyly syndrome, 175700<br>Pallister-Hall syndrome, 146510<br>Polydactyly, preaxial, type IV, 174700<br>Polydactyly, postaxial, types A1 and B, 174200<br>{Hypothalamic hamartomas, somatic}, 241800 |
| GM2A | 118.6 | 100% | 100% | GM2-gangliosidosis, AB variant, 272750 |
| GMPPA | 138.9 | 100% | 100% | Alacrima, achalasia and mental retardation syndrome, 615510 |
| GMPPB | 134.2 | 100% | 100% | Muscular dystrophy-dystroglycanopathy (with brain and eye anomalies), type A,14, 6135350<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 |
| GNAO1 | 125.8 | 100% | 100% | Epileptic encephalopathy, early infantile, 17, 615473 |
| GNAS | 141 | 99% | 97% | Pseudohypoparathyroidism Ia, 103580<br>McCune-Albright syndrome, 174800<br>Pseudohypoparathyroidism Ic, 612462<br>Osseous heteroplasia, progressive, 166350<br>Pseudohypoparathyroidism Ib, 603233 |
| GNPAT | 131.8 | 100% | 100% | Chondrodysplasia punctata, rhizomelic, type 2, 222765 |
| GNS | 87.3 | 89% | 87% | Mucopolysaccharidosis type IIID, 252940 |
| GPC3 | 112 | 100% | 100% | Simpson-Golabi-Behmel syndrome, type 1, 312870<br>Wilms tumor, somatic, 194070 |
| GPHN | 125.5 | 100% | 100% | Molybdenum cofactor deficiency, type C, 252150 |
| GPR56 | 113.7 | 100% | 99% | Polymicrogyria, bilateral frontoparietal, 606854 |
| GPT2 | 98.9 | 100% | 89% | ?Mental retardation, autosomal recessive 49, 616281 |
| GRIA3 | 115.4 | 100% | 98% | Mental retardation, X-linked 94, 300699 |

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| GRID2 | 145.5 | 100% | 100% | Spinocerebellar ataxia, autosomal recessive 18, 616204 |
| GRIK2 | 121.2 | 96% | 96% | Mental retardation, autosomal recessive, 6, 611092 |
| GRIN1 | 98.6 | 100% | 99% | Mental retardation, autosomal dominant 8, 614254 |
| GRIN2A | 155.6 | 100% | 99% | Epilepsy with neurodevelopmental defects, 613971 |
| GRIN2B | 155.1 | 100% | 99% | Mental retardation, autosomal dominant 6, 613970 |
| GRIN3B | 73.3 | 85% | 72% | No OMIM phenotype |
| GRM1 | 152.9 | 100% | 99% | Spinocerebellar ataxia, autosomal recessive 13, 614831 |
| GSE1 | 95.8 | 96% | 93% | No OMIM phenotype |
| GSS | 96 | 100% | 98% | Hemolytic anemia due to glutathione synthetase deficiency, 231900 |
| GTF2H5 | 92.1 | 100% | 100% | Trichothiodystrophy, complementation group A, 601675 |
| GUSB | 72.4 | 91% | 86% | Mucopolysaccharidosis VII, 253220 |
| HAX1 | 155.1 | 100% | 100% | Neutropenia, severe congenital 3, autosomal recessive, 610738 |
| HCCS | 127.9 | 100% | 99% | Microphthalmia, syndromic 7, 309801 |
| HCFC1 | 85.9 | 99% | 94% | Mental retardation, X-linked 3, 309541 |
| HCN1 | 114.3 | 100% | 100% | Epileptic encephalopathy, early infantile, 24, 615871 |
| HDAC4 | 80.9 | 91% | 89% | Brachydactyly-mental retardation syndrome, 600430 |
| HDAC6 | 136.9 | 99% | 96% | Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863 |
| HDAC8 | 114 | 100% | 99% | Wilson-Turner syndrome, 309585<br>Cornelia de Lange syndrome 5, 300882 |
| HECTD1 | 138.8 | 100% | 99% | No OMIM phenotype |
| HERC1 | 124.2 | 100% | 98% | No OMIM phenotype |
| HERC2 | 70.4 | 63% | 60% | Mental retardation, autosomal recessive 38, 615516<br>[Skin/hair/eye pigmentation 1, blond/brown hair], 227220<br>[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 |
| HESX1 | 111 | 100% | 98% | Septooptic dysplasia, 182230<br>Pituitary hormone deficiency, combined, 5, 182230<br>Growth hormone deficiency with pituitary anomalies, 182230 |
| HEXA | 111 | 100% | 100% | Tay-Sachs disease, 272800<br>GM2-gangliosidosis, several forms, 272800<br>[Hex A pseudodeficiency], 272800 |
| HEXB | 121.1 | 100% | 100% | Sandhoff disease, infantile, juvenile, and adult forms, 268800 |
| HIVEP2 | 180.5 | 100% | 100% | No OMIM phenotype |
| HLCS | 153.3 | 100% | 100% | Holocarboxylase synthetase deficiency, 253270 |

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| HOXA1 | 144.6 | 100% | 100% | Bosley-Salih-Alorainy syndrome, 601536<br>Athabaskan brainstem dysgenesis syndrome, 601536 |
| HPD | 113.9 | 100% | 99% | Tyrosinemia, type III, 276710<br>Hawkinsinuria, 140350 |
| HPRT1 | 98.1 | 100% | 98% | Lesch-Nyhan syndrome, 300322 |
| HRAS | 120.2 | 100% | 100% | {Bladder cancer, somatic}, 109800<br>Costello syndrome, 218040<br>{Thyroid carcinoma, follicular, somatic}, 188470<br>{Nevus sebaceous, somatic}, 162900 |
| HSD17B10 | 120.2 | 99% | 99% | 17-beta-hydroxysteroid dehydrogenase X deficiency, 300438<br>Mental retardation, X-linked syndromic 10, 300220<br>Mental retardation, X-linked 17/31, microduplication, 300705 |
| HSPD1 | 16 | 62% | 38% | Spastic paraplegia 13, autosomal dominant, 605280<br>Leukodystrophy, hypomyelinating, 4, 612233 |
| HUWE1 | 107.5 | 100% | 99% | Mental retardation, X-linked syndromic, Turner type, 300706 |
| IDS | 121.5 | 91% | 87% | Mucopolysaccharidosis II, 309900 |
| IDUA | 101.7 | 95% | 91% | Mucopolysaccharidosis Ih, 607014<br>Mucopolysaccharidosis Is, 607016<br>Mucopolysaccharidosis Ih/s, 607015 |
| IER3IP1 | 71.3 | 100% | 99% | Microcephaly, epilepsy, and diabetes syndrome, 614231 |
| IFT172 | 109.4 | 100% | 97% | Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 |
| IGBP1 | 107.1 | 96% | 89% | Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472 |
| IGF1 | 147.1 | 100% | 100% | Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747 |
| IKBKG | 31.4 | 26% | 26% | Incontinentia pigmenti, type II, 308300<br>Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291<br>Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301<br>Immunodeficiency, isolated, 300584 |
| IL1RAPL1 | 146.5 | 100% | 100% | Mental retardation, X-linked 21/34, 300143 |
| INPP5E | 93.8 | 100% | 99% | Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156<br>Joubert syndrome 1, 213300 |
| IQSEC2 | 91.5 | 95% | 90% | Mental retardation, X-linked 1, 309530 |
| ISPD | 96.8 | 97% | 95% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 |
| ITPR1 | 114.8 | 100% | 98% | Spinocerebellar ataxia 15, 606658<br>Spinocerebellar ataxia 29, congenital nonprogressive, 117360 |

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| JAG1 | 108.4 | 100% | 97% | Alagille syndrome, 118450<br>Deafness, congenital heart defects and posterior embryotoxon<br>Tetralogy of Fallot, 187500 |
| JAM3 | 76.5 | 99% | 90% | Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 |
| KANK1 | 164.1 | 100% | 100% | Cerebral palsy, spastic quadriplegic, 2, 612900 |
| KANSL1 | 69.8 | 94% | 87% | Koolen-De Vries syndrome, 610443 |
| KAT6A | 142.7 | 100% | 99% | Mental retardation, autosomal dominant 32, 616268 |
| KAT6B | 157.8 | 100% | 100% | SBBYSS syndrome, 603736<br>Genitopatellar syndrome, 606170 |
| KCNC3 | 78.4 | 80% | 70% | Spinocerebellar ataxia 13,605259 |
| KCNH1 | 128.3 | 100% | 99% | Temple-Baraitser syndrome, 611816<br>Zimmermann-Laband syndrome 1, 135500 |
| KCNJ10 | 176.9 | 100% | 100% | SESAME syndrome, 612780<br>Enlarged vestibular aqueduct, digenic, 600791 |
| KCNJ11 | 147.9 | 100% | 100% | Hyperinsulinemic hypoglycemia, familial, 2, 601820<br>Diabetes, permanent neonatal, 606176<br>Diabetes mellitus, permanent neonatal, with neurologic features, 606176<br>{Diabetes mellitus, type 2, susceptibility to}, 125853 |
| KCNK9 | 137.5 | 100% | 100% | Birk-Barel mental retardation dysmorphism syndrome, 612292 |
| KCNQ2 | 80.4 | 98% | 96% | Seizures, benign neonatal, 1, 121200<br>Myokymia, 121200<br>Epileptic encephalopathy, early infantile, 7, 613720 |
| KCNQ5 | 147.4 | 99% | 97% | No OMIM phenotype |
| KCNT1 | 87.2 | 96% | 94% | Epileptic encephalopathy, early infantile, 14, 614959<br>Epilepsy, nocturnal frontal lobe, 5, 615005 |
| KCTD7 | 93.2 | 70% | 69% | Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 |
| KDM5C | 130.2 | 100% | 100% | Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 |
| KDM6A | 134 | 100% | 99% | Kabuki syndrome 2, 300867 |
| KIAA0226 | 102.7 | 99% | 97% | ?Spinocerebellar ataxia, autosomal recessive 15, 615705 |
| KIAA1033 | 121.9 | 100% | 98% | ?Mental retardation, autosomal recessive 43, 615817 |
| KIAA1109 | 136.7 | 100% | 99% | no OMIM phenotype |
| KIAA1279 | 125.6 | 100% | 98% | Goldberg-Shprintzen megacolon syndrome, 609460 |
| KIAA2022 | 181.6 | 100% | 100% | Mental retardation, X-linked 98, 300912 |
| KIF11 | 108.9 | 100% | 100% | Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 |

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| KIF1A | 79.4 | 99% | 94% | Spastic paraplegia 30, autosomal recessive, 610357<br>Neuropathy, hereditary sensory, type IIC, 614213<br>Mental retardation, autosomal dominant 9, 614255 |
| KIF4A | 143 | 97% | 94% | ?Mental retardation,X-linked 100,300923 |
| KIF5C | 92 | 99% | 92% | Cortical dysplasia, complex, with other brain malformations 2, 615282 |
| KIF7 | 80.9 | 93% | 86% | Hydrolethalus syndrome 2, 614120<br>Acrocallosal syndrome, 200990<br>Joubert syndrome 12, 200990 |
| KIRREL3 | 83.3 | 100% | 97% | Mental retardation, autosomal dominant 4, 612581 |
| KMT2A | 152.4 | 99% | 98% | Leukemia, myeloid/lymphoid or mixed-lineage, 159555<br>Wiedemann-Steiner syndrome, 605130 |
| KMT2D | 118.1 | 99% | 98% | Kabuki syndrome 1, 147920 |
| KPTN | 100.5 | 100% | 100% | Mental retardation, autosomal recessive 41, 615637 |
| KRAS | 69.8 | 97% | 92% | Noonan syndrome 3, 609942<br>Bladder cancer, somatic, 109800<br>Breast cancer, somatic, 114480<br>Cardiofaciocutaneous syndrome 2, 615278<br>Gastric cancer, somatic, 137215<br>Lung cancer, somatic, 211980<br>Pancreatic carcinoma, somatic, 260350 |
| KRBOX4 | 141.5 | 100% | 100% | No OMIM phenotype<br>nonsyndromic X-linked mental retardation (Lugtenberg et al. 2006) |
| L1CAM | 146.8 | 100% | 100% | Hydrocephalus due to aqueductal stenosis, 307000<br>MASA syndrome, 303350<br>CRASH syndrome, 303350<br>Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 |
| L2HGDH | 83.2 | 94% | 91% | L-2-hydroxyglutaric aciduria, 236792 |
| LAMA1 | 104.1 | 99% | 96% | Poretti-Boltshauser syndrome, 615960 |
| LAMA2 | 108.4 | 100% | 98% | Muscular dystrophy, congenital merosin-deficient, 607855<br>Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855 |
| LAMC3 | 113 | 99% | 97% | Cortical malformations, occipital, 614115 |
| LAMP2 | 124.5 | 100% | 99% | Danon disease, 300257 |
| LARGE | 111 | 98% | 96% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 |

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| LARP7 | 122.5 | 100% | 100% | Alazami syndrome, 615071 |
| LIG4 | 188.1 | 100% | 100% | LIG4 syndrome, 606593<br>{Multiple myeloma, resistance to}, 254500<br>Severe combined immunodeficiency with sensitivity to ionizing radiation, 602450 |
| LINS | 124.7 | 100% | 100% | Mental retardation, autosomal recessive 27, 614340 |
| LRP2 | 115.9 | 100% | 99% | Donnai-Barrow syndrome, 222448 |
| LRPPRC | 106.5 | 98% | 97% | Leigh syndrome, French-Canadian type, 220111 |
| MAGEL2 | 147.9 | 100% | 100% | Prader-Willi-like syndrome, 615547 |
| MAGT1 | 114 | 98% | 98% | Mental retardation, X-linked 95, 300716<br>Immunodeficiency, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 |
| MAN1B1 | 108.4 | 100% | 99% | Mental retardation, autosomal recessive 15, 614202 |
| MAN2B1 | 91 | 99% | 94% | Mannosidosis, alpha-, types I and II, 248500 |
| MANBA | 102.6 | 100% | 99% | Mannosidosis, beta, 248510 |
| MAOA | 114.4 | 100% | 100% | Brunner syndrome, 300615 |
| MAP2K1 | 110 | 93% | 80% | Cardiofaciocutaneous syndrome 3, 615279 |
| MAP2K2 | 128.3 | 100% | 99% | Cardiofaciocutaneous syndrome 4, 615280 |
| MAT1A | 99.9 | 100% | 98% | Hypermethioninemia, persistent,due to methionine adenosyltransferase I/III deficiency, 250850<br>Methionine adenosyltransferase deficiency, autosomal recessive, 250850 |
| MBD5 | 155.7 | 100% | 100% | Mental retardation, autosomal dominant 1, 156200 |
| MBTPS2 | 152.1 | 100% | 100% | IFAP syndrome with or without BRESHECK syndrome, 308205<br>Keratosis follicularis spinulosa decalvans, X-linked, 308800 |
| MCCC1 | 107.4 | 100% | 98% | 3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 |
| MCCC2 | 123.8 | 94% | 92% | 3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 |
| MCOLN1 | 111.4 | 98% | 92% | Mucolipidosis IV, 252650 |
| MCPH1 | 136.8 | 100% | 100% | Microcephaly 1, primary, autosomal recessive, 251200 |
| MECP2 | 204.3 | 100% | 99% | Rett syndrome, 312750<br>Mental retardation, X-linked, syndromic 13, 300055<br>Rett syndrome, preserved speech variant, 312750<br>Encephalopathy, neonatal severe, 300673<br>{Autism susceptibility, X-linked 3}, 300496<br>Angelman syndrome, 105830 |
| MED12 | 137.1 | 99% | 96% | Opitz-Kaveggia syndrome, 305450<br>Lujan-Fryns syndrome, 309520<br>Ohdo syndrome, X-linked, 300895 |

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| MED13L | 129.5 | 99% | 98% | Transposition of the great arteries, dextro-looped 1, 608808 |
| MED17 | 157.1 | 100% | 100% | Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 |
| MED23 | 127.3 | 99% | 99% | Mental retardation, autosomal recessive 18, 614249 |
| MEF2C | 121 | 100% | 99% | Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443<br>Chromosome 5q14.3 deletion syndrome, 613443 |
| METTL23 | 191.6 | 100% | 100% | Mental retardation, autosomal recessive 44, 615942 |
| MGAT2 | 239.8 | 100% | 100% | Congenital disorder of glycosylation, type IIa, 212066 |
| MID1 | 166 | 100% | 100% | Opitz GBBB syndrome, type I, 300000 |
| MID2 | 136.5 | 99% | 99% | ?Mental retardation,X-linked 101,300928 |
| MKKS | 132.7 | 89% | 89% | McKusick-Kaufman syndrome, 236700<br>Bardet-Biedl syndrome 6, 209900 |
| MLYCD | 84.4 | 94% | 89% | Malonyl-CoA decarboxylase deficiency, 248360 |
| MMAA | 187.9 | 100% | 100% | Methylmalonic aciduria, vitamin B12-responsive, 251100 |
| MMACHC | 186 | 100% | 100% | Methylmalonic aciduria and homocystinuria, cblC type, 277400 |
| MMADHC | 70.5 | 89% | 89% | Homocystinuria, cbID type, 277410 |
| MOCS1 | 93.5 | 98% | 96% | Molybdenum cofactor deficiency, type A, 252150 |
| MOCS2 | 131.3 | 99% | 99% | Molybdenum cofactor deficiency, type B, 252150 |
| MOGS | 153.5 | 100% | 100% | Congenital disorder of gycosylation, type 2b, 606056 |
| MPDU1 | 136.5 | 100% | 100% | Congenital disorder of glycosylation, type If, 609180 |
| MPDZ | 119.3 | 97% | 97% | Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219 |
| MPLKIP | 98.6 | 100% | 100% | Trichothiodystrophy, nonphotosensitive 1, 234050 |
| MRPS22 | 109 | 100% | 100% | Combined oxidative phosphorylation deficiency 5, 611719 |
| MTHFR | 107.1 | 100% | 99% | Homocystinuria due to MTHFR deficiency, 236250<br>{Neural tube defects, susceptibility to}, 601634<br>{Schizophrenia, susceptibility to}, 181500<br>{Thromboembolism, susceptibility to}, 188050 |
| MTR | 116.6 | 99% | 98% | Homocystinuria-megaloblastic anemia, cblG complementation type, 250940<br>{Neural tube defects, folate-sensitive, susceptibility to}, 601634 |
| MTRR | 119.3 | 100% | 100% | Homocystinuria-megaloblastic anemia, cbl E type, 236270 |
| MUT | 130.4 | 100% | 100% | Methylmalonic aciduria, mut(0) type, 251000 |
| MVK | 99.7 | 100% | 100% | Mevalonic aciduria, 610377<br>Hyper-IgD syndrome, 260920<br>Porokeratosis 3, disseminated superficial actinic, 175900 |
| MYCN | 136.6 | 100% | 96% | Feingold syndrome, 164280 |

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| MYH9 | 106.8 | 100% | 99% | Deafness, autosomal dominant 17, 603622<br>Epstein syndrome, 153650<br>Fechtner syndrome, 153640<br>Macrothrombocytopenia and progressive sensorineural deafness, 600208<br>May-Hegglin anomaly, 155100<br>Sebastian syndrome, 605249 |
| MYO5A | 103.9 | 99% | 98% | Griscelli syndrome, type 1, 214450 |
| MYT1L | 113.5 | 99% | 98% | Mental retardation,autosomal dominant 39,616521 |
| NAA10 | 115.8 | 97% | 97% | N-terminal acetyltransferase deficiency, 300855 |
| NAGA | 86.2 | 100% | 99% | Schindler disease, type I, 609241<br>Kanzaki disease, 609242<br>Schindler disease, type III, 609241 |
| NAGLU | 80.6 | 96% | 91% | Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 |
| NALCN | 113.2 | 99% | 97% | ?Neuroaxonal neurodegeneration, infantile, with facial dysmophism, 615419 |
| NBN | 129.6 | 99% | 97% | Aplastic anemia, 609135<br>Leukemia, acute lymphoblastic, 613065<br>Nijmegen breakage syndrome, 251260 |
| NDE1 | 109.1 | 100% | 99% | Lissencephaly 4 (with microcephaly), 614019 |
| NDP | 106.7 | 99% | 94% | Norrie disease, 310600<br>Exudative vitreoretinopathy, X-linked, 305390 |
| NDST1 | 116.1 | 100% | 100% | Mental retardation, autosomal recessive 46, 616116 |
| NDUFA1 | 216.5 | 100% | 100% | Mitochondrial complex I deficiency, 252010 |
| NDUFA11 | 127.5 | 99% | 95% | Mitochondrial complex I deficiency, 252010 |
| NDUFA12 | 89.3 | 100% | 100% | Leigh syndrome due to mitochondrial complex 1 deficiency, 256000 |
| NDUFS1 | 91.4 | 100% | 100% | Mitochondrial complex I deficiency, 252010 |
| NDUFS2 | 132.2 | 100% | 100% | Mitochondrial complex I deficiency, 252010 |
| NDUFS3 | 154.6 | 98% | 91% | Leigh syndrome due to mitochondrial complex I deficiency, 256000<br>Mitochondrial complex I deficiency, 252010 |
| NDUFS4 | 142.4 | 100% | 100% | Leigh syndrome, 256000<br>Mitochondrial complex I deficiency, 252010 |
| NDUFS7 | 121.2 | 100% | 100% | Leigh syndrome, 256000 |
| NDUFS8 | 123.4 | 100% | 99% | Leigh syndrome due to mitochondrial complex I deficiency, 256000 |
| NDUFV1 | 68.1 | 98% | 93% | Mitochondrial complex I deficiency, 252010 |
| NEDD4L | 119.9 | 100% | 99% | No OMIM phenotype |

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|---|---|---|---|---|
| NEU1 | 17.8 | 65% | 41% | Sialidosis, type I, 256550<br>Sialidosis, type II, 256550 |
| NF1 | 89.9 | 83% | 82% | Neurofibromatosis, type 1, 162200<br>Leukemia, juvenile myelomonocytic, 607785<br>Melanoma, desmoplastic neurotrophic (2)<br>Neurofibromatosis, familial spinal, 162210<br>Neurofibromatosis-Noonan syndrome, 601321<br>Watson syndrome, 193520 |
| NFATC1 | 110.6 | 100% | 100% | No OMIM phenotype |
| NFIA | 126 | 100% | 100% | No OMIM phenotype |
| NFIX | 145.5 | 98% | 98% | Marshall-Smith syndrome, 602535<br>Sotos syndrome 2, 614753 |
| NHS | 140.2 | 95% | 94% | Nance-Horan syndrome, 302350<br>Cataract 40, X-linked, 302200 |
| NIPBL | 129.7 | 98% | 98% | Cornelia de Lange syndrome 1, 122470 |
| NKX2-1 | 110.7 | 100% | 99% | Goiter, familial, due to TTF-1 defect (1)<br>Chorea, hereditary benign, 118700<br>Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 |
| NLGN3 | 143.5 | 100% | 100% | {Asperger syndrome susceptibility, X-linked 1}, 300494<br>{Autism susceptibility, X-linked 1}, 300425 |
| NLGN4X | 62.1 | 76% | 68% | Mental retardation, X-linked, 300495<br>{Asperger syndrome susceptibility, X-linked 2}, 300497 |
| NLRP3 | 130.8 | 100% | 100% | Cold-induced autoinflammatory syndrome, familial, 120100<br>Muckle-Wells syndrome, 191900<br>CINCA syndrome, 607115 |
| NPHP1 | 123.1 | 100% | 100% | hronophthisis 1, juvenile, 256100<br>Senior-Loken syndrome-1, 266900<br>Joubert syndrome 4, 609583 |
| NR2F1 | 174.2 | 100% | 100% | Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 |
| NRAS | 143.9 | 100% | 100% | Autoimmune lymphoproliferative syndrome type IV, 614470<br>Noonan syndrome 6, 613224<br>Epidermal nevus, somatic, 162900<br>Thyroid carcinoma, follicular, somatic, 188470<br>Colorectal cancer, somatic, 114500 |

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|---|---|---|---|---|
| NRXN1 | 132.9 | 100% | 97% | Pitt-Hopkins-like syndrome 2, 614325<br>{Schizophrenia, susceptibility to, 17}, 614332 |
| NSD1 | 137.6 | 100% | 99% | Sotos syndrome 1, 117550<br>Leukemia, acute myeloid, 601626 (1)<br>Beckwith-Wiedemann syndrome, 130650 |
| NSDHL | 113.8 | 100% | 98% | CHILD syndrome, 308050<br>CK syndrome, 300831 |
| NSUN2 | 131.8 | 100% | 97% | Mental retardation, autosomal recessive 5, 611091 |
| NTRK1 | 77.3 | 99% | 95% | Insensitivity to pain, congenital, with anhidrosis, 256800<br>Medullary thyroid carcinoma, familial, 155240 |
| OCLN | 98.5 | 72% | 72% | Band-like calcification with simplified gyration and polymicrogyria, 251290 |
| OCRL | 133.1 | 100% | 97% | Lowe syndrome, 309000<br>Dent disease 2, 300555 |
| ODC1 | 135.7 | 100% | 100% | {Colonic adenoma recurrence,reduced risk of},114500 |
| OFD1 | 78.7 | 98% | 91% | Oral-facial-digital syndrome 1, 311200<br>Simpson-Golabi-Behmel syndrome, type 2, 300209<br>Joubert syndrome 10, 300804 |
| OPHN1 | 113.8 | 100% | 99% | Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486 |
| ORC1 | 113.1 | 100% | 98% | Meier-Gorlin syndrome 1, 224690 |
| OTC | 113.1 | 100% | 99% | CGD Ornithine transcarbamylase deficiency, 311250 |
| PACS1 | 120.7 | 100% | 98% | Mental retardation, autosomal dominant 17, 615009 |
| PAFAH1B1 | 81.6 | 92% | 82% | Lissencephaly, 607432<br>Subcortical laminar heterotopia, 607432 |
| PAH | 91.8 | 96% | 91% | Phenylketonuria, 261600<br>[Hyperphenylalaninemia, non-PKU mild], 261600 |
| PAK3 | 110.7 | 100% | 100% | Mental retardation, X-linked 30/47, 300558 |
| PANK2 | 123.3 | 100% | 98% | Neurodegeneration with brain iron accumulation 1, 234200<br>HARP syndrome, 607236 |
| PAX1 | 100.3 | 96% | 88% | ?Orofaciocervical syndrome 2, 615560 |

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|---|---|---|---|---|
| PAX6 | 102.7 | 100% | 100% | Aniridia, 106210<br>Peters anomaly, 604229<br>Cataract with late-onset corneal dystrophy, 106210<br>Keratitis, 148190<br>Foveal hyperplasia, 136520<br>Morning glory disc anomaly, 120430<br>Optic nerve hypoplasia, 165550 |
| PAX8 | 72.9 | 97% | 89% | Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 |
| PC | 110.8 | 99% | 93% | Pyruvate carboxylase deficiency, 266150 |
| PCDH19 | 159.2 | 100% | 100% | Epileptic encephalopathy, early infantile, 9, 300088 |
| PCGF2 | 67.4 | 100% | 89% | no OMIM phenotype |
| PCNT | 112.6 | 99% | 95% | Microcephalic osteodysplastic primordial dwarfism, type II, 210720 |
| PDE4D | 125.2 | 99% | 95% | Acrocydostosis 2 with or without hormone resistance, 614613 |
| PDHA1 | 134.1 | 100% | 99% | Pyruvate dehydrogenase E1-alpha deficiency, 312170<br>Leigh syndrome, X-linked, 308930 |
| PDSS1 | 106.4 | 93% | 88% | Coenzyme Q10 deficiency, primary, 2, 614651 |
| PDSS2 | 105.9 | 100% | 99% | Coenzyme Q10 deficiency, primary, 3, 614652 |
| PEPD | 80 | 95% | 95% | Prolidase deficiency, 170100 |
| PEX1 | 135.7 | 100% | 100% | Peroxisome biogenesis disorder 1A (Zellweger), 214100<br>Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 |
| PEX10 | 79.1 | 93% | 86% | Peroxisome biogenesis disorder 6A (Zellweger), 614870<br>Peroxisome biogenesis disorder 6B, 614871 |
| PEX11B | 187.2 | 100% | 100% | Peroxisome biogenesis disorder 14B, 614920 |
| PEX12 | 141 | 100% | 100% | Peroxisome biogenesis disorder 3A (Zellweger), 614859<br>Peroxisome biogenesis disorder 3B, 266510 |
| PEX13 | 141.8 | 96% | 94% | Peroxisome biogenesis disorder 11A (Zellweger), 614883<br>Peroxisome biogenesis disorder 11B, 614885 |
| PEX16 | 103.1 | 86% | 86% | Peroxisome biogenesis disorder 8A, (Zellweger), 614876<br>Peroxisome biogenesis disorder 8B, 614877 |
| PEX19 | 117.7 | 100% | 100% | Peroxisome biogenesis disorder 12A (Zellweger), 614886 |
| PEX2 | 174.8 | 100% | 100% | Peroxisome biogenesis disorder 5A (Zellweger), 614866<br>Peroxisome biogenesis disorder 5B, 614867 |
| PEX26 | 123.7 | 100% | 100% | Peroxisome biogenesis disorder 7A (Zellweger), 614872<br>Peroxisome biogenesis disorder 7B, 614873 |

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| PEX3 | 146.3 | 100% | 100% | Peroxisome biogenesis disorder 10A (Zellweger), 614882 |
| PEX5 | 94 | 98% | 97% | Peroxisome biogenesis disorder 2A (Zellweger), 214110<br>Peroxisome biogenesis disorder 2B, 202370 |
| PEX6 | 108.2 | 100% | 93% | Peroxisome biogenesis disorder 4A (Zellweger), 614862<br>Peroxisome biogenesis disorder 4B, 614863 |
| PEX7 | 103.8 | 91% | 89% | Rhizomelic chondrodysplasia punctata, type 1, 215100<br>Peroxisome biogenesis disorder 9B, 614879 |
| PGAP1 | 128.6 | 100% | 100% | ?Mental retardation, autosomal recessive 42, 615802 |
| PGAP2 | 130.5 | 99% | 97% | Hyperphosphatasia with mental retardation syndrome 3, 614207 |
| PGAP3 | 69.4 | 100% | 88% | Hyperphosphatasia with mental retardation syndrome 4, 615716 |
| PGK1 | 87.9 | 85% | 79% | Phosphoglycerate kinase 1 deficiency, 300653 |
| PHF6 | 144.1 | 100% | 100% | Borjeson-Forssman-Lehmann syndrome, 301900 |
| PHF8 | 112.9 | 100% | 99% | Mental retardation syndrome, X-linked, Siderius type, 300263 |
| PHGDH | 101.2 | 100% | 100% | Phosphoglycerate dehydrogenase deficiency, 601815 |
| PHIP | 131.3 | 99% | 98% | No OMIM phenotype |
| PIGL | 102.4 | 100% | 100% | CHIME syndrome, 280000 |
| PIGN | 113.1 | 100% | 100% | Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 |
| PIGO | 130.9 | 100% | 100% | Hyperphosphatasia with mental retardation syndrome 2, 614749 |
| PIGT | 141.3 | 100% | 99% | ?Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398<br>?Paroxysmal nocturnal hemoglobinuria 2, 615399 |
| PIGV | 196.9 | 100% | 100% | Hyperphosphatasia with mental retardation syndrome 1, 239300 |
| PIK3R2 | 96.6 | 93% | 86% | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome, 603387 |
| PLA2G6 | 87.3 | 99% | 91% | Infantile neuroaxonal dystrophy 1, 256600<br>Neurodegeneration with brain iron accumulation 2B, 610217<br>Parkinson disease 14, 612953 |
| PLCB1 | 121.5 | 99% | 98% | Epileptic encephalopathy, early infantile, 12, 613722 |
| PLIN4 | 208.1 | 100% | 98% | No OMIM phenotype |
| PLP1 | 88 | 100% | 99% | Pelizaeus-Merzbacher disease, 312080<br>Spastic paraplegia 2, X-linked, 312920 |
| PLXND1 | 84.7 | 98% | 94% | No OMIM phenotype |
| PMM2 | 105.6 | 100% | 100% | Congenital disorder of glycosylation, type Ia, 212065 |
| PNKP | 78.8 | 100% | 99% | Epileptic encephalopathy, early infantile, 10, 613402 |
| PNP | 128.6 | 100% | 100% | Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 |
| POC1A | 111.3 | 98% | 95% | Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 |

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|---|---|---|---|---|
| POGZ | 142.3 | 100% | 99% | Autism (Neale (2012) Nature 485, 242)<br>Intellectual disability (Gilissen (2014) Nature 511, 344)<br>Schizophrenia (Fromer (2014) Nature 506, 179) |
| POLG | 95.9 | 100% | 94% | Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700<br>Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662<br>Mitochondrial recessive ataxia syndrome, 607459<br>Progressive external ophthalmoplegia, autosomal dominant, 157640 |
| POLR3A | 96 | 99% | 96% | Leukodystrophy, hypomyelinating, 7,oligodontia and hypogonadotropic hypogonadism, 607694 |
| POLR3B | 115.6 | 98% | 98% | Leukodystrophy, hypomyelinating, 8,oligodontia and hypogonadotropic hypogonadism, 614381 |
| POMGNT1 | 114.7 | 100% | 99% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 |
| POMT1 | 115.5 | 100% | 98% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 |
| POMT2 | 77.6 | 99% | 91% | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150<br>Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 |
| PORCN | 127 | 92% | 92% | Focal dermal hypoplasia, 305600 |
| POU1F1 | 109.4 | 100% | 100% | Pituitary hormone deficiency, combined, 1, 613038 |
| PPOX | 122.9 | 100% | 98% | Porphyria variegata, 176200 |
| PPP2R1A | 102.2 | 93% | 92% | Mental retardation, autosomal dominant 36, 616362 |
| PPP2R5D | 147.4 | 98% | 97% | Mental retardation, autosomal dominant 35, 616355 |
| PPT1 | 76 | 100% | 99% | Ceroid lipofuscinosis, neuronal, 1, 256730 |
| PQBP1 | 148.6 | 100% | 99% | Renpenning syndrome, 309500 |
| PRODH | 53.4 | 89% | 70% | Hyperprolinemia, type I, 239500<br>{Schizophrenia, susceptibility to, 4}, 600850 |
| PRPS1 | 137.7 | 100% | 100% | Gout, PRPS-related, 300661<br>Phosphoribosylpyrophosphate synthetase superactivity, 300661<br>Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070<br>Arts syndrome, 301835<br>Deafness, X-linked 1, 304500 |
| PRSS12 | 114.7 | 100% | 97% | Mental retardation, autosomal recessive 1, 249500 |
| PSAP | 94.3 | 100% | 99% | Metachromatic leukodystrophy due to SAP-b deficiency, 249900<br>Gaucher disease, atypical, 610539<br>Combined SAP deficiency, 611721<br>Krabbe disease, atypical, 611722 |

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|---|---|---|---|---|
| PSEN1 | 107 | 100% | 99% | Acne inversa, familial, 3, 613737<br>Alzheimer disease, type 3, 607822<br>Cardiomyopathy, dilated, 1U, 61 |
| PTCH1 | 93.4 | 99% | 97% | Basal cell nevus syndrome, 109400<br>Basal cell carcinoma, somatic, 605462<br>Holoprosencephaly-7, 610828 |
| PTCHD1 | 183.3 | 100% | 100% | {Autism,susceptibility to,X-linked 4},300830 |
| PTDSS1 | 138.3 | 100% | 100% | Lenz-Majewski hyperostotic dwarfism, 151050 |
| PTEN | 141.6 | 100% | 99% | Cowden syndrome 1, 158350<br>Bannayan-Riley-Ruvalcaba syndrome, 153480<br>Macrocephaly/autism syndrome, 605309<br>PTEN hamartoma tumor syndrome |
| PTPN11 | 49.2 | 94% | 69% | Noonan syndrome 1, 163950<br>LEOPARD syndrome 1, 151100<br>Leukemia, juvenile myelomonocytic, 607785<br>Metachondromatosis, 156250 |
| PUF60 | 144.1 | 98% | 95% | Verheij syndrome, 615583 |
| PURA | 125.4 | 100% | 99% | Mental retardation, autosomal dominant 31, 616158 |
| PUS1 | 81.9 | 100% | 100% | Mitochondrial myopathy and sideroblastic anemia 1, 600462 |
| PYCR1 | 101.4 | 100% | 100% | Cutis laxa, autosomal recessive, type IIB, 612940<br>Cutis laxa, autosomal recessive, type IIIB, 614438 |
| RAB18 | 129.3 | 100% | 100% | Warburg micro syndrome 3, 614222 |
| RAB27A | 142.7 | 100% | 100% | Griscelli syndrome, type 2, 607624 |
| RAB39B | 174.3 | 100% | 100% | Mental retardation, X-linked 72, 300271 |
| RAB3GAP1 | 133.1 | 98% | 98% | Warburg micro syndrome 1, 600118 |
| RAB3GAP2 | 121 | 100% | 99% | Martsolf syndrome, 212720<br>Warburg micro syndrome 2, 614225 |
| RAB40AL | 34.7 | 98% | 79% | Mental retardation, X-linked, syndromic, Martin-Probst type, 300519 |
| RAC1 | 41.2 | 64% | 55% | No OMIM phenotype |
| RAD21 | 97.2 | 99% | 96% | Cornelia de Lange syndrome 4, 614701 |
| RAF1 | 99.8 | 100% | 99% | Noonan syndrome 5, 611553<br>LEOPARD syndrome 2, 611554 |
| RAI1 | 151.6 | 99% | 99% | Immunodeficiency 9, 612782<br>Smith-Magenis syndrome, 182290 |

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|---|---|---|---|---|
| RARS2 | 94.9 | 100% | 100% | Pontocerebellar hypoplasia, type 6, 611523 |
| RBM10 | 125.2 | 100% | 98% | TARP syndrome, 311900 |
| RBM28 | 108.3 | 100% | 98% | Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 |
| RELN | 115.3 | 99% | 98% | Lissencephaly 2 (Norman-Roberts type), 257320 |
| REV3L | 160.2 | 100% | 100% | No OMIM phenotype |
| RFT1 | 84.6 | 100% | 95% | Congenital disorder of glycosylation, type In, 612015 |
| RHEB | 58.2 | 73% | 68% | No OMIM phenotype |
| RIT1 | 139.9 | 100% | 100% | Noonan syndrome 8, 615355 |
| RMND1 | 79.8 | 95% | 92% | Combined oxidative phosphorylation deficiency 11, 614922 |
| RNASEH2A | 107.1 | 100% | 98% | Aicardi-Goutieres syndrome 4, 610333 |
| RNASEH2B | 112.3 | 100% | 100% | Aicardi-Goutieres syndrome 2, 610181 |
| RNASEH2C | 163.4 | 100% | 100% | Aicardi-Goutieres syndrome 3, 610329 |
| RNASET2 | 110.5 | 100% | 98% | Leukoencephalopathy, cystic, without megalencephaly, 612951 |
| ROGDI | 113.1 | 95% | 95% | Kohlschutter-Tonz syndrome, 226750 |
| RPGRIP1L | 110.8 | 99% | 98% | Joubert syndrome 7, 611560<br>Meckel syndrome 5, 611561<br>COACH syndrome, 216360 |
| RPL10 | 98.7 | 100% | 98% | {Autism, susceptibility to, X-linked 5}, 300847 |
| RPS6KA3 | 106.3 | 100% | 100% | Coffin-Lowry syndrome, 303600<br>Mental retardation, X-linked 19, 300844 |
| RTEL1 | 94.7 | 99% | 98% | Dyskeratosis congenita, autosomal dominant 4, 615190<br>Dyskeratosis congenita, autosomal recessive 5, 615190 |
| SALL1 | 162.3 | 99% | 98% | Townes-Brocks syndrome, 107480<br>Townes-Brocks branchiootorenal-like syndrome, 107480 |
| SATB2 | 128.8 | 100% | 99% | Cleft palate and mental retardation, 119540 |
| SC5D | 171.6 | 100% | 100% | Lathosterolosis, 607330 |
| SCN1A | 129.1 | 100% | 98% | Epilepsy, generalized, with febrile seizures plus, type 2, 604403<br>Dravet syndrome, 607208<br>Migraine, familial hemiplegic, 3, 609634<br>Febrile seizures, familial, 3A, 604403 |
| SCN2A | 136.3 | 100% | 99% | Seizures, benign familial infantile, 3, 607745<br>Epileptic encephalopathy, early infantile, 11, 613721 |
| SCN8A | 144.9 | 100% | 99% | Cognitive impairment with or without cerebellar ataxia, 614306<br>Epileptic encephalopathy, early infantile, 13, 614558 |

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| SCO2 | 98 | 100% | 100% | Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377<br>Myopia 6, 608908 |
| SDHA | 10.4 | 34% | 17% | Leigh syndrome, 256000<br>Mitochondrial respiratory chain complex II deficiency, 252011<br>Cardiomyopathy, dilated, 1GG, 613642<br>Paragangliomas 5, 614165 |
| SERAC1 | 100.6 | 100% | 100% | 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 |
| SETBP1 | 171.1 | 98% | 97% | Schinzel-Giedion midface retraction syndrome, 269150 |
| SETD2 | 155.1 | 100% | 100% | No OMIM phenotype |
| SETD5 | 169 | 100% | 98% | Mental retardation,autosomal dominant 24,615761 |
| SF1 | 78.2 | 90% | 87% | No OMIM phenotype |
| SGSH | 92.3 | 94% | 93% | Mucopolysaccharidisis type 3A (Sanfilippo A), 252900 |
| SHANK2 | 104.1 | 99% | 95% | {Autism susceptibility 17}, 613436 |
| SHANK3 | 84.6 | 95% | 82% | Phelan-McDermid syndrome, 606232<br>{Schizophrenia 15}, 613950 |
| SHH | 118.3 | 99% | 96% | Holoprosencephaly-3, 142945<br>Single median maxillary central incisor, 147250<br>Microphthalmia with coloboma 5, 611638<br>Schizencephaly, 269160 |
| SHOC2 | 128.3 | 100% | 100% | Noonan-like syndrome with loose anagen hair, 607721 |
| SHROOM4 | 140.4 | 100% | 99% | Stocco dos Santos X-linked mental retardation syndrome, 300434 |
| SIL1 | 107 | 100% | 100% | Marinesco-Sjogren syndrome, 248800 |
| SIN3A | 115.9 | 95% | 94% | No OMIM phenotype |
| SIX3 | 139.2 | 100% | 100% | Holoprosencephaly-2, 157170<br>Schizensephaly, 269160 |
| SKI | 76.3 | 95% | 84% | Shprintzen-Goldberg syndrome, 182212 |
| SLC12A6 | 103.7 | 100% | 99% | Agenesis of the corpus callosum with peripheral neuropathy, 218000 |
| SLC16A2 | 106.7 | 99% | 97% | Allan-Herndon-Dudley syndrome, 300523 |
| SLC17A5 | 104.9 | 100% | 100% | Salla disease, 604369<br>Sialic acid storage disorder, infantile, 269920 |
| SLC19A3 | 125.4 | 100% | 100% | Thiamine metabolism dysfunction syndrome 2,607483 |
| SLC1A1 | 128.9 | 100% | 100% | Dicarboxylic aminoaciduria, 222730<br>{?Schizophrenia susceptibility 18}, 615232 |
| SLC1A4 | 108.1 | 100% | 98% | Spastic tetraplegia,thin corpus callosum,and progressive microcephaly,616657 |

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| SLC25A15 | 104.7 | 91% | 79% | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 |
| SLC25A22 | 85 | 100% | 95% | Epileptic encephalopathy, early infantile, 3, 609304 |
| SLC2A1 | 106 | 100% | 100% | GLUT1 deficiency syndrome 1, 606777<br>GLUT1 deficiency syndrome 2, 612126<br>{Epilepsy, idiopathic generalized, suscpetibility to, 12}, 614847<br>Dystonia 9, 601042 |
| SLC33A1 | 106.7 | 100% | 98% | Spastic paraplegia 42, autosomal dominant, 612539<br>Congenital cataracts, hearing loss, and neurodegeneration, 614482 |
| SLC35A2 | 113.8 | 100% | 100% | Congenital disorder of glycosylation, type 2m, 300896 |
| SLC35C1 | 116.4 | 100% | 100% | Congenital disorder of glycosylation, type IIc, 266265 |
| SLC39A12 | 89.2 | 97% | 96% | No OMIM phenotype |
| SLC4A4 | 116.2 | 100% | 100% | Renal tubular acidosis, proximal, with ocular abnormalities, 604278 |
| SLC6A17 | 92.2 | 99% | 95% | Mental retardation, autosomal recessive 48, 616269 |
| SLC6A3 | 88.7 | 100% | 100% | Parkinsonism -dystonia, infantile, 613135<br>{Nicotine dependence, protection against}, 188890 |
| SLC6A8 | 11.4 | 32% | 15% | Cerebral creatine deficiency syndrome 1, 300352 |
| SLC7A7 | 104.4 | 100% | 98% | Lysinuric protein intolerance, 222700 |
| SLC9A6 | 131.1 | 100% | 99% | Mental retardation, X-linked syndromic, Christianson type, 300243 |
| SMAD4 | 147.6 | 100% | 100% | Pancreatic cancer<br>Polyposis, juvenile intestinal, 174900<br>Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050<br>Myhre syndrome, 139210 |
| SMARCA2 | 99.3 | 98% | 96% | Nicolaides-Baraitser syndrome, 601358 |
| SMARCA4 | 94.7 | 97% | 94% | Rhabdoid tumor predisposition syndrome 2, 613325<br>Mental retardation, autosomal dominant 16, 614609 |
| SMARCB1 | 141.4 | 100% | 100% | Rhabdoid tumors, somatic, 609322<br>Mental retardation, autosomal dominant 15, 614608 |
| SMARCC2 | 99.7 | 98% | 94% | No OMIM phenotype |
| SMARCE1 | 72.3 | 84% | 75% | {Meningioma, familial, susceptibility to}, 607174 |
| SMC1A | 150.2 | 98% | 96% | Cornelia de Lange syndrome 2, 300590 |
| SMC3 | 128.4 | 99% | 97% | Cornelia de Lange syndrome 3, 610759 |
| SMOC1 | 84.3 | 100% | 98% | Microphthalmia with limb anomalies, 206920 |
| SMPD1 | 113.6 | 100% | 97% | Niemann-Pick disease, type A, 257200<br>Niemann-Pick disease, type B, 607616 |

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|---|---|---|---|---|
| SMS | 38.2 | 90% | 68% | Mental retardation, X-linked, Snyder-Robinson type, 309583 |
| SNAP29 | 137.5 | 100% | 100% | Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 |
| SNIP1 | 153.5 | 100% | 98% | Psychomotor retardation, epilepsy and craniofacial dysmorphism, 614501 |
| SNX14 | 113.9 | 97% | 96% | Spinocerebellar ataxia,autosomal recessive 20,616354 |
| SOBP | 132 | 99% | 94% | Mental retardation, anterior maxillary protrusion, and strabismus, 613671 |
| SON | 140.7 | 100% | 99% | No OMIM phenotype |
| SOS1 | 124.5 | 100% | 99% | Fibromatosis, gingival, 135300<br>Noonan syndrome 4, 610733 |
| SOX10 | 82.5 | 100% | 100% | Waardenburg syndrome, type 4C, 613266<br>Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584<br>PCWH syndrome, 609136 |
| SOX11 | 138.6 | 100% | 100% | Mental retardation, autosomal dominant, 27, 615866 |
| SOX2 | 168.7 | 100% | 100% | Microphthalmia, syndromic 3, 206900<br>Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 |
| SOX3 | 110.3 | 99% | 96% | Mental retardation, X-linked, with isolated growth hormone deficiency, 300123<br>Panhypopituitarism, X-linked, 312000 |
| SOX5 | 92.9 | 99% | 99% | No OMIM phenotype |
| SPG11 | 121.6 | 100% | 99% | Spastic paraplegia 11, autosomal recessive, 604360 |
| SPRED1 | 146 | 100% | 100% | Legius syndrome, 611431 |
| SPTAN1 | 110.6 | 100% | 99% | Epileptic encephalopathy, early infantile, 5 |
| SRCAP | 149 | 100% | 99% | Floating-Harbor syndrome, 136140 |
| SRD5A3 | 133.3 | 100% | 100% | Congenital disorder of glycosylation, type Iq, 612379<br>Kahrizi syndrome, 612713 |
| SRPX2 | 92.4 | 100% | 99% | Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 |
| ST3GAL3 | 125.3 | 100% | 100% | Mental retardation, autosomal recessive 12, 611090<br>Epileptic encephalopathy, early infantile, 15, 615006 |
| ST3GAL5 | 128.2 | 94% | 92% | Amish infantile epilepsy syndrome, 609056 |
| STAG1 | 102.9 | 100% | 98% | No OMIM phenotype |
| STIL | 155.2 | 100% | 100% | Microcephaly 7, primary, autosomal recessive, 612703 |
| STRA6 | 79 | 100% | 97% | Microphthalmia, syndromic 9, 601186<br>Microphthalmia, isolated, with coloboma 8, 601186 |
| STT3A | 123.1 | 100% | 98% | ?Congenital disorder of glycosylation, type Iw, 615596 |
| STT3B | 111.9 | 100% | 97% | ?Congenital disorder of glycosylation, type Ix, 615597 |
| STX1B | 107.5 | 100% | 98% | Generalized epilepsy with febrile seizures plus,type 9,616172 |

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|---|---|---|---|---|
| STXBP1 | 106 | 100% | 100% | Epileptic encephalopathy, early infantile, 4, 612164 |
| SUCLA2 | 83.2 | 94% | 87% | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 |
| SUOX | 191.2 | 100% | 100% | Sulfite oxidase deficiency, 272300 |
| SURF1 | 97.4 | 88% | 88% | Leigh syndrome, due to COX deficiency, 256000 |
| SYN1 | 71.6 | 97% | 79% | Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 |
| SYNCRIP | 102.7 | 97% | 93% | No OMIM phenotype |
| SYNE1 | 121.9 | 99% | 98% | Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998<br>Spinocerebellar ataxia, autosomal recessive 8, 610743 |
| SYNGAP1 | 58.1 | 91% | 79% | Mental retardation, autosomal dominant 5, 612621 |
| SYP | 98.2 | 100% | 99% | Mental retardation, X-linked 96, 300802 |
| SYT14 | 150.3 | 93% | 93% | Spinocerebellar ataxia, autosomal recessive 11, 614229 |
| TAF2 | 119.4 | 100% | 100% | Mental retardation, autosomal recessive 40, 615599 |
| TAT | 111.1 | 100% | 100% | Tyrosinemia, type II, 276600 |
| TBC1D24 | 125.5 | 100% | 100% | Myoclonic epilepsy, infantile, familial, 605021<br>Epileptic encephalopathy, early infantile, 16, 615338 |
| TBC1D7 | 148.7 | 100% | 100% | Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000 |
| TBCE | 127.6 | 100% | 100% | Kenny-Caffey syndrome-1, 244460<br>Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 |
| TBR1 | 97.8 | 99% | 98% | No OMIM phenotype |
| TCF20 | 167.1 | 100% | 99% | No OMIM phenotype |
| TCF4 | 107.5 | 97% | 97% | Pitt-Hopkins syndrome, 610954 |
| TCF7L2 | 130.2 | 100% | 99% | {Diabetes mellitus,type 2,susceptibility to},125853 |
| TECR | 100.2 | 100% | 92% | Mental retardation, autosomal recessive 14, 614020 |
| TFAP2A | 81.6 | 96% | 88% | Branchiooculofacial syndrome, 113620 |
| TGFBR1 | 136 | 96% | 93% | Loeys-Dietz syndrome, type 1A, 609192<br>Loeys-Dietz syndrome, type 2A, 608967<br>{Multiple self-healing squamous epithelioma, susceptiblity to}, 132800 |
| TGFBR2 | 102.9 | 100% | 99% | Colorectal cancer, hereditary nonpolyposis, type 6, 614331<br>Esophageal cancer, somatic, 133239<br>Loeys-Dietz syndrome, type 1B, 610168<br>Loeys-Dietz syndrome, type 2B, 610380 |
| TGIF1 | 190.6 | 100% | 100% | Holoprosencephaly-4, 142946 |
| TH | 98.3 | 95% | 91% | Segawa syndrome,recessive,605407 |

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|---|---|---|---|---|
| THOC6 | 179.7 | 100% | 98% | Beaulieu-Boycott-Innes syndrome, 613680 |
| THRB | 132.4 | 100% | 100% | Thyroid hormone resistance, 188570<br>Thyroid hormone resistance, autosomal recessive, 274300<br>Thyroid hormone resistance, selective pituitary, 145650 |
| TIMM8A | 68.7 | 99% | 79% | Deafness, X-linked 1, progressive<br>Mohr-Tranebjaerg syndrome, 304700<br>Jensen syndrome, 311150 |
| TLK2 | 52.1 | 64% | 57% | No OMIM phenotype |
| TMCO1 | 83.6 | 100% | 95% | Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 614132 |
| TMEM165 | 100.2 | 100% | 100% | Congenital disorder of glycosylation, type IIk, 614727 |
| TMEM231 | 85.5 | 99% | 88% | Joubert syndrome 20, 614970<br>Meckel syndrome, type 11, 615397 |
| TMEM237 | 101.4 | 100% | 97% | Joubert syndrome 14, 614424 |
| TMEM67 | 125.1 | 100% | 100% | Meckel syndrome 3, 607361<br>Joubert syndrome 6, 610688<br>{Bardet-Biedl syndrome 14, modifier of}, 209900<br>COACH syndrome, 216360<br>Nephronophthisis 11, 613550 |
| TMLHE | 68.2 | 85% | 78% | Epsilon-trimethyllysine hydrolylase deficiency, 300872 |
| TPP1 | 149.2 | 100% | 100% | Ceroid lipofuscinosis, neuronal, 2, 204500 |
| TRAPPC11 | 129.3 | 100% | 100% | Muscular dystrophy, limb-girdle, type 2S |
| TRAPPC9 | 77.4 | 97% | 94% | Mental retardation, autosomal recessive 13, 613192 |
| TREX1 | 160.8 | 100% | 100% | Aicardi-Goutieres syndrome 1, dominant and recessive, 225750<br>Chilblain lupus, 610448<br>Vasculopathy, retinal, with cerebral leukodystrophy, 192315<br>{Systemic lupus erythematosus, susceptibility to}, 152700 |
| TRIM32 | 135.8 | 100% | 100% | ?Bardet-Biedl syndrome 11, 615988<br>Muscular dystrophy, limb-girdle, type 2H, 254110 |
| TRIO | 115.4 | 97% | 94% | No OMIM phenotype |
| TRIP12 | 124.2 | 100% | 100% | No OMIM phenotype |
| TRMT10A | 128.5 | 100% | 100% | Microcephaly, short stature and impaired glucose metabolism, 616033 |
| TSC1 | 105.4 | 100% | 99% | Tuberous sclerosis-1, 191100<br>Lymphangioleiomyomatosis, 606690<br>Focal cortical dysplasia, Taylor balloon cell type, 607341 |

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|---|---|---|---|---|
| TSC2 | 99.3 | 99% | 97% | Tuberous sclerosis-2, 613254<br>Lymphangioleiomyomatosis, somatic, 606690 |
| TSEN54 | 115.1 | 98% | 96% | Pontocerebellar hypoplasia type 2A, 277470<br>Pontocerebellar hypoplasia type 4, 225753 |
| TSPAN7 | 108.2 | 100% | 99% | Mental retardation, X-linked 58, 300210 |
| TTC8 | 114.5 | 100% | 100% | Bardet-Biedl syndrome 8, 209900<br>Retinitis pigmentosa 51, 613464 |
| TTI2 | 109 | 100% | 100% | Mental retardation, autosomal recessive 39, 615541 |
| TUBA1A | 28.2 | 97% | 64% | Lissencephaly 3, 611603 |
| TUBA8 | 109.3 | 100% | 97% | Polymicrogyria with optic nerve hypoplasia, 613180 |
| TUBB2B | 52 | 100% | 97% | Polymicrogyria, symmetric or asymmetric, 610031 |
| TUBGCP6 | 146.6 | 100% | 98% | Microcephaly and chorioretinopathy, autosomal recessive 1, 251270 |
| TUSC3 | 131 | 100% | 99% | Mental retardation, autosomal recessive 7, 611093 |
| TWIST1 | 137.6 | 100% | 99% | Craniosynostosis, type 1, 123100<br>Robinow-Sorauf syndrome, 180750<br>Saethre-Chotzen syndrome, 101400<br>Saethre-Chotzen syndrome with eyelid anomalies, 101400 |
| UBE2A | 116.8 | 100% | 100% | Mental retardation, X-linked syndromic, Nascimento-type, 300860 |
| UBE3A | 109.7 | 100% | 99% | Angelman syndrome, 105830 |
| UBE3B | 108.8 | 95% | 93% | Blepharophimosis-ptosis-intellectual disability syndrome, 615057 |
| UBR1 | 112.8 | 100% | 100% | Johanson-Blizzard syndrome, 243800 |
| UPB1 | 150.3 | 100% | 98% | Beta-ureidopropionase deficiency, 613161 |
| UPF3B | 111.9 | 100% | 98% | Mental retardation, X-linked, syndromic 14, 300676 |
| USP7 | 93.7 | 100% | 97% | No OMIM phenotype |
| USP9X | 138.8 | 100% | 99% | Mental retardation, X-linked 99, 300919 |
| VLDLR | 124 | 100% | 99% | Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 |
| VPS13B | 119.9 | 99% | 99% | Cohen syndrome, 216550 |
| VRK1 | 143.4 | 100% | 100% | Pontocerebellar hypoplasia type 1A, 607596 |
| WAC | 128.5 | 100% | 97% | No OMIM phenotype |
| WDR13 | 121.3 | 100% | 99% | No OMIM phenotype |
| WDR19 | 137 | 100% | 100% | Nephronophtisis 13, 614377<br>Senior-Loken syndrome 8, 616307<br>?Cranioectodermal dysplasia 4, 614378<br>?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 |

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|---|---|---|---|---|
| WDR45 | 96.9 | 99% | 93% | Neurodegeneration with brain iron accululation 5, 300894 |
| WDR62 | 119.3 | 99% | 96% | Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 |
| WDR73 | 172.5 | 100% | 100% | Galloway-Mowat syndrome,251300 |
| WDR81 | 133.5 | 99% | 98% | Cerebellar ataxia, mental retardation and dysequilibrium syndrome 2, 610185 |
| WWOX | 111.9 | 97% | 97% | Epileptic encephalopathy, early infantile, 28, 616211<br>Esophageal squamous cell carcinoma, somatic, 133239<br>Spinocerebellar ataxia, autosomal recessive, 12, 614322 |
| XPA | 99.5 | 100% | 97% | Xeroderma pigmentosum, group A, 278700 |
| XPNPEP3 | 128.6 | 97% | 95% | Nephronophthisis-like nephropathy 1, 613159 |
| XYLT1 | 123.2 | 98% | 91% | Desbuquois dysplasia 2, 615777<br>{Pseudoxanthoma elasticum, modifier of severity of}, 264800 |
| YAP1 | 77.5 | 96% | 90% | Coloboma, ocular with or without hearing impairment, cleft lip/palate and mental retardation, 120433 |
| YWHAE | 43 | 77% | 66% | No OMIM phenotype |
| YY1 | 94.1 | 100% | 100% | No OMIM phenotype |
| ZBTB16 | 137.9 | 100% | 98% | Leukemia, acute promyelocytic, PL2F/RARA type<br>Skeletal defects, genital hypoplasia, and mental retardation, 612447 |
| ZBTB18 | 186.6 | 100% | 99% | ?Mental retardation, autosomal dominant 22, 612337 |
| ZDHHC15 | 126 | 100% | 100% | ?Mental retardation, X-linked 91, 300577 |
| ZDHHC9 | 96.8 | 100% | 99% | Mental retardation, X-linked syndromic, Raymond type, 300799 |
| ZEB2 | 162.9 | 100% | 100% | Mowat-Wilson syndrome, 235730 |
| ZFYVE26 | 102.6 | 97% | 95% | Spastic paraplegia 15, autosomal recessive, 270700 |
| ZIC2 | 79.9 | 94% | 85% | Holoprosencephaly-5, 609637 |
| ZMYND11 | 132 | 100% | 100% | Mental retardation, autosomal dominant 30, 616083 |
| ZNF292 | 168.3 | 99% | 97% | No OMIM phenotype |
| ZNF41 | 140.7 | 100% | 100% | Mental retardation, X-linked 89, 300848 |
| ZNF592 | 122.7 | 93% | 91% | Spinocerebellar ataxia, autosomal recessive 5, 606937 |
| ZNF674 | 129.8 | 100% | 99% | Mental retardation, X-linked 92, 300851 |
| ZNF711 | 161.5 | 100% | 100% | Mental retardation, X-linked 97, 300803 |
| ZNF81 | 109.9 | 100% | 99% | Mental retardation, X-linked 45, 300498 |

*Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.*
*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 : November 15th, 2015*
*This list is accurate for all panel versions starting with DG 2.4. (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*