## CHU Liège : Gene Panel - Intellectual Disability v4 (859 genes)

| Gene (HGNC) | OMIM (gene) | OMIM (disease) | OMIM (phenotype) | Fraction of Length Covered Above 30x (%) |
|---|---|---|---|---|
| A2ML1 | 610627 | - | Vissers et al., EJHG 2015 : Noonan syndrome | 99,35 |
| ABCC9 | 601439 | 239850 | Hypertrichotic osteochondrodysplasia | 99,41 |
| ABCD1 | 300371 | 300100 | Adrenoleukodystrophy | 99,49 |
| ABHD5 | 604780 | 275630 | Chanarin-Dorfman syndrome | 98,48 |
| ACAD9 | 611103 | 611126 | Mitochondrial complex I deficiency due to ACAD9 deficiency | 99,57 |
| ACO2 | 100850 | 614559 | Infantile cerebellar-retinal degeneration | 99,66 |
| ACOX1 | 609751 | 264470 | Peroxisomal acyl-CoA oxidase deficiency | 99,49 |
| ACSL4 | 300157 | 300387 | Mental retardation, X-linked 63 | 91,09 |
| ACTB | 102630 | 243310 | Baraitser-Winter syndrome 1 | 99,34 |
| ACTG1 | 102560 | 614583 | Baraitser-Winter syndrome 2 | 99,69 |
| ACY1 | 104620 | 609924 | Aminoacylase 1 deficiency | 99,67 |
| ADAR | 146920 | 615010 | Aicardi-Goutieres syndrome 6 | 99,45 |
| ADAT3 | 615302 | 615286 | Mental retardation, autosomal recessive 36 | 81,40 |
| ADGRG1 | 604110 | 606854 | Polymicrogyria, bilateral frontoparietal | 99,67 |
| ADK | 102750 | 614300 | Hypermethioninemia due to adenosine kinase deficiency | 98,74 |
| ADNP | 611386 | 615873 | Helsmoortel-van der Aa syndrome | 99,55 |
| ADSL | 608222 | 103050 | Adenylosuccinase deficiency | 99,58 |
| AFF2 | 300806 | 309548 | Mental retardation, X-linked, FRAXE type | 90,80 |
| AGA | 613228 | 208400 | Aspartylglucosaminuria | 99,60 |
| AGPAT2 | 603100 | 608594 | Lipodystrophy, congenital generalized, type 1 | 91,26 |
| AHCY | 180960 | 613752 | Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase | 99,67 |
| AHDC1 | 615790 | 615829 | Xia-Gibbs syndrome | 99,68 |
| AHI1 | 608894 | 608629 | Joubert syndrome 3 | 99,05 |
| AIMP1 | 603605 | 260600 | Leukodystrophy, hypomyelinating, 3 | 96,88 |
| AKT3 | 611223 | 615937 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2 | 98,82 |
| ALDH18A1 | 138250 | 219150 | Cutis laxa, autosomal recessive, type IIIA | 99,66 |
| ALDH3A2 | 609523 | 270200 | Sjogren-Larsson syndrome | 99,31 |
| ALDH4A1 | 606811 | 239510 | Hyperprolinemia, type II | 96,44 |
| ALDH5A1 | 610045 | 271980 | Succinic semialdehyde dehydrogenase deficiency | 93,04 |
| ALDH7A1 | 107323 | 266100 | Epilepsy, pyridoxine-dependent | 99,00 |
| ALG1 | 605907 | 608540 | Congenital disorder of glycosylation, type Ik | 99,43 |
| ALG11 | 613666 | 613661 | Congenital disorder of glycosylation, type Ip | 99,57 |
| ALG12 | 607144 | 607143 | Congenital disorder of glycosylation, type Ig | 99,69 |
| ALG13 | 300776 | 300884 | Epileptic encephalopathy, early infantile, 36 | 92,88 |
| ALG2 | 607905 | 616228 | Myasthenic syndrome, congenital, 14, with tubular aggregates | 97,14 |
| ALG3 | 608750 | 601110 | Congenital disorder of glycosylation, type Id | 99,59 |
| ALG6 | 604566 | 603147 | Congenital disorder of glycosylation, type Ic | 98,36 |
| ALG9 | 606941 | 263210 | Gillessen-Kaesbach-Nishimura syndrome | 96,33 |
| ALMS1 | 606844 | 203800 | Alstrom syndrome | 98,27 |
| ALX1 | 601527 | 613456 | Frontonasal dysplasia 3 | 99,52 |
| ALX4 | 605420 | 613451 | Frontonasal dysplasia 2 | 99,07 |
| AMER1 | 300647 | 300373 | Osteopathia striata with cranial sclerosis | 99,96 |
| AMPD2 | 102771 | 615809 | Pontocerebellar hypoplasia, type 9 | 99,69 |
| AMT | 238310 | 605899 | Glycine encephalopathy | 99,69 |
| ANK3 | 600465 | 615493 | Mental retardation, autosomal recessive, 37 | 99,47 |
| ANKH | 605145 | 123000 | Craniometaphyseal dysplasia | 99,64 |
| ANKRD11 | 611192 | 148050 | KBG syndrome | 99,68 |
| AP1S1 | 603531 | 609313 | MEDNIK syndrome | 99,50 |
| AP1S2 | 300629 | 304340 | Mental retardation, X-linked syndromic 5 | 88,78 |
| AP4B1 | 607245 | 614066 | Spastic paraplegia 47, autosomal recessive | 99,61 |
| AP4M1 | 602296 | 612936 | Spastic paraplegia 50, autosomal recessive | 99,68 |
| AP4S1 | 607243 | 614067 | Spastic paraplegia 52, autosomal recessive | 99,48 |
| APTX | 606350 | 208920 | Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia | 99,02 |
| ARCN1 | 600820 | 617164 | Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay | 99,27 |
| ARFGEF2 | 605371 | 608097 | Periventricular heterotopia with microcephaly | 99,31 |
| ARG1 | 608313 | 207800 | Argininemia | 99,53 |
| ARHGAP31 | 610911 | 100300 | Adams-Oliver syndrome 1 | 99,60 |
| ARHGEF2 | 607560 | 617523 | Neurodevelopmental disorder with midbrain and hindbrain malformations | 99,64 |
| ARHGEF9 | 300429 | 300607 | Epileptic encephalopathy, early infantile, 8 | 97,26 |
| ARID1A | 603024 | 614607 | Coffin-Siris syndrome 2 | 97,02 |
| ARID1B | 614556 | 135900 | Coffin-Siris syndrome 1 | 97,57 |
| ARID2 | 609539 | 617808 | Coffis-Siris syndrome 6 | 99,33 |
| ARL13B | 608922 | 612291 | Joubert syndrome 8 | 98,36 |
| ARL6 | 608845 | 600151 | Bardet-Biedl syndrome 3 | 99,09 |
| ARSL | 300180 | 302950 | Chondrodysplasia punctata, X-linked recessive | 90,38 |
| ARX | 300382 | 300419 | Mental retardation, X-linked 29 and others | 65,25 |
| ASH1L | 607999 | - | Okamoto et al., AJMG 2017 : MCA/ID syndrome | 99,53 |
| ASNS | 108370 | 615574 | Asparagine synthetase deficiency | 99,42 |
| ASPA | 608034 | 271900 | Canavan disease | 99,17 |
| ASPM | 605481 | 608716 | Microcephaly 5, primary, autosomal recessive | 98,50 |
| ASXL1 | 612990 | 605039 | Bohring-Opitz syndrome | 95,03 |
| ASXL3 | 615115 | 615485 | Bainbridge-Ropers syndrome | 98,20 |
| ATIC | 601731 | 608688 | AICA-ribosiduria due to ATIC deficiency | 96,73 |
| ATP6V0A2 | 611716 | 219200 | Cutis laxa, autosomal recessive, type IIA | 99,12 |
| ATP7A | 300011 | 309400 | Menkes disease | 94,32 |
| ATR | 601215 | 210600 | Seckel syndrome 1 | 99,17 |
| ATRIP | 606605 | - | Ogi et al., PLoS Genet 2012 : Seckel syndrome | 95,19 |
| ATRX | 300032 | 309580 | Mental retardation-hypotonic facies syndrome, X-linked | 92,10 |
| AUH | 600529 | 250950 | 3-methylglutaconic aciduria, type I | 96,75 |
| AUTS2 | 607270 | 615834 | Mental retardation, autosomal dominant 26 | 97,55 |
| B3GLCT | 610308 | 261540 | Peters-plus syndrome | 92,21 |
| B4GALT1 | 137060 | 607091 | Congenital disorder of glycosylation, type IId | 98,41 |
| B4GALT7 | 604327 | 130070 | Ehlers-Danlos syndrome with short stature and limb anomalies | 83,36 |
| BBIP1 | 613605 | 615995 | Bardet-Biedl syndrome 18 | 99,64 |
| BBS1 | 209901 | 209900 | Bardet-Biedl syndrome 1 | 99,66 |
| BBS10 | 610148 | 615987 | Bardet-Biedl syndrome 10 | 99,57 |
| BBS12 | 610683 | 615989 | Bardet-Biedl syndrome 12 | 99,52 |
| BBS2 | 606151 | 615981 | Bardet-Biedl syndrome 2 | 99,32 |
| BBS4 | 600374 | 615982 | Bardet-Biedl syndrome 4 | 99,49 |
| BBS5 | 603650 | 615983 | Bardet-Biedl syndrome 5 | 98,49 |
| BBS7 | 607590 | 615984 | Bardet-Biedl syndrome 7 | 99,00 |
| BBS9 | 615986 | 615986 | Bardet-Biedl syndrome 9 | 98,68 |
| BCKDHB | 248611 | 248600 | Maple syrup urine disease, type Ib | 98,82 |
| BCL11A | 606557 | 617101 | Dias-Logan syndrome | 99,16 |
| BCOR | 300485 | 300166 | Microphthalmia, syndromic 2 | 96,85 |
| BCS1L | 603647 | 256000 | Leigh syndrome | 99,69 |
| BLM | 210900 | 210900 | Bloom syndrome | 99,07 |
| BRAF | 164757 | 613706 | Noonan syndrome 7 | 94,78 |
| BRPF1 | 602410 | 617333 | Intellectual developmental disorder with dysmorphic facies and ptosis | 99,67 |
| BRWD3 | 300553 | 300659 | Mental retardation, X-linked 93 | 93,74 |
| BSCL2 | 606158 | 615924 | Encephalopathy, progressive, with or without lipodystrophy | 99,67 |
| BTD | 609019 | 253260 | Biotinidase deficiency | 99,68 |
| BUB1B | 602860 | 257300 | Mosaic variegated aneuploidy syndrome 1 | 99,40 |

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|---|---|---|---|---|
| C12ORF57 | 615640 | - | Platzer et al., AJMG 2014 : RM, hypoplasie corps caleux, colobome, épilepsie. | 99,69 |
| C19ORF12 | 614297 | 614298 | Neurodegeneration with brain iron accumulation 4 | 99,69 |
| C2CD3 | 615944 | 615948 | Orofaciodigital syndrome XIV | 99,44 |
| C5ORF42 | 614571 | 614615 | Joubert syndrome 17 | 98,89 |
| CA8 | 114815 | 613227 | Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3 | 99,19 |
| CACNA1A | 601011 | 617106 | Epileptic encephalopathy, early infantile, 42 | 99,12 |
| CACNA1C | 114205 | 601005 | Timothy syndrome | 99,65 |
| CACNA1D | 114206 | 615474 | Primary aldosteronism, seizures, and neurologic abnormalities | 99,50 |
| CAMK2A | 114078 | 617798 | Mental retardation, autosomal dominant 53 | 99,67 |
| CAMK2B | 607707 | 617799 | Mental retardation, autosomal dominant 54 | 96,34 |
| CANT1 | 613165 | 251450 | Desbuquois dysplasia 1 | 99,60 |
| CASK | 300172 | 300749 | Mental retardation and microcephaly with pontine and cerebellar hypoplasia | 95,36 |
| CAV1 | 601047 | 606721 | Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome | 99,67 |
| CAVIN1 | 603198 | 613327 | Lipodystrophy, congenital generalized, type 4 | 99,31 |
| CBL | 165360 | 613563 | Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia | 99,54 |
| CBS | 613381 | 236200 | Homocystinuria, B6-responsive and nonresponsive types | 99,69 |
| CC2D1A | 610055 | 608443 | Mental retardation, autosomal recessive 3 | 99,64 |
| CC2D2A | 612013 | 612285 | Joubert syndrome 9 | 99,11 |
| CCDC22 | 300859 | 300963 | Ritscher-Schinzel syndrome 2 | 99,91 |
| CCDC39 | 613798 | 613807 | Ciliary dyskinesia, primary, 14 | 98,07 |
| CCDC78 | 614666 | 614807 | Myopathy, centronuclear, 4 | 99,69 |
| CCDC88C | 611204 | 236600 | Hydrocephalus, nonsyndromic, autosomal recessive | 99,38 |
| CD96 | 606037 | 211750 | C syndrome | 99,13 |
| CDC45 | 603465 | 617063 | Meier-Gorlin syndrome 7 | 99,57 |
| CDC6 | 602627 | 613805 | Meier-Gorlin syndrome 5 | 99,32 |
| CDH15 | 114019 | 612580 | Mental retardation, autosomal dominant 3 | 96,19 |
| CDK13 | 603309 | 617360 | Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder | 95,39 |
| CDK5RAP2 | 608201 | 604804 | Microcephaly 3, primary, autosomal recessive | 99,45 |
| CDKL5 | 300203 | 300672 | Epileptic encephalopathy, early infantile, 2 | 87,62 |
| CDON | 608707 | 614226 | Holoprosencephaly 11 | 98,90 |
| CDT1 | 605525 | 613804 | Meier-Gorlin syndrome 4 | 89,72 |
| CENPJ | 609279 | 608393 | Microcephaly 6, primary, autosomal recessive | 99,19 |
| CEP135 | 611423 | 614673 | Microcephaly 8, primary, autosomal recessive | 97,81 |
| CEP152 | 613529 | 614852 | Microcephaly 9, primary, autosomal recessive | 98,40 |
| CEP290 | 610142 | 610188 | Joubert syndrome 5 | 97,64 |
| CEP41 | 610523 | 614464 | Joubert syndrome 15 | 99,07 |
| CEP57 | 607951 | 614114 | Mosaic variegated aneuploidy syndrome 2 | 99,09 |
| CEP63 | 614724 | 614728 | Seckel syndrome 6 | 99,06 |
| CHAMP1 | 616327 | 616579 | Mental retardation, autosomal dominant 40 | 99,63 |
| CHAT | 118490 | 254210 | Myasthenic syndrome, congenital, 6, presynaptic | 97,70 |
| CHD2 | 602119 | 615369 | Epileptic encephalopathy, childhood-onset | 99,18 |
| CHD4 | 603277 | 617159 | Sifrim-Hitz-Weiss syndrome | 99,65 |
| CHD7 | 608892 | 214800 | CHARGE syndrome | 99,53 |
| CHD8 | 610528 | 615032 | Autism, susceptibility to, 18 | 99,58 |
| CHKB | 612395 | 602541 | Muscular dystrophy, congenital, megaconial type | 97,97 |
| CHMP1A | 164010 | 614961 | Pontocerebellar hypoplasia, type 8 | 99,59 |
| CHRNA4 | 118504 | 600513 | Epilepsy, nocturnal frontal lobe, 1 | 90,96 |
| CHRND | 100720 | 616322 | Myasthenic syndrome, congenital, 3B, fast-channel | 99,68 |
| CISD2 | 611507 | 604928 | Wolfram syndrome 2 | 98,61 |
| CLCN4 | 302910 | 300114 | Mental retardation, X-linked 49/15 | 99,27 |
| CLN3 | 607042 | 204200 | Ceroid lipofuscinosis, neuronal, 3 | 99,69 |
| CLN5 | 608102 | 256731 | Ceroid lipofuscinosis, neuronal, 5 | 97,69 |
| CLN6 | 606725 | 601780 | Ceroid lipofuscinosis, neuronal, 6 | 85,06 |
| CLN8 | 607837 | 600143 | Ceroid lipofuscinosis, neuronal, 8 | 99,69 |
| CLP1 | 608757 | 615803 | Pontocerebellar hypoplasia, type 10 | 99,68 |
| CLPB | 616254 | 616271 | 3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia | 99,62 |
| CNKSR2 | 300724 | - | Houge et al., Mol Syndromol 2012 : ID | 91,50 |
| CNTNAP2 | 604569 | 610042 | Pitt-Hopkins like syndrome 1 | 99,39 |
| COASY | 609855 | 615643 | Neurodegeneration with brain iron accumulation 6 | 99,68 |
| COG1 | 606973 | 611209 | Congenital disorder of glycosylation, type IIg | 98,34 |
| COG5 | 606821 | 613612 | Congenital disorder of glycosylation, type IIi | 99,11 |
| COG6 | 606977 | 614576 | Congenital disorder of glycosylation, type III | 98,39 |
| COG7 | 606978 | 608779 | Congenital disorder of glycosylation, type IIe | 99,66 |
| COG8 | 606979 | 611182 | Congenital disorder of glycosylation, type IIh | 97,38 |
| COL4A1 | 120130 | 175780 | Porencephaly 1 | 97,86 |
| COL4A2 | 120090 | 614483 | Porencephaly 2 | 99,62 |
| COL4A3BP | 604677 | 616351 | Mental retardation, autosomal dominant 34 | 99,15 |
| COLEC10 | 607620 | 616351 | Mental retardation, autosomal dominant 34 | 99,16 |
| COLEC11 | 612502 | 265050 | 3MC syndrome 2 | 99,67 |
| COQ2 | 609825 | 607426 | Coenzyme Q10 deficiency, primary, 1 | 94,52 |
| COQ4 | 612898 | 616276 | Coenzyme Q10 deficiency, primary, 7 | 98,74 |
| COQ6 | 614647 | 614650 | Coenzyme Q10 deficiency, primary, 6 | 99,60 |
| COQ7 | 601683 | 616733 | Coenzyme Q10 deficiency, primary, 8 | 99,40 |
| COQ8A | 606980 | 612016 | Coenzyme Q10 deficiency, primary, 4 | 99,69 |
| COQ9 | 612837 | 614654 | Coenzyme Q10 deficiency, primary, 5 | 98,23 |
| COX10 | 602125 | 256000 | Leigh syndrome due to mitochondrial COX4 deficiency | 99,65 |
| CRADD | 603454 | 614499 | Mental retardation, autosomal recessive 34, with variant lissencephaly | 99,63 |
| CRBN | 609262 | 607417 | Mental retardation, autosomal recessive 2 | 98,86 |
| CREBBP | 600140 | 180849 | Rubinstein-Taybi syndrome 1 | 99,62 |
| CSNK2A1 | 115440 | 617062 | Okur-Chung neurodevelopmental syndrome | 99,32 |
| CSPP1 | 611654 | 615636 | Joubert syndrome 21 | 98,95 |
| CTC1 | 613129 | 612199 | Cerebroretinal microangiopathy with calcifications and cysts | 99,57 |
| CTCF | 604167 | 615502 | Mental retardation, autosomal dominant 21 | 99,65 |
| CTNNB1 | 116806 | 615075 | Mental retardation, autosomal dominant 19 | 99,65 |
| CTSA | 613111 | 256540 | Galactosialidosis | 99,59 |
| CTSD | 116840 | 610127 | Ceroid lipofuscinosis, neuronal, 10 | 94,51 |
| CUL4B | 300304 | 300354 | Mental retardation, X-linked, syndromic 15 (Cabezas type) | 84,65 |
| CYB5R3 | 613213 | 250800 | Methemoglobinemia, type I | 88,44 |
| CYP27A1 | 606530 | 213700 | Cerebrotendinous xanthomatosis | 97,94 |
| D2HGDH | 609186 | 600721 | D-2-hydroxyglutaric aciduria | 98,50 |
| DAG1 | 128239 | 616538 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9 | 99,69 |
| DARS2 | 610956 | 611105 | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation | 99,20 |
| DCAF17 | 612515 | 241080 | Woodhouse-Sakati syndrome | 93,81 |
| DCC | 120470 | 157600 | Mirror movements 1 and/or agenesis of the corpus callosum | 99,55 |
| DCPS | 610534 | 616459 | Al-Raqad syndrome | 99,69 |
| DCX | 300121 | 300067 | Lissencephaly, X-linked | 96,94 |
| DDB2 | 600811 | 278740 | Xeroderma pigmentosum, group E, DDB-negative subtype | 99,69 |
| DDHD2 | 615003 | 615033 | Spastic paraplegia 54, autosomal recessive | 97,74 |
| DDX11 | 601150 | 613398 | Warsaw breakage syndrome | 99,66 |
| DDX3X | 300160 | 300958 | Mental retardation, X-linked 102 | 96,52 |
| DEAF1 | 602635 | 615828 | Mental retardation, autosomal dominant 24 | 92,63 |
| DHCR24 | 606418 | 602398 | Desmosterolosis | 99,34 |
| DHCR7 | 602858 | 270400 | Smith-Lemli-Opitz syndrome | 99,68 |
| DHFR | 126060 | 613839 | Megaloblastic anemia due to dihydrofolate reductase deficiency | 98,50 |

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| DHX30 | 616423 | - | Lessel et al., AJHG 2017 : global developmental delay (GDD), intellectual disability (ID), severe speech impairment and gait abnormalities. | 99,51 |
| DIAPH1 | 602121 | 616632 | Seizures, cortical blindness, microcephaly syndrome | 95,85 |
| DKC1 | 300126 | 305000 | Dyskeratosis congenita, X-linked | 91,60 |
| DLD | 238331 | 246900 | Dihydrolipoamide dehydrogenase deficiency | 99,08 |
| DLG3 | 300189 | 300850 | Mental retardation, X-linked 90 | 98,91 |
| DLL4 | 605185 | 616589 | Adams-Oliver syndrome 6 | 99,61 |
| DNAJC19 | 608977 | 610198 | 3-methylglutaconic aciduria, type V | 97,60 |
| DNM1 | 602377 | 616346 | Epileptic encephalopathy, early infantile, 31 | 96,84 |
| DNMT3A | 602769 | 615879 | Tatton-Brown-Rahman syndrome | 99,69 |
| DNMT3B | 602900 | 242860 | Immunodeficiency-centromeric instability-facial anomalies syndrome 1 | 99,69 |
| DOCK6 | 614194 | 614219 | Adams-Oliver syndrome 2 | 97,92 |
| DOCK8 | 611432 | 243700 | Hyper-IgE recurrent infection syndrome, autosomal recessive | 99,24 |
| DPAGT1 | 191350 | 608093 | Congenital disorder of glycosylation, type Ij | 99,68 |
| DPH1 | 603527 | 616901 | Developmental delay with short stature, dysmorphic features, and sparse hair | 99,67 |
| DPM1 | 603503 | 608799 | Congenital disorder of glycosylation, type Ie | 99,27 |
| DPP6 | 126141 | 616311 | Mental retardation, autosomal dominant 33 | 99,57 |
| DSCAM | 602523 | - | Wang et al., Nat Commun 2016 : Autisme | 99,29 |
| DYNC1H1 | 600112 | 614563 | Mental retardation, autosomal dominant 13 | 99,50 |
| DYRK1A | 600855 | 614104 | Mental retardation, autosomal dominant 7 | 99,53 |
| EARS2 | 612799 | 614924 | Combined oxidative phosphorylation deficiency 12 | 99,64 |
| EBF3 | 607407 | 617330 | Hypotonia, ataxia, and delayed development syndrome | 98,59 |
| EBP | 300205 | 300960 | MEND syndrome | 99,89 |
| ECHS1 | 602292 | 616277 | Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency | 98,49 |
| EDC3 | 609842 | 616460 | Mental retardation, autosomal recessive 50 | 99,68 |
| EEF1A2 | 602959 | 616393 | Mental retardation, autosomal dominant 38 | 88,74 |
| EFNB1 | 300035 | 304110 | Craniofrontonasal dysplasia | 99,39 |
| EFTUD2 | 603892 | 610536 | Mandibulofacial dysostosis, Guion-Almeida type | 99,44 |
| EHMT1 | 607001 | 610253 | Kleefstra syndrome | 95,40 |
| EIF2AK3 | 604032 | 226980 | Wolcott-Rallison syndrome | 94,12 |
| EIF2S3 | 300161 | 300987 | Mental retardation, X-linked, syndromic, Borck type | 97,88 |
| ELOVL4 | 605512 | 614457 | Ichthyosis, spastic quadriplegia, and mental retardation | 99,45 |
| ELP2 | 616054 | 617270 | Mental retardation, autosomal recessive 58 | 98,99 |
| EML1 | 602033 | 600348 | Band heterotopia | 96,28 |
| EMX2 | 600035 | 269160 | Schizencephaly | 99,48 |
| ENTPD1 | 601752 | 615683 | Spastic paraplegia 64, autosomal recessive | 99,05 |
| EOGT | 614789 | 615297 | Adams-Oliver syndrome 4 | 99,33 |
| EP300 | 602700 | 613684 | Rubinstein-Taybi syndrome 2 | 99,56 |
| EPB41L1 | 602879 | 614257 | Mental retardation, autosomal dominant 11 | 99,69 |
| EPG5 | 615068 | 242840 | Vici syndrome | 99,47 |
| ERCC2 | 126340 | 610756 | Cerebrooculofacioskeletal syndrome 2 | 99,67 |
| ERCC3 | 133510 | 610651 | Xeroderma pigmentosum, group B | 99,68 |
| ERCC4 | 133520 | 278760 | Xeroderma pigmentosum, type F/Cockayne syndrome | 99,35 |
| ERCC5 | 133530 | 278780 | Xeroderma pigmentosum, group G/Cockayne syndrome | 99,48 |
| ERCC6 | 609413 | 214150 | Cerebrooculofacioskeletal syndrome 1 | 99,07 |
| ERCC8 | 609412 | 216400 | Cockayne syndrome, type A | 99,27 |
| ERLIN2 | 611605 | 611225 | Spastic paraplegia 18, autosomal recessive | 99,32 |
| ESCO2 | 609353 | 268300 | Roberts syndrome | 98,50 |
| ETHE1 | 608451 | 602473 | Ethylmalonic encephalopathy | 99,60 |
| EXOSC3 | 606489 | 614678 | Pontocerebellar hypoplasia, type 1B | 99,53 |
| EZH2 | 601573 | 277590 | Weaver syndrome | 99,41 |
| EZR | 123900 | - | Riecken et al., Hum Mut 2015 : ID | 99,63 |
| FAM126A | 610531 | 610532 | Leukodystrophy, hypomyelinating, 5 | 98,81 |
| FARS2 | 611592 | 614946 | Combined oxidative phosphorylation deficiency 14 | 99,56 |
| FAT4 | 612411 | 615546 | Van Maldergem syndrome 2 | 99,39 |
| FBXL4 | 605654 | 615471 | Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type) | 99,46 |
| FBXO31 | 609102 | 615979 | Mental retardation, autosomal recessive 45 | 93,34 |
| FGD1 | 300546 | 305400 | Mental retardation, X-linked syndromic 16 | 98,23 |
| FGFR1 | 136350 | 147950 | Hypogonadotropic hypogonadism 2 with or without anosmia | 99,69 |
| FGFR2 | 176943 | 101400 | Saethre-Chotzen syndrome | 99,67 |
| FGFR3 | 134934 | 612247 | Crouzon syndrome with acanthosis nigricans | 95,86 |
| FH | 136850 | 606812 | Fumarase deficiency | 99,29 |
| FKRP | 606596 | 606612 | Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5 | 85,03 |
| FKTN | 607440 | 613152 | Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4 | 99,13 |
| FLNA | 300017 | 300049 | Heterotopia, periventricular | 99,73 |
| FMN2 | 606373 | 616193 | Mental retardation, autosomal recessive 47 | 99,05 |
| FMR1 | 309550 | 300623 | Fragile X tremor/ataxia syndrome | 94,48 |
| FOLR1 | 136430 | 613068 | Neurodegeneration due to cerebral folate transport deficiency | 99,69 |
| FOXG1 | 164874 | 613454 | Rett syndrome, congenital variant | 84,20 |
| FOXP1 | 605515 | 613670 | Mental retardation with language impairment and with or without autistic features | 99,55 |
| FOXP2 | 605317 | 602081 | Speech-language disorder-1 | 99,34 |
| FOXRED1 | 613622 | 256000 | Leigh syndrome due to mitochondrial complex I deficiency | 99,63 |
| FRAS1 | 607830 | 219000 | Fraser syndrome 1 | 99,57 |
| FREM2 | 608945 | 617666 | Fraser syndrome 2 | 99,23 |
| FRMPD4 | 300838 | 300983 | Mental retardation, X-linked 104 | 96,56 |
| FTCD | 606806 | 229100 | Glutamate formiminotransferase deficiency | 95,82 |
| FTL | 134790 | 606159 | Neurodegeneration with brain iron accumulation 3 | 99,55 |
| FTO | 610966 | 612938 | Growth retardation, developmental delay, facial dysmorphism | 99,47 |
| FTSJ1 | 300499 | 309549 | Mental retardation, X-linked 9/44 | 99,69 |
| FUCA1 | 612280 | 230000 | Fucosidosis | 98,18 |
| GABRA3 | 305660 | - | Niturad et al., Brain 2017 : range of epileptic seizure types, a varying degree of intellectual disability and developmental delay, sometimes with dysmorphic features or nystagmus. | 87,08 |
| GABRB3 | 137192 | 617113 | Epileptic encephalopathy, early infantile, 43 | 94,66 |
| GABRG2 | 137164 | 611277 | Epilepsy, generalized, with febrile seizures plus, type 3 | 99,57 |
| GALT | 606999 | 230400 | Galactosemia | 99,68 |
| GAMT | 601240 | 612736 | Cerebral creatine deficiency syndrome 2 | 93,42 |
| GAN | 605379 | 256850 | Giant axonal neuropathy-1 | 98,90 |
| GATAD2B | 614998 | 615074 | Mental retardation, autosomal dominant 18 | 99,69 |
| GATM | 602360 | 612718 | Cerebral creatine deficiency syndrome 3 | 94,01 |
| GBE1 | 607839 | 232500 | Glycogen storage disease IV | 98,82 |
| GCH1 | 600225 | 128230 | Dystonia, DOPA-responsive, with or without hyperphenylalaninemia | 93,92 |
| GCSH | 238330 | 605899 | Glycine encephalopathy | 86,02 |
| GDI1 | 300104 | 300849 | Mental retardation, X-linked 41 | 95,21 |
| GFAP | 137780 | 203450 | Alexander disease | 99,69 |
| GJC2 | 608803 | 608804 | Leukodystrophy, hypomyelinating, 2 | 73,70 |
| GLB1 | 611458 | 230500 | GM1-gangliosidosis, type I | 99,56 |
| GLDC | 238300 | 605899 | Glycine encephalopathy | 97,83 |
| GLI2 | 165230 | 610829 | Holoprosencephaly 9 | 98,60 |
| GLI3 | 165240 | 175700 | Greig cephalopolysyndactyly syndrome | 99,55 |
| GLUL | 138290 | 610015 | Glutamine deficiency, congenital | 99,68 |
| GLYCTK | 610516 | 220120 | D-glyceric aciduria | 99,69 |
| GM2A | 613109 | 272750 | GM2-gangliosidosis, AB variant | 99,68 |
| GMPPA | 615495 | 615510 | Alacrima, achalasia, and mental retardation syndrome | 99,69 |
| GMPPB | 615320 | 615351 | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14 | 99,64 |

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|---|---|---|---|---|
| GNAI3 | 139370 | 602483 | Auriculocondylar syndrome 1 | 99,08 |
| GNAO1 | 139311 | 615473 | Epileptic encephalopathy, early infantile, 17 | 99,66 |
| GNAS | 139320 | 103580 | Pseudohypoparathyroidism Ia | 97,96 |
| GNPAT | 602744 | 222765 | Rhizomelic chondrodysplasia punctata, type 2 | 97,06 |
| GNPTAB | 607840 | 252500 | Mucolipidosis II alpha/beta | 99,49 |
| GNPTG | 607838 | 252605 | Mucolipidosis III gamma | 90,19 |
| GNS | 607664 | 252940 | Mucopolysaccharidosis type IIID | 99,55 |
| GPC3 | 300037 | 312870 | Simpson-Golabi-Behmel syndrome, type 1 | 91,66 |
| GPHN | 603930 | 615501 | Molybdenum cofactor deficiency C | 99,58 |
| GPSM2 | 609245 | 604213 | Chudley-McCullough syndrome | 99,10 |
| GPT2 | 138210 | 616281 | Mental retardation, autosomal recessive 49 | 95,20 |
| GRIA3 | 305915 | 300699 | Mental retardation, X-linked 94 | 89,99 |
| GRID2 | 602368 | 616204 | Spinocerebellar ataxia, autosomal recessive 18 | 99,47 |
| GRIK2 | 138244 | 611092 | Mental retardation, autosomal recessive, 6 | 99,13 |
| GRIN1 | 138249 | 614254 | Mental retardation, autosomal dominant 8 | 99,54 |
| GRIN2A | 138253 | 245570 | Epilepsy, focal, with speech disorder and with or without mental retardation | 99,68 |
| GRIN2B | 138252 | 613970 | Mental retardation, autosomal dominant 6 | 99,66 |
| GRIP1 | 604597 | 617667 | Fraser syndrome 3 | 99,65 |
| GRM1 | 604473 | 617691 | Spinocerebellar ataxia 44 | 99,46 |
| GSS | 601002 | 266130 | Glutathione synthetase deficiency | 99,59 |
| GTF2E2 | 189964 | 616943 | Trichothiodystrophy 6, nonphotosensitive | 98,86 |
| GTF2H5 | 608780 | 616395 | Trichothiodystrophy 3, photosensitive | 99,64 |
| GTPBP3 | 608536 | 616198 | Combined oxidative phosphorylation deficiency 23 | 97,37 |
| GUSB | 611499 | 253220 | Mucopolysaccharidosis VII | 98,73 |
| HACE1 | 610876 | 616756 | Spastic paraplegia and psychomotor retardation with or without seizures | 96,37 |
| HCFC1 | 300019 | 616756 | Spastic paraplegia and psychomotor retardation with or without seizures | 99,76 |
| HCN1 | 602780 | 615871 | Epileptic encephalopathy, early infantile, 24 | 94,14 |
| HDAC4 | 605314 | - | Fisch et al., AJMG 2016 : ID | 99,68 |
| HDAC8 | 300269 | 300882 | Cornelia de Lange syndrome 5 | 99,17 |
| HECW2 | 617245 | 617268 | Neurodevelopmental disorder with hypotonia, seizures, and absent language | 99,50 |
| HEPACAM | 611642 | 613926 | Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation | 91,30 |
| HERC1 | 605109 | 617011 | Macrocephaly, dysmorphic facies, and psychomotor retardation | 99,49 |
| HERC2 | 605837 | 615516 | Mental retardation, autosomal recessive 38 | 99,66 |
| HESX1 | 601802 | 182230 | Growth hormone deficiency with pituitary anomalies | 98,36 |
| HEXA | 606869 | 272800 | Tay-Sachs disease | 99,67 |
| HEXB | 606873 | 268800 | Sandhoff disease, infantile, juvenile, and adult forms | 97,84 |
| HGSNAT | 610453 | 252930 | Mucopolysaccharidosis type IIIC (Sanfilippo C) | 94,00 |
| HIVEP2 | 143054 | 616977 | Mental retardation, autosomal dominant 43 | 99,60 |
| HLCS | 609018 | 253270 | Holocarboxylase synthetase deficiency | 99,60 |
| HNMT | 605238 | 616739 | Mental retardation, autosomal recessive 51 | 99,26 |
| HNRNPH2 | 300610 | 300986 | Mental retardation, X-linked, syndromic, Bain type | 99,85 |
| HNRNPK | 600712 | 616580 | Au-Kline syndrome | 99,17 |
| HNRNPU | 602869 | 617391 | Epileptic encephalopathy, early infantile, 54 | 98,33 |
| HOXA1 | 142955 | 601536 | Athabaskan brainstem dysgenesis syndrome | 99,65 |
| HPRT1 | 308000 | 300322 | Lesch-Nyhan syndrome | 75,78 |
| HRAS | 190020 | 218040 | Costello syndrome | 99,69 |
| HSD17B10 | 300256 | 300438 | HSD10 mitochondrial disease | 99,97 |
| HSPG2 | 142461 | 224410 | Dyssegmental dysplasia, Silverman-Handmaker type | 98,62 |
| HUWE1 | 300697 | 300706 | Mental retardation, X-linked syndromic, Turner type | 97,92 |
| IARS1 | 600709 | 617093 | Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy | 99,32 |
| IARS2 | 612801 | 616007 | Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia | 99,11 |
| IDS | 300823 | 309900 | Mucopolysaccharidosis II | 97,07 |
| IDUA | 252800 | 607015 | Mucopolysaccharidosis Ih/s | 77,72 |
| IER3IP1 | 609382 | 614231 | Microcephaly, epilepsy, and diabetes syndrome | 98,86 |
| IFIH1 | 606951 | 615846 | Aicardi-Goutieres syndrome 7 | 99,07 |
| IFT172 | 607386 | 615630 | Short-rib thoracic dysplasia 10 with or without polydactyly | 99,62 |
| IFT27 | 615870 | 615996 | Bardet-Biedl syndrome 19 | 99,69 |
| IFT57 | 606621 | - | Thevenon et al., Clin Genet 2016 : Oro-facial-digital syndrome | 98,66 |
| IGF1 | 147440 | 608747 | Growth retardation with deafness and mental retardation due to IGF1 deficiency | 99,63 |
| IL1RAPL1 | 300206 | 300143 | Mental retardation, X-linked 21/34 | 93,86 |
| INPP5E | 613037 | 213300 | Joubert syndrome 1 | 98,14 |
| INTU | 610621 | - | Bruel et al., JMG 2017 : oro-facial-digital syndrome | 98,49 |
| IQSEC2 | 300522 | 309530 | Mental retardation, X-linked 1/78 | 97,05 |
| IRX5 | 606195 | 611174 | Hamamy syndrome | 89,09 |
| ISPD | 614631 | 614643 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7 | 92,51 |
| ITGA7 | 600536 | 613204 | Muscular dystrophy, congenital, due to ITGA7 deficiency | 99,67 |
| ITPR1 | 147265 | 117360 | Spinocerebellar ataxia 29, congenital nonprogressive | 99,38 |
| JAM3 | 606871 | 613730 | Hemorrhagic destruction of the brain, subependymal calcification, and cataracts | 99,20 |
| KANSL1 | 612452 | 610443 | Koolen-De Vries syndrome | 99,63 |
| KAT6A | 601408 | 616268 | Mental retardation, autosomal dominant 32 | 99,60 |
| KAT6B | 605880 | 606170 | Genitopatellar syndrome | 99,21 |
| KCNC1 | 176258 | 616187 | Epilepsy, progressive myoclonic 7 | 99,05 |
| KCNC3 | 176264 | 605259 | Spinocerebellar ataxia 13 | 76,28 |
| KCNH1 | 603305 | 611816 | Temple-Baraitser syndrome | 98,56 |
| KCNJ10 | 602208 | 612780 | SESAME syndrome | 99,68 |
| KCNJ11 | 600937 | 606176 | Diabetes, permanent neonatal, with or without neurologic features | 99,69 |
| KCNK9 | 605874 | 612292 | Birk-Barel mental retardation dysmorphism syndrome | 99,67 |
| KCNMA1 | 600150 | 609446 | Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy | 99,54 |
| KCNQ2 | 602235 | 613720 | Epileptic encephalopathy, early infantile, 7 | 98,93 |
| KCNQ5 | 607357 | 617601 | Mental retardation, autosomal dominant 46 | 96,45 |
| KCNT1 | 608167 | 614959 | Epileptic encephalopathy, early infantile, 14 | 98,70 |
| KCTD7 | 611725 | 611726 | Epilepsy, progressive myoclonic 3, with or without intracellular inclusions | 95,20 |
| KDM1A | 609132 | 616728 | Cleft palate, psychomotor retardation, and distinctive facial features | 96,83 |
| KDM5C | 314690 | 300534 | Mental retardation, X-linked, syndromic, Claes-Jensen type | 99,87 |
| KDM6A | 300128 | 300867 | Kabuki syndrome 2 | 94,07 |
| KIAA0586 | 610178 | 616490 | Joubert syndrome 23 | 92,34 |
| KIAA0753 | 617112 | 617127 | Orofaciodigital syndrome XV | 99,23 |
| KIAA2022 | 300524 | 300912 | Mental retardation, X-linked 98 | 97,66 |
| KIF11 | 148760 | 152950 | Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation | 98,74 |
| KIF1A | 601255 | 614255 | Mental retardation, autosomal dominant 9 | 99,66 |
| KIFBP | 609367 | 609460 | Goldberg-Shprintzen megacolon syndrome | 96,44 |
| KIF4A | 300521 | 300923 | Mental retardation, X-linked 100 | 99,41 |
| KIF5A | 602821 | 617235 | Myoclonus, intractable, neonatal | 99,35 |
| KIF5C | 604593 | 615282 | Cortical dysplasia, complex, with other brain malformations 2 | 96,19 |
| KIF7 | 611254 | 200990 | Joubert syndrome 12 | 99,42 |
| KIRREL3 | 607761 | 612581 | Mental retardation, autosomal dominant 4 | 99,66 |
| KLC2 | 611729 | 609541 | Spastic paraplegia, optic atrophy, and neuropathy | 99,69 |
| KMT2A | 159555 | 605130 | Wiedemann-Steiner syndrome | 97,93 |
| KMT2C | 606833 | 617768 | Kleefstra syndrome 2 | 99,31 |
| KMT2D | 602113 | 147920 | Kabuki syndrome 1 | 99,67 |

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|---|---|---|---|---|
| KMT2E | 608444 | - | Dong et al., Cell Rep 2014 : trouble du spectre autistique | 98,54 |
| KMT5B | 610881 | - | Stessman et al., Nat Genet 2017 : autisme et retard développemental | 99,31 |
| KNL1 | 609173 | 604321 | Microcephaly 4, primary, autosomal recessive | 92,70 |
| KPTN | 615620 | 615637 | Mental retardation, autosomal recessive 41 | 99,37 |
| KRAS | 190070 | 609942 | Noonan syndrome 3 | 99,32 |
| L1CAM | 308840 | 303350 | CRASH syndrome | 99,66 |
| L2HGDH | 609584 | 236792 | L-2-hydroxyglutaric aciduria | 98,45 |
| LAMA1 | 150320 | 615960 | Poretti-Boltshauser syndrome | 99,13 |
| LAMA2 | 156225 | 607855 | Muscular dystrophy, congenital, due to partial LAMA2 deficiency | 99,46 |
| LAMB1 | 150240 | 615191 | Lissencephaly 5 | 99,50 |
| LAMC3 | 604349 | 614115 | Cortical malformations, occipital | 97,99 |
| LAMP2 | 309060 | 300257 | Danon disease | 93,32 |
| LARGE1 | 603590 | 608840 | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6 | 99,64 |
| LARP7 | 612026 | 615071 | Alazami syndrome | 97,40 |
| LAS1L | 300964 | 309585 | Wilson-Turner syndrome | 99,76 |
| LIAS | 607031 | 614462 | Hyperglycinemia, lactic acidosis, and seizures | 98,94 |
| LIG4 | 601837 | 606593 | LIG4 syndrome | 99,55 |
| LINS1 | 610350 | 614340 | Mental retardation, autosomal recessive 27 | 98,65 |
| LIPT1 | 610284 | 616299 | Lipoyltransferase 1 deficiency | 98,69 |
| LMBRD1 | 612625 | 277380 | Methylmalonic aciduria and homocystinuria, cblF type | 96,94 |
| LMNA | 150330 | 616516 | Emery-Dreifuss muscular dystrophy 3, AR | 99,49 |
| LRP2 | 600073 | 222448 | Donnai-Barrow syndrome | 99,40 |
| LZTFL1 | 606568 | 615994 | Bardet-Biedl syndrome 17 | 98,67 |
| LZTR1 | 600574 | 616564 | Noonan syndrome 10 | 99,56 |
| MAGEL2 | 605283 | 615547 | Schaaf-Yang syndrome | 98,06 |
| MAN1B1 | 604346 | 614202 | Mental retardation, autosomal recessive 15 | 98,88 |
| MAN2B1 | 609458 | 248500 | Mannosidosis, alpha-, types I and II | 99,40 |
| MANBA | 609489 | 248510 | Mannosidosis, beta | 99,01 |
| MAOA | 309850 | 300615 | Brunner syndrome | 85,51 |
| MAP2K1 | 176872 | 615279 | Cardiofaciocutaneous syndrome 3 | 99,62 |
| MAP2K2 | 601263 | 615280 | Cardiofaciocutaneous syndrome 4 | 99,01 |
| MASP1 | 600521 | 257920 | 3MC syndrome 1 | 99,59 |
| MAT1A | 610550 | 250850 | Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency | 99,66 |
| MBD5 | 611472 | 156200 | Mental retardation, autosomal dominant 1 | 99,52 |
| MBOAT7 | 606048 | 617188 | Mental retardation, autosomal recessive 57 | 99,50 |
| MBTPS2 | 300294 | 308205 | IFAP syndrome with or without BRESHECK syndrome | 96,46 |
| MCCC1 | 609010 | 210200 | 3-Methylcrotonyl-CoA carboxylase 1 deficiency | 99,50 |
| MCOLN1 | 605248 | 252650 | Mucolipidosis IV | 93,43 |
| MCPH1 | 607117 | 251200 | Microcephaly 1, primary, autosomal recessive | 98,40 |
| MECP2 | 300005 | 312750 | Rett syndrome | 89,77 |
| MED12 | 300188 | 305450 | Opitz-Kaveggia syndrome | 99,82 |
| MED13L | 608771 | 616789 | Mental retardation and distinctive facial features with or without cardiac defects | 99,52 |
| MED17 | 603810 | 613668 | Microcephaly, postnatal progressive, with seizures and brain atrophy | 98,79 |
| MED23 | 605042 | 614249 | Mental retardation, autosomal recessive 18 | 98,87 |
| MEF2C | 600662 | 613443 | Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations | 99,52 |
| MEIS2 | 601740 | 600987 | Cleft palate, cardiac defects, and mental retardation | 99,65 |
| METTL23 | 615262 | 615942 | Mental retardation, autosomal recessive 44 | 99,62 |
| MFSD8 | 611124 | 610951 | Ceroid lipofuscinosis, neuronal, 7 | 99,30 |
| MID1 | 300552 | 300000 | Opitz GBBB syndrome, type I | 97,46 |
| MKKS | 604896 | 605231 | Bardet-Biedl syndrome 6 | 99,58 |
| MKS1 | 609883 | 615990 | Bardet-Biedl syndrome 13 | 99,54 |
| MLYCD | 606761 | 248360 | Malonyl-CoA decarboxylase deficiency | 87,33 |
| MMACHC | 609831 | 277400 | Methylmalonic aciduria and homocystinuria, cblC type | 99,68 |
| MOCS1 | 603707 | 252150 | Molybdenum cofactor deficiency A | 99,68 |
| MOCS2 | 603708 | 252160 | Molybdenum cofactor deficiency B | 99,48 |
| MOGS | 601336 | 606056 | Congenital disorder of glycosylation, type IIb | 92,25 |
| MPDU1 | 604041 | 609180 | Congenital disorder of glycosylation, type If | 99,69 |
| MPDZ | 603785 | 615219 | Hydrocephalus, nonsyndromic, autosomal recessive 2 | 99,22 |
| MPLKIP | 609188 | 234050 | Trichothiodystrophy 4, nonphotosensitive | 99,63 |
| MTFMT | 611766 | 614947 | Combined oxidative phosphorylation deficiency 15 | 97,00 |
| MTHFR | 607093 | 236250 | Homocystinuria due to MTHFR deficiency | 99,58 |
| MTM1 | 300415 | 310400 | Myotubular myopathy, X-linked | 94,11 |
| MTOR | 601231 | 616638 | Smith-Kingsmore syndrome | 99,61 |
| MTR | 156570 | 250940 | Homocystinuria-megaloblastic anemia, cblG complementation type | 99,55 |
| MTRR | 602568 | 236270 | Homocystinuria-megaloblastic anemia, cbl E type | 99,33 |
| MVK | 251170 | 610377 | Mevalonic aciduria | 99,69 |
| MYCN | 164840 | 164280 | Feingold syndrome 1 | 82,91 |
| MYO5A | 160777 | 214450 | Griscelli syndrome, type 1 | 97,60 |
| MYT1L | 613084 | 616521 | Mental retardation, autosomal dominant 39 | 99,59 |
| NAA10 | 300013 | 300855 | Ogden syndrome | 87,47 |
| NACC1 | 610672 | 617393 | Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination | 99,66 |
| NAGA | 104170 | 609241 | Schindler disease, type I | 99,67 |
| NAGLU | 609701 | 252920 | Mucopolysaccharidosis type IIIB (Sanfilippo B) | 84,28 |
| NALCN | 611549 | 616266 | Congenital contractures of the limbs and face, hypotonia, and developmental delay | 99,38 |
| NARS2 | 612803 | 616239 | Combined oxidative phosphorylation deficiency 24 | 98,96 |
| NBN | 602667 | 251260 | Nijmegen breakage syndrome | 98,67 |
| NDE1 | 609449 | 614019 | Lissencephaly 4 (with microcephaly) | 99,69 |
| NDP | 300658 | 310600 | Norrie disease | 99,56 |
| NDST1 | 600853 | 616116 | Mental retardation, autosomal recessive 46 | 99,67 |
| NDUFA1 | 300078 | 252010 | Mitochondrial complex I deficiency | 97,69 |
| NDUFA11 | 612638 | 252010 | Mitochondrial complex I deficiency | 99,69 |
| NDUFA12 | 614530 | 256000 | Leigh syndrome due to mitochondrial complex 1 deficiency | 99,64 |
| NDUFA2 | 602137 | 256000 | Leigh syndrome due to mitochondrial complex I deficiency | 99,69 |
| NDUFA9 | 603834 | 256000 | Leigh syndrome due to mitochondrial complex I deficiency | 99,27 |
| NDUFAF6 | 612392 | 256000 | Leigh syndrome due to mitochondrial complex I deficiency | 89,55 |
| NDUFS2 | 602985 | 252010 | Mitochondrial complex I deficiency | 99,25 |
| NDUFS3 | 603846 | 256000 | Leigh syndrome due to mitochondrial complex I deficiency | 99,67 |
| NDUFS4 | 602694 | 252010 | Mitochondrial complex I deficiency | 99,05 |
| NDUFS7 | 601825 | 256000 | Leigh syndrome | 99,38 |
| NEDD4L | 606384 | 617201 | Periventricular nodular heterotopia 7 | 98,92 |
| NFIA | 600727 | 613735 | Brain malformations and urinary tract defects | 99,44 |
| NFIX | 164005 | 614753 | Sotos syndrome 2 | 99,64 |
| NGF | 162030 | 608654 | Neuropathy, hereditary sensory and autonomic, type V | 99,66 |
| NHEJ1 | 611290 | 611291 | Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation | 99,67 |
| NHP2 | 606470 | 613987 | Dyskeratosis congenita, autosomal recessive 2 | 99,64 |
| NHS | 300457 | 302350 | Nance-Horan syndrome | 88,25 |
| NIN | 608684 | 614851 | Seckel syndrome 7 | 99,20 |
| NIPBL | 608667 | 122470 | Cornelia de Lange syndrome 1 | 98,53 |
| NKX2-1 | 600635 | 610978 | Choreoathetosis, hypothyroidism, and neonatal respiratory distress | 92,05 |
| NONO | 300084 | 300967 | Mental retardation, X-linked, syndromic 34 | 98,37 |
| NOTCH1 | 190198 | 616028 | Adams-Oliver syndrome 5 | 96,74 |
| NPC1 | 607623 | 257220 | Niemann-Pick disease, type C1 | 98,34 |
| NPC2 | 601015 | 607625 | Niemann-pick disease, type C2 | 99,67 |
| NPHP1 | 607100 | 609583 | Joubert syndrome 4 | 99,24 |
| NPHP3 | 608002 | 267010 | Meckel syndrome 7 | 98,15 |

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|---|---|---|---|---|
| NR2F1 | 132890 | 615722 | Bosch-Boonstra-Schaaf optic atrophy syndrome | 86,66 |
| NR5A1 | 184757 | 612964 | Adrenocortical insufficiency | 99,25 |
| NRAS | 164790 | 613224 | Noonan syndrome 6 | 99,61 |
| NRXN1 | 600565 | 614325 | Pitt-Hopkins-like syndrome 2 | 99,52 |
| NSD1 | 606681 | 117550 | Sotos syndrome 1 | 99,26 |
| NSDHL | 300275 | 308050 | CHILD syndrome | 99,39 |
| NSUN2 | 610916 | 611091 | Mental retardation, autosomal recessive 5 | 95,86 |
| OCLN | 602876 | 251290 | Pseudo-TORCH syndrome 1 | 99,47 |
| OCRL | 300535 | 309000 | Lowe syndrome | 92,17 |
| OFD1 | 300170 | 311200 | Orofaciodigital syndrome I | 92,99 |
| OGT | 300255 | 300997 | Mental retardation, X-linked 106 | 96,41 |
| OPHN1 | 300127 | 300486 | Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance | 90,64 |
| ORC1 | 601902 | 224690 | Meier-Gorlin syndrome 1 | 99,49 |
| ORC4 | 603056 | 613800 | Meier-Gorlin syndrome 2 | 98,60 |
| ORC6 | 607213 | 613803 | Meier-Gorlin syndrome 3 | 98,10 |
| OSGEP | 610107 | - | Braun et al., Nat Genet 2017 : Galloway-Mowat syndrome | 99,65 |
| OTC | 300461 | 311250 | Ornithine transcarbamylase deficiency | 92,10 |
| OTUD6B | 612021 | 617452 | Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies | 97,03 |
| PACS1 | 607492 | 615009 | Schuurs-Hoeijmakers syndrome | 97,17 |
| PAFAH1B1 | 601545 | 607432 | Lissencephaly 1 | 98,98 |
| PAH | 612349 | 261600 | Phenylketonuria | 99,66 |
| PAK3 | 300142 | 300558 | Mental retardation, X-linked 30/47 | 87,10 |
| PANK2 | 606157 | 234200 | Neurodegeneration with brain iron accumulation 1 | 95,75 |
| PAX1 | 167411 | 615560 | Otofaciocervical syndrome 2 | 82,57 |
| PAX3 | 606597 | 122880 | Craniofacial-deafness-hand syndrome | 99,68 |
| PAX6 | 607108 | 106210 | Aniridia | 99,51 |
| PC | 608786 | 266150 | Pyruvate carboxylase deficiency | 99,69 |
| PCDH19 | 300460 | 300088 | Epileptic encephalopathy, early infantile, 9 | 99,22 |
| PCGF2 | 600346 | - | Fitzgerald et al., Nature 2015 : retard mental, retard dévelopemental | 99,69 |
| PCNT | 605925 | 210720 | Microcephalic osteodysplastic primordial dwarfism, type II | 99,50 |
| PDE4D | 600129 | 614613 | Acrodysostosis 2, with or without hormone resistance | 97,76 |
| PDHA1 | 300502 | 312170 | Pyruvate dehydrogenase E1-alpha deficiency | 97,16 |
| PDHB | 179060 | 614111 | Pyruvate dehydrogenase E1-beta deficiency | 99,40 |
| PDHX | 608769 | 245349 | Lacticacidemia due to PDX1 deficiency | 98,49 |
| PDSS1 | 607429 | 614651 | Coenzyme Q10 deficiency, primary, 2 | 82,84 |
| PDSS2 | 610564 | 614652 | Coenzyme Q10 deficiency, primary, 3 | 98,95 |
| PEPD | 613230 | 170100 | Prolidase deficiency | 99,61 |
| PEX1 | 602136 | 214100 | Peroxisome biogenesis disorder 1A (Zellweger) | 98,83 |
| PEX10 | 602859 | 614870 | Peroxisome biogenesis disorder 6A (Zellweger) | 83,86 |
| PEX11B | 603867 | 614920 | Peroxisome biogenesis disorder 14B | 99,63 |
| PEX12 | 601758 | 614859 | Peroxisome biogenesis disorder 3A (Zellweger) | 99,40 |
| PEX13 | 601789 | 614883 | Peroxisome biogenesis disorder 11A (Zellweger) | 99,25 |
| PEX14 | 601791 | 614887 | Peroxisome biogenesis disorder 13A (Zellweger) | 99,66 |
| PEX16 | 603360 | 614876 | Peroxisome biogenesis disorder 8A (Zellweger) | 99,69 |
| PEX19 | 600279 | 614886 | Peroxisome biogenesis disorder 12A (Zellweger) | 99,60 |
| PEX2 | 170993 | 614866 | Peroxisome biogenesis disorder 5A (Zellweger) | 99,69 |
| PEX26 | 608666 | 614872 | Peroxisome biogenesis disorder 7A (Zellweger) | 96,96 |
| PEX3 | 603164 | 614882 | Peroxisome biogenesis disorder 10A (Zellweger) | 98,89 |
| PEX5 | 600414 | 214110 | Peroxisome biogenesis disorder 2A (Zellweger) | 99,15 |
| PEX6 | 601498 | 614862 | Peroxisome biogenesis disorder 4A (Zellweger) | 98,57 |
| PEX7 | 601757 | 614879 | Peroxisome biogenesis disorder 9B | 90,36 |
| PGAP1 | 611655 | 615802 | Mental retardation, autosomal recessive 42 | 97,25 |
| PGAP2 | 615187 | 614207 | Hyperphosphatasia with mental retardation syndrome 3 | 99,68 |
| PGAP3 | 611801 | 615716 | Hyperphosphatasia with mental retardation syndrome 4 | 99,15 |
| PGK1 | 311800 | 300653 | Phosphoglycerate kinase 1 deficiency | 99,16 |
| PHC1 | 602978 | 615414 | Microcephaly 11, primary, autosomal recessive | 99,66 |
| PHF6 | 300414 | 301900 | Borjeson-Forssman-Lehmann syndrome | 84,23 |
| PHF8 | 300560 | 300263 | Mental retardation syndrome, X-linked, Siderius type | 99,04 |
| PHGDH | 606879 | 601815 | Phosphoglycerate dehydrogenase deficiency | 99,67 |
| PHIP | 612870 | - | Webster et al., Cold Spring Harb Mol Case Stud. 2016 : DD, ID, obésité et dysmorphies | 95,93 |
| PIGA | 311770 | 300868 | Multiple congenital anomalies-hypotonia-seizures syndrome 2 | 92,30 |
| PIGG | 616918 | 616917 | Mental retardation, autosomal recessive 53 | 99,54 |
| PIGL | 605947 | 280000 | CHIME syndrome | 99,59 |
| PIGN | 606097 | 614080 | Multiple congenital anomalies-hypotonia-seizures syndrome 1 | 97,78 |
| PIGO | 614730 | 614749 | Hyperphosphatasia with mental retardation syndrome 2 | 99,67 |
| PIGT | 610272 | 615398 | Multiple congenital anomalies-hypotonia-seizures syndrome 3 | 99,63 |
| PIGV | 610274 | 239300 | Hyperphosphatasia with mental retardation syndrome 1 | 99,66 |
| PIGW | 610275 | 616025 | Hyperphosphatasia with mental retardation syndrome 5 | 99,68 |
| PIGY | 610662 | 616809 | Hyperphosphatasia with mental retardation syndrome 6 | 99,66 |
| PIK3R2 | 603157 | 603387 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1 | 91,70 |
| PLA2G6 | 603604 | 610217 | Neurodegeneration with brain iron accumulation 2B | 99,69 |
| PLCB1 | 607120 | 613722 | Epileptic encephalopathy, early infantile, 12 | 99,15 |
| PLCB4 | 600810 | 614669 | Auriculocondylar syndrome 2 | 99,09 |
| PLOD1 | 153454 | 225400 | Ehlers-Danlos syndrome, type VI | 98,58 |
| PLP1 | 300401 | 312080 | Pelizaeus-Merzbacher disease | 99,86 |
| PMM2 | 601785 | 212065 | Congenital disorder of glycosylation, type Ia | 98,47 |
| PNKP | 605610 | 613402 | Microcephaly, seizures, and developmental delay | 99,68 |
| PNP | 164050 | 613179 | Immunodeficiency due to purine nucleoside phosphorylase deficiency | 99,66 |
| POC1A | 614783 | 614813 | Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis | 99,26 |
| POGZ | 614787 | 616364 | White-Sutton syndrome | 98,81 |
| POLG | 174763 | 613662 | Mitochondrial DNA depletion syndrome 4B (MNGIE type) | 99,59 |
| POLH | 603968 | 278750 | Xeroderma pigmentosum, variant type | 99,55 |
| POLR3A | 614258 | 607694 | Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism | 99,40 |
| POLR3B | 614366 | 614381 | Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism | 99,29 |
| POMGNT1 | 606822 | 613151 | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3 | 99,66 |
| POMGNT2 | 614828 | 614830 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8 | 99,69 |
| POMK | 615247 | 615249 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12 | 99,66 |
| POMT1 | 607423 | 613155 | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1 | 99,56 |
| POMT2 | 607439 | 613156 | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2 | 98,42 |
| PORCN | 300651 | 305600 | Focal dermal hypoplasia | 99,91 |
| PPM1D | 605100 | 617450 | Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold | 98,20 |
| PPP1CB | 600590 | 617506 | Noonan syndrome-like disorder with loose anagen hair 2 | 99,40 |
| PPP2R1A | 605983 | 616362 | Mental retardation, autosomal dominant 36 | 99,69 |
| PPP2R5D | 601646 | 616355 | Mental retardation, autosomal dominant 35 | 99,64 |
| PPT1 | 600722 | 256730 | Ceroid lipofuscinosis, neuronal, 1 | 99,67 |
| PQBP1 | 300463 | 309500 | Renpenning syndrome | 99,90 |
| PRICKLE1 | 608500 | 612437 | Epilepsy, progressive myoclonic 1B | 99,67 |
| PRMT7 | 610087 | 617157 | Short stature, brachydactyly, intellectual developmental disability, and seizures | 99,21 |
| PRPS1 | 311850 | 301835 | Arts syndrome | 98,61 |
| PRSS12 | 606709 | 249500 | Mental retardation, autosomal recessive 1 | 98,69 |

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|---|---|---|---|---|
| PSAP | 176801 | 611722 | Krabbe disease, atypical | 99,68 |
| PSMD12 | 604450 | 617516 | Stankiewicz-Isidor syndrome | 99,44 |
| PTCH1 | 601309 | 610828 | Holoprosencephaly 7 | 97,64 |
| PTCHD1 | 300828 | 300830 | Autism, susceptibility to, X-linked 4 | 99,37 |
| PTDSS1 | 612792 | 151050 | Lenz-Majewski hyperostotic dwarfism | 99,56 |
| PTEN | 601728 | 158350 | Cowden syndrome 1 | 98,68 |
| PTPN11 | 176876 | 151100 | LEOPARD syndrome 1 | 97,30 |
| PUF60 | 604819 | 615583 | Verheij syndrome | 99,56 |
| PURA | 600473 | 616158 | Mental retardation, autosomal dominant 31 | 84,89 |
| PUS1 | 608109 | 600462 | Myopathy, lactic acidosis, and sideroblastic anemia 1 | 93,31 |
| PYCR1 | 179035 | 612940 | Cutis laxa, autosomal recessive, type IIB | 99,67 |
| QDPR | 612676 | 261630 | Hyperphenylalaninemia, BH4-deficient, C | 96,67 |
| RAB18 | 602207 | 614222 | Warburg micro syndrome 3 | 98,43 |
| RAB27A | 603868 | 607624 | Griscelli syndrome, type 2 | 95,80 |
| RAB39B | 300774 | 300271 | Mental retardation, X-linked 72 | 99,96 |
| RAB3GAP1 | 602536 | 600118 | Warburg micro syndrome 1 | 99,24 |
| RAB3GAP2 | 609275 | 212720 | Martsolf syndrome | 99,14 |
| RAC1 | 602048 | 617751 | Mental retardation, autosomal dominant 48 | 87,43 |
| RAD21 | 606462 | 614701 | Cornelia de Lange syndrome 4 | 99,34 |
| RAF1 | 164760 | 611553 | Noonan syndrome 5 | 99,65 |
| RAI1 | 607642 | 182290 | Smith-Magenis syndrome | 97,85 |
| RARB | 180220 | 615524 | Microphthalmia, syndromic 12 | 99,52 |
| RARS2 | 611524 | 611523 | Pontocerebellar hypoplasia, type 6 | 99,20 |
| RASA2 | 601589 | - | Aoki et al., JHG 2016 : RASopathie | 95,15 |
| RAX | 601881 | 611038 | Microphthalmia, isolated 3 | 78,51 |
| RBBP8 | 604124 | 606744 | Seckel syndrome 2 | 96,69 |
| RBM10 | 300080 | 311900 | TARP syndrome | 99,86 |
| RBM28 | 612074 | 612079 | Alopecia, neurologic defects, and endocrinopathy syndrome | 99,55 |
| RBM8A | 605313 | 274000 | Thrombocytopenia-absent radius syndrome | 99,68 |
| RBPJ | 147183 | 614814 | Adams-Oliver syndrome 3 | 99,36 |
| RECQL4 | 603780 | 218600 | Baller-Gerold syndrome | 91,13 |
| RELN | 600514 | 257320 | Lissencephaly 2 (Norman-Roberts type) | 99,47 |
| RERE | 605226 | 616975 | Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart | 99,45 |
| RFT1 | 611908 | 612015 | Congenital disorder of glycosylation, type In | 97,48 |
| RIT1 | 609591 | 615355 | Noonan syndrome 8 | 99,66 |
| RMND1 | 614917 | 614922 | Combined oxidative phosphorylation deficiency 11 | 98,79 |
| RNASEH2A | 606034 | 610333 | Aicardi-Goutieres syndrome 4 | 99,69 |
| RNASEH2B | 610326 | 610181 | Aicardi-Goutieres syndrome 2 | 90,13 |
| RNASEH2C | 610330 | 610329 | Aicardi-Goutieres syndrome 3 | 96,74 |
| RNASET2 | 612944 | 612951 | Leukoencephalopathy, cystic, without megalencephaly | 99,45 |
| RNF113A | 300951 | 300953 | Trichothiodystrophy 5, nonphotosensitive | 99,85 |
| ROGDI | 614574 | 226750 | Kohlschutter-Tonz syndrome | 90,00 |
| RPGRIP1L | 610937 | 611560 | Joubert syndrome 7 | 95,44 |
| RPIA | 180430 | 608611 | Ribose 5-phosphate isomerase deficiency | 98,01 |
| RPS6KA3 | 300075 | 303600 | Coffin-Lowry syndrome | 85,79 |
| RRAS | 165090 | - | Flex et al., HMG 2014 : RASopathie | 94,39 |
| RTEL1 | 608833 | 615190 | Dyskeratosis congenita, autosomal recessive 5 | 99,69 |
| RTTN | 610436 | 614833 | Microcephaly, short stature, and polymicrogyria with seizures | 98,71 |
| SALL1 | 602218 | 107480 | Townes-Brocks branchiootorenal-like syndrome | 99,68 |
| SAMHD1 | 606754 | 612952 | Aicardi-Goutieres syndrome 5 | 99,47 |
| SATB2 | 608148 | 612313 | Glass syndrome | 99,46 |
| SC5D | 602286 | 607330 | Lathosterolosis | 99,08 |
| SCN1A | 182389 | 607208 | Epileptic encephalopathy, early infantile, 6 (Dravet syndrome) | 98,95 |
| SCN2A | 182390 | 613721 | Epileptic encephalopathy, early infantile, 11 | 98,85 |
| SCN8A | 600702 | 614558 | Epileptic encephalopathy, early infantile, 13 | 99,37 |
| SCO2 | 604272 | 604377 | Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1 | 99,68 |
| SDCCAG8 | 613524 | 615993 | Bardet-Biedl syndrome 16 | 99,19 |
| SDHAF1 | 612848 | 252011 | Mitochondrial complex II deficiency | 66,11 |
| SEPSECS | 613009 | 613811 | Pontocerebellar hypoplasia type 2D | 99,07 |
| SERAC1 | 614725 | 614739 | 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome | 98,98 |
| SETBP1 | 611060 | 616078 | Mental retardation, autosomal dominant 29 | 98,39 |
| SETD2 | 612778 | 616831 | Luscan-Lumish syndrome | 99,30 |
| SETD5 | 615743 | 615761 | Mental retardation, autosomal dominant 23 | 99,55 |
| SGSH | 605270 | 252900 | Mucopolysaccharidosis type IIIA (Sanfilippo A) | 96,47 |
| SHANK2 | 603290 | 613436 | Autism susceptibility 17 | 83,77 |
| SHANK3 | 606230 | 606232 | Phelan-McDermid syndrome | 78,63 |
| SHH | 600725 | 142945 | Holoprosencephaly 3 | 90,25 |
| SHOC2 | 602775 | 607721 | Noonan-like syndrome with loose anagen hair | 97,89 |
| SHROOM4 | 300579 | 300434 | Stocco dos Santos X-linked mental retardation syndrome | 99,52 |
| SIL1 | 608005 | 248800 | Marinesco-Sjogren syndrome | 99,65 |
| SIN3A | 607776 | 613406 | Witteveen-Kolk syndrome | 99,56 |
| SIX3 | 603714 | 157170 | Holoprosencephaly 2 | 89,54 |
| SKI | 164780 | 182212 | Shprintzen-Goldberg syndrome | 93,23 |
| SLC12A6 | 604878 | 218000 | Agenesis of the corpus callosum with peripheral neuropathy | 99,13 |
| SLC16A2 | 300095 | 300523 | Allan-Herndon-Dudley syndrome | 99,71 |
| SLC17A5 | 604322 | 604369 | Salla disease | 98,53 |
| SLC19A3 | 606152 | 607483 | Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2) | 99,50 |
| SLC1A4 | 600229 | 616657 | Spastic tetraplegia, thin corpus callosum, and progressive microcephaly | 97,40 |
| SLC25A1 | 190315 | 615182 | Combined D-2- and L-2-hydroxyglutaric aciduria | 86,05 |
| SLC25A12 | 603667 | 612949 | Epileptic encephalopathy, early infantile, 39 | 99,45 |
| SLC25A15 | 603861 | 238970 | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome | 99,57 |
| SLC25A19 | 606521 | 607196 | Microcephaly, Amish type | 99,66 |
| SLC25A22 | 609302 | 609304 | Epileptic encephalopathy, early infantile, 3 | 99,68 |
| SLC2A1 | 138140 | 606777 | GLUT1 deficiency syndrome 1, infantile onset, severe | 93,34 |
| SLC33A1 | 603690 | 614482 | Congenital cataracts, hearing loss, and neurodegeneration | 98,70 |
| SLC35A2 | 314375 | 300896 | Congenital disorder of glycosylation, type IIm | 99,98 |
| SLC35C1 | 605881 | 266265 | Congenital disorder of glycosylation, type IIc | 99,69 |
| SLC45A1 | 605763 | 617532 | Intellectual developmental disorder with neuropsychiatric features | 92,61 |
| SLC46A1 | 611672 | 229050 | Folate malabsorption, hereditary | 97,75 |
| SLC4A4 | 603345 | 604278 | Renal tubular acidosis, proximal, with ocular abnormalities | 99,23 |
| SLC6A1 | 137165 | 616421 | Myoclonic-atonic epilepsy | 99,61 |
| SLC6A17 | 610299 | 616269 | Mental retardation, autosomal recessive 48 | 99,68 |
| SLC6A3 | 126455 | 613135 | Parkinsonism-dystonia, infantile | 99,68 |
| SLC6A8 | 300036 | 300352 | Cerebral creatine deficiency syndrome 1 | 92,97 |
| SLC7A7 | 603593 | 222700 | Lysinuric protein intolerance | 99,67 |
| SLC9A6 | 300231 | 300243 | Mental retardation, X-linked syndromic, Christianson type | 80,47 |
| SMARCA2 | 600014 | 601358 | Nicolaides-Baraitser syndrome | 97,63 |
| SMARCA4 | 603254 | 614609 | Coffin-Siris syndrome 4 | 99,68 |
| SMARCB1 | 601607 | 614608 | Coffin-Siris syndrome 3 | 99,47 |
| SMARCE1 | 603111 | 616938 | Coffin-Siris syndrome 5 | 98,87 |
| SMC1A | 300040 | 300590 | Cornelia de Lange syndrome 2 | 97,41 |
| SMC3 | 606062 | 610759 | Cornelia de Lange syndrome 3 | 98,62 |
| SMOC1 | 608488 | 206920 | Microphthalmia with limb anomalies | 98,99 |
| SMPD1 | 607608 | 257200 | Niemann-Pick disease, type A | 99,68 |
| SMS | 300105 | 309583 | Mental retardation, X-linked, Snyder-Robinson type | 83,70 |
| SNAP29 | 604202 | 609528 | Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome | 98,97 |
| SNIP1 | 608241 | 614501 | Psychomotor retardation, epilepsy, and craniofacial dysmorphism | 99,59 |

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|---|---|---|---|---|
| SNX14 | 616105 | 616354 | Spinocerebellar ataxia, autosomal recessive 20 | 97,83 |
| SOBP | 613667 | 613671 | Mental retardation, anterior maxillary protrusion, and strabismus | 96,13 |
| SON | 182465 | 617140 | ZTTK syndrome | 99,42 |
| SOS1 | 182530 | 610733 | Noonan syndrome 4 | 98,22 |
| SOS2 | 601247 | 616559 | Noonan syndrome 9 | 98,32 |
| SOX10 | 602229 | 611584 | Waardenburg syndrome, type 2E, with or without neurologic involvement | 93,26 |
| SOX11 | 600898 | 615866 | Mental retardation, autosomal dominant, 27 | 84,37 |
| SOX2 | 184429 | 206900 | Optic nerve hypoplasia and abnormalities of the central nervous system | 95,94 |
| SOX5 | 604975 | 616803 | Lamb-Shaffer syndrome | 99,56 |
| SPATA5 | 613940 | 616577 | Epilepsy, hearing loss, and mental retardation syndrome | 98,88 |
| SPECC1L | 614140 | 145410 | Opitz GBBB syndrome, type II | 99,49 |
| SPR | 182125 | 612716 | Dystonia, dopa-responsive, due to sepiapterin reductase deficiency | 77,10 |
| SPRED1 | 609291 | 611431 | Legius syndrome | 99,22 |
| SPTAN1 | 182810 | 613477 | Epileptic encephalopathy, early infantile, 5 | 99,48 |
| SRCAP | 611421 | 136140 | Floating-Harbor syndrome | 99,65 |
| SRD5A3 | 611715 | 612379 | Congenital disorder of glycosylation, type Iq | 96,86 |
| ST3GAL3 | 606494 | 611090 | Mental retardation, autosomal recessive 12 | 99,66 |
| ST3GAL5 | 604402 | 609056 | Salt and pepper developmental regression syndrome | 85,32 |
| STAG1 | 604358 | 617635 | Mental retardation, autosomal dominant 47 | 98,01 |
| STAMBP | 606247 | 614261 | Microcephaly-capillary malformation syndrome | 99,58 |
| STIL | 181590 | 612703 | Microcephaly 7, primary, autosomal recessive | 99,24 |
| STRA6 | 610745 | 601186 | Microphthalmia, syndromic 9 | 99,67 |
| STT3A | 601134 | 615596 | Congenital disorder of glycosylation, type Iw | 99,55 |
| STT3B | 608605 | 615597 | Congenital disorder of glycosylation, type Ix | 95,54 |
| STX1B | 601485 | 616172 | Generalized epilepsy with febrile seizures plus, type 9 | 99,68 |
| STXBP1 | 602926 | 612164 | Epileptic encephalopathy, early infantile, 4 | 99,55 |
| SUCLG1 | 611224 | 245400 | Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria) | 98,06 |
| SUOX | 606887 | 272300 | Sulfite oxidase deficiency | 99,68 |
| SURF1 | 185620 | 256000 | Leigh syndrome, due to COX IV deficiency | 86,96 |
| SYN1 | 313440 | 300491 | Epilepsy, X-linked, with variable learning disabilities and behavior disorders | 88,25 |
| SYNCRIP | 616686 | - | Lelieveld et al., Nat Neurosci 2016 : ID | 99,45 |
| SYNE1 | 608441 | 610743 | Spinocerebellar ataxia, autosomal recessive 8 | 99,44 |
| SYNGAP1 | 603384 | 612621 | Mental retardation, autosomal dominant 5 | 99,68 |
| SYP | 313475 | 300802 | Mental retardation, X-linked 96 | 93,14 |
| SYT14 | 610949 | 614229 | Spinocerebellar ataxia, autosomal recessive 11 | 88,69 |
| TAF1 | 313650 | 300966 | Mental retardation, X-linked, syndromic 33 | 97,05 |
| TAF2 | 604912 | 615599 | Mental retardation, autosomal recessive 40 | 98,57 |
| TBC1D23 | 617687 | 617695 | Pontocerebellar hypoplasia, type 11 | 97,70 |
| TBC1D24 | 613577 | 615338 | Epileptic encephalopathy, early infantile, 16 | 99,68 |
| TBC1D7 | 612655 | 248000 | Macrocephaly/megalencephaly syndrome, autosomal recessive | 98,94 |
| TBCE | 604934 | 241410 | Hypoparathyroidism-retardation-dysmorphism syndrome | 98,41 |
| TBCK | 616899 | 616900 | Hypotonia, infantile, with psychomotor retardation and characteristic facies 3 | 98,05 |
| TBL1XR1 | 608628 | 616944 | Mental retardation, autosomal dominant 41 | 98,00 |
| TCF20 | 603107 | - | Schäfgen et al., EJHG 2016 : TSA, DI, croissance excessive et hypotonie | 99,69 |
| TCF4 | 602272 | 610954 | Pitt-Hopkins syndrome | 99,65 |
| TCOF1 | 606847 | 154500 | Treacher Collins syndrome 1 | 99,61 |
| TCTN1 | 609863 | 614173 | Joubert syndrome 13 | 99,08 |
| TCTN2 | 613846 | 616654 | Joubert syndrome 24 | 99,05 |
| TCTN3 | 613847 | 614815 | Joubert syndrome 18 | 99,61 |
| TECPR2 | 615000 | 615031 | Spastic paraplegia 49, autosomal recessive | 99,41 |
| TECR | 610057 | 614020 | Mental retardation, autosomal recessive 14 | 99,69 |
| TFAP2A | 107580 | 113620 | Branchiooculofacial syndrome | 99,48 |
| TGIF1 | 602630 | 142946 | Holoprosencephaly 4 | 99,69 |
| TH | 191290 | 605407 | Segawa syndrome, recessive | 98,57 |
| THOC2 | 300395 | 300957 | Mental retardation, X-linked 12/35 | 91,08 |
| THOC6 | 615403 | 613680 | Beaulieu-Boycott-Innes syndrome | 99,66 |
| THRA | 190120 | 614450 | Hypothyroidism, congenital, nongoitrous, 6 | 99,60 |
| TIMM8A | 300356 | 304700 | Mohr-Tranebjaerg syndrome | 99,66 |
| TINF2 | 604319 | 613990 | Dyskeratosis congenita, autosomal dominant 3 | 99,67 |
| TMCO1 | 614123 | 213980 | Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome | 95,48 |
| TMEM107 | 616183 | 617563 | Orofaciodigital syndrome XVI | 99,69 |
| TMEM138 | 614459 | 614465 | Joubert syndrome 16 | 99,68 |
| TMEM165 | 614726 | 614727 | Congenital disorder of glycosylation, type IIk | 87,14 |
| TMEM216 | 613277 | 608091 | Joubert syndrome 2 | 99,69 |
| TMEM231 | 614949 | 614970 | Joubert syndrome 20 | 99,62 |
| TMEM237 | 614423 | 614424 | Joubert syndrome 14 | 97,88 |
| TMEM67 | 609884 | 610688 | Joubert syndrome 6 | 97,57 |
| TMEM70 | 612418 | 614052 | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2 | 97,01 |
| TNK2 | 606994 | - | Hitomi et al., Ann Neurol 2013 : severe autosomal recessive infantile-onset epilepsy and ID | 99,30 |
| TPK1 | 606370 | 614458 | Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type) | 98,92 |
| TPP1 | 607998 | 609270 | Spinocerebellar ataxia, autosomal recessive 7 | 99,65 |
| TRAPPC11 | 614138 | 615356 | Muscular dystrophy, limb-girdle, type 2S | 98,70 |
| TRAPPC9 | 611966 | 613192 | Mental retardation, autosomal recessive 13 | 99,57 |
| TREX1 | 606609 | 225750 | Aicardi-Goutieres syndrome 1, dominant and recessive | 99,69 |
| TRIM32 | 602290 | 615988 | Bardet-Biedl syndrome 11 | 99,68 |
| TRIO | 601893 | 617061 | Mental retardation, autosomal dominant 44 | 97,17 |
| TRIP12 | 604506 | - | Zhang et al., Hum Gen 2017 : intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features. | 99,18 |
| TRMT10A | 616013 | 616033 | Microcephaly, short stature, and impaired glucose metabolism 1 | 99,08 |
| TRMU | 610230 | 613070 | Liver failure, transient infantile | 94,99 |
| TRPV4 | 605427 | 184252 | Spondylometaphyseal dysplasia, Kozlowski type | 99,64 |
| TSEN2 | 608753 | 612389 | Pontocerebellar hypoplasia type 2B | 99,60 |
| TSEN54 | 608755 | 277470 | Pontocerebellar hypoplasia type 2A | 87,28 |
| TSFM | 604723 | 610505 | Combined oxidative phosphorylation deficiency 3 | 85,15 |
| TSPAN7 | 300096 | 300210 | Mental retardation, X-linked 58 | 98,69 |
| TTC37 | 614589 | 222470 | Trichohepatoenteric syndrome 1 | 99,17 |
| TTC8 | 608132 | 615985 | Bardet-Biedl syndrome 8 | 98,67 |
| TTI2 | 614426 | 615541 | Mental retardation, autosomal recessive 39 | 99,57 |
| TUBA1A | 602529 | 611603 | Lissencephaly 3 | 99,69 |
| TUBA8 | 605742 | 613180 | Cortical dysplasia, complex, with other brain malformations 8 | 97,24 |
| TUBB2A | 615101 | 615763 | Cortical dysplasia, complex, with other brain malformations 5 | 95,11 |
| TUBB2B | 612850 | 610031 | Cortical dysplasia, complex, with other brain malformations 7 | 99,68 |
| TUBB3 | 602661 | 614039 | Cortical dysplasia, complex, with other brain malformations 1 | 94,69 |
| TUBB4A | 602662 | 612438 | Leukodystrophy, hypomyelinating, 6 | 98,34 |
| TUBG1 | 191135 | 615412 | Cortical dysplasia, complex, with other brain malformations 4 | 99,69 |
| TUBGCP6 | 610053 | 251270 | Microcephaly and chorioretinopathy, autosomal recessive, 1 | 99,69 |
| TUSC3 | 601385 | 611093 | Mental retardation, autosomal recessive 7 | 99,45 |
| TWIST1 | 601622 | 101400 | Saethre-Chotzen syndrome | 59,51 |
| UBE2A | 312180 | 300860 | Mental retardation, X-linked syndromic, Nascimento-type | 81,32 |
| UBE3A | 601623 | 105830 | Angelman syndrome | 98,84 |
| UBE3B | 608047 | 244450 | Kaufman oculocerebrofacial syndrome | 99,43 |
| UBR1 | 605981 | 243800 | Johanson-Blizzard syndrome | 98,99 |

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| UNC80 | 612636 | 616801 | Hypotonia, infantile, with psychomotor retardation and characteristic facies 2 | 99,46 |
| UPF3B | 300298 | 300676 | Mental retardation, X-linked, syndromic 14 | 90,82 |
| UQCRQ | 612080 | 615159 | Mitochondrial complex III deficiency, nuclear type 4 | 99,62 |
| UROC1 | 613012 | 276880 | Urocanase deficiency | 99,69 |
| USP18 | 607057 | 617397 | Pseudo-TORCH syndrome 2 | 99,69 |
| USP9X | 300072 | 300919 | Mental retardation, X-linked 99 | 93,59 |
| VLDLR | 192977 | 224050 | Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1 | 97,10 |
| VPS13B | 607817 | 216550 | Cohen syndrome | 98,95 |
| VRK1 | 602168 | 607596 | Pontocerebellar hypoplasia type 1A | 99,08 |
| WAC | 615049 | 616708 | Desanto-Shinawi syndrome | 97,24 |
| WDPCP | 613580 | 615992 | Bardet-Biedl syndrome 15 | 98,77 |
| WDR26 | 617424 | 617616 | Skraban-Deardorff syndrome | 99,46 |
| WDR45 | 300526 | 300894 | Neurodegeneration with brain iron accumulation 5 | 99,94 |
| WDR45B | 609226 | - | Suleiman et al., Clin Genet 2017 : intellectual disability, spastic quadriplegia, epilepsy, and cerebral hypoplasia | 98,84 |
| WDR62 | 613583 | 604317 | Microcephaly 2, primary, autosomal recessive, with or without cortical malformations | 99,58 |
| WDR73 | 616144 | 251300 | Galloway-Mowat syndrome | 99,54 |
| WDR81 | 614218 | 610185 | Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2 | 99,65 |
| WWOX | 605131 | 616211 | Epileptic encephalopathy, early infantile, 28 | 99,63 |
| XPA | 611153 | 278700 | Xeroderma pigmentosum, group A | 95,49 |
| XPC | 613208 | 278720 | Xeroderma pigmentosum, group C | 99,66 |
| XYLT1 | 608124 | 615777 | Desbuquois dysplasia 2 | 91,40 |
| YAP1 | 606608 | 120433 | Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation | 95,59 |
| YY1 | 600013 | 617557 | Gabriele-de Vries syndrome | 97,61 |
| ZBTB16 | 176797 | 612447 | Skeletal defects, genital hypoplasia, and mental retardation | 99,69 |
| ZBTB18 | 608433 | 612337 | Mental retardation, autosomal dominant 22 | 99,10 |
| ZBTB20 | 606025 | 259050 | Primrose syndrome | 99,63 |
| ZBTB24 | 614064 | 614069 | Immunodeficiency-centromeric instability-facial anomalies syndrome-2 | 99,63 |
| ZC4H2 | 300897 | 314580 | Wieacker-Wolff syndrome | 97,81 |
| ZDHHC9 | 300646 | 300799 | Mental retardation, X-linked syndromic, Raymond type | 98,90 |
| ZEB2 | 605802 | 235730 | Mowat-Wilson syndrome | 99,54 |
| ZFYVE26 | 612012 | 270700 | Spastic paraplegia 15, autosomal recessive | 99,53 |
| ZIC2 | 603073 | 609637 | Holoprosencephaly 5 | 67,20 |
| ZMPSTE24 | 606480 | 608612 | Mandibuloacral dysplasia with type B lipodystrophy | 98,68 |
| ZMYND11 | 608668 | 616083 | Mental retardation, autosomal dominant 30 | 99,50 |
| ZNF335 | 610827 | 615095 | Microcephaly 10, primary, autosomal recessive | 99,63 |
| ZNF41 | 314995 | - | Shoichet et al., AJHG 2013 : X-Linked Mental Retardation | 99,90 |
| ZNF711 | 314990 | 300803 | Mental retardation, X-linked 97 | 89,61 |

La nomenclature des gènes suit les recommandations HGCN : Genomics 79(4):464-470 (2002) [updated October 2013].
Les codes OMIM (gene, disease, phenotype) affichés ont été repris à partir de la version datant du 15 Novembre 2017.
La couverture moyenne avec une profondeur de lecture de 30x [Length Covered Above 30x] est calculée sur base de 256 exomes.

Gene symbols used follow HGCN guidelines : Genomics 79(4):464-470 (2002) [updated October 2013].
OMIM release used for OMIM disease identifiers and descriptions : 15th November 2017.
[Length Covered Above 30x] describes the average coverage of a gene across 256 exomes in percentile.