## Supplementary data:

# Coverage analysis of lists of genes involved in heterogeneous genetic diseases following benchtop exome sequencing using the ion proton

Caroline Lacoste, Jean-Pierre Desvignes, David Salgado, Christophe Pecheux, Laurent Villard, Marc Bartoli, Christophe Beroud, Nicolas Levy, Catherine Badens and Martin Krahn

*J. Genet.* **95**, 203–208

**Table 1.** Gene lists of interest for sequence coverage analysis and corresponding diseases as referenced in OMIM (Amberger *et al.* 2009).

**Genes involved in myopathies:** 82 genes, based on the disease groups 'muscular dystrophies', 'congenital muscular dystrophies', 'congenital myopathies', 'distal myopathies' and 'other myopathies' according to the Gene Table of Neuromuscular Disorders; www.musclegenetable.fr (Kaplan and Hamroun 2013).

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *ACTA1* | 102610 | Myopathy, actin, congenital, with cores | 161800 |
| | | Myopathy, actin, congenital, with excess of thin myofilaments | 161800 |
| | | Myopathy, congenital, with fiber-type disproportion 1 | 255310 |
| | | Nemaline myopathy 3, autosomal dominant or recessive | 161800 |
| *ACVR1* | 102576 | Fibrodysplasia ossificans progressiva | 135100 |
| *ALG13* | 300776 | Congenital disorder of glycosylation, type Is | 300884 |
| *ANO5* | 608662 | Gnathodiaphyseal dysplasia | 166260 |
| | | Miyoshi muscular dystrophy 3 | 613319 |
| | | Muscular dystrophy, limb-girdle, type 2L | 611307 |
| *B3GALNT2* | 610194 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11 | 615181 |
| *B3GNT1* | 605517 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13 | 615287 |
| *BAG3* | 603883 | Cardiomyopathy, dilated, 1HH | 613881 |
| | | Myopathy, myofibrillar, 6 | 612954 |
| *BIN1* | 601248 | Myopathy, centronuclear, autosomal recessive | 255200 |
| *CAPN3* | | Muscular dystrophy, limb-girdle, type 2A | 253600 |
| *CAV3* | 601253 | Cardiomyopathy, familial hypertrophic | 192600 |
| | | Creatine phosphokinase, elevated serum | 123320 |
| | | Long QT syndrome 9 | 611818 |
| | | Muscular dystrophy, limb-girdle, type IC | 607801 |
| | | Myopathy, distal, Tateyama type | 614321 |
| | | Rippling muscle disease | 606072 |
| *CFL2* | 601443 | Nemaline myopathy 7, autosomal recessive | 610687 |
| *CHKB* | 612395 | Muscular dystrophy, congenital, megaconial type | 602541 |
| *CNTN1* | 600016 | Myopathy, congenital, Compton-North | 612540 |
| *COL6A1* | 120220 | Bethlem myopathy | 158810 |
| | | Ullrich congenital muscular dystrophy | 254090 |
| | | {Ossification of the posterior longitudinal spinal ligaments} | 602475 |
| *COL6A2* | 120240 | Bethlem myopathy | 158810 |
| | | Myosclerosis, congenital | 255600 |
| | | Ullrich congenital muscular dystrophy | 254090 |
| *COL6A3* | 120250 | Bethlem myopathy | 158810 |
| | | Ullrich congenital muscular dystrophy | 254090 |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *CRYAB* | 123590 | Cardiomyopathy, dilated, 1II | 615184 |
| | | Cataract 16, multiple types | 613763 |
| | | Myopathy, myofibrillar, 2 | 608810 |
| | | Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related | 613869 |
| *DAG1* | 128239 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9 | 613818 |
| *DES* | 125660 | ?Muscular dystrophy, limb-girdle, type 2R | 615325 |
| | | Cardiomyopathy, dilated, 1I | 604765 |
| | | Myopathy, myofibrillar, 1 | 601419 |
| | | Scapuloperoneal syndrome, neurogenic, Kaeser type | 181400 |
| *DMD* | 300377 | Becker muscular dystrophy | 300376 |
| | | Cardiomyopathy, dilated, 3B | 302045 |
| | | Duchenne muscular dystrophy | 310200 |
| *DNAJB6* | 611332 | Muscular dystrophy, limb-girdle, type 1E | 603511 |
| *DNM2* | 602378 | Charcot-Marie-Tooth disease, axonal, type 2M | 606482 |
| | | Charcot-Marie-Tooth disease, dominant intermediate B | 606482 |
| | | Lethal congenital contracture syndrome 5 | 615368 |
| | | Myopathy, centronuclear | 160150 |
| *DPM1* | 603503 | Congenital disorder of glycosylation, type Ie | 608799 |
| *DPM2* | 603564 | Congenital disorder of glycosylation, type Iu | 615042 |
| *DPM3* | 605951 | Congenital disorder of glycosylation, type Io | 612937 |
| *DYSF* | 603009 | Miyoshi muscular dystrophy 1 | 254130 |
| | | Muscular dystrophy, limb-girdle, type 2B | 253601 |
| | | Myopathy, distal, with anterior tibial onset | 606768 |
| *EMD* | 300384 | Emery-Dreifuss muscular dystrophy 1, X-linked | 310300 |
| *FHL1* | 300163 | Emery-Dreifuss muscular dystrophy 6, X-linked | 300696 |
| | | Myopathy, reducing body, X-linked, childhood-onset | 300718 |
| | | Myopathy, reducing body, X-linked, severe early-onset | 300717 |
| | | Myopathy, X-linked, with postural muscle atrophy | 300696 |
| | | Scapuloperoneal myopathy, X-linked dominant | 300695 |
| *FKRP* | 606596 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5 | 613153 |
| | | Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5 | 606612 |
| | | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5 | 607155 |
| *FKTN* | 607440 | Cardiomyopathy, dilated, 1X | 611615 |
| | | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4 | 253800 |
| | | Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4 | 613152 |
| | | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4 | 611588 |
| *FLNC* | 102565 | Myopathy, distal, 4 | 614065 |
| | | Myopathy, myofibrillar, 5 | 609524 |
| *GMPPB* | 615320 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14 | 615350 |
| | | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14 | 615351 |
| | | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14 | 615352 |
| *GNE* | 603824 | Inclusion body myopathy, autosomal recessive | 600737 |
| | | Nonaka myopathy | 605820 |
| | | Sialuria | 269921 |
| *ISCU* | 611911 | Myopathy with lactic acidosis, hereditary | 255125 |
| *ITGA7* | 600536 | Muscular dystrophy, congenital, due to ITGA7 deficiency | 613204 |
| *KBTBD13* | 613727 | Nemaline myopathy 6, autosomal dominant | 609273 |
| *KLHL40* | 615340 | Nemaline myopathy 8, autosomal recessive | 615348 |
| *KLHL9* | 611201 | / | / |
| *LAMA2* | 156225 | Muscular dystrophy, congenital merosin-deficient | 607855 |
| | | Muscular dystrophy, congenital, due to partial LAMA2 deficiency | 607855 |
| *LAMP2* | 309060 | Danon disease | 300257 |
| *LARGE* | 603590 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6 | 613154 |
| | | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6 | 608840 |
| *LDB3* | 605906 | Cardiomyopathy, dilated 1C | 601493 |
| | | Left ventricular noncompaction 3, with or without dilated cardiomyopathy | 601493 |
| | | Myopathy, myofibrillar, 4 | 609452 |
| *LMNA* | 150330 | Cardiomyopathy, dilated, 1A | 115200 |
| | | Charcot-Marie-Tooth disease, type 2B1 | 605588 |
| | | Emery-Dreifuss muscular dystrophy 2, AD | 181350 |
| | | Emery-Dreifuss muscular dystrophy 3, AR | 181350 |
| | | Heart-hand syndrome, Slovenian type | 610140 |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| | | Hutchinson-Gilford progeria | 176670 |
| | | Lipodystrophy, familial partial, 2 | 151660 |
| | | Malouf syndrome | 212112 |
| | | Mandibuloacral dysplasia | 248370 |
| | | Muscular dystrophy, congenital | 613205 |
| | | Muscular dystrophy, limb-girdle, type 1B | 159001 |
| | | Restrictive dermopathy, lethal | 275210 |
| *MATR3* | 164015 | Amyotrophic lateral sclerosis 21 | 606070 |
| *MEGF10* | 612453 | Myopathy, areflexia, respiratory distress, and dysphagia, early-onset | 614399 |
| | | Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant | 614399 |
| *MSTN* | 601788 | Muscle hypertrophy | 614160 |
| *MTM1* | 300415 | Myotubular myopathy, X-linked | 310400 |
| *MYBPC3* | 600958 | Cardiomyopathy, dilated, 1MM | 615396 |
| | | Cardiomyopathy, familial hypertrophic, 4 | 115197 |
| | | Left ventricular noncompaction 10 | 615396 |
| *MYH2* | 160740 | Inclusion body myopathy-3 | 605637 |
| *MYH7* | 160760 | Cardiomyopathy, dilated, 1S | 613426 |
| | | Cardiomyopathy, familial hypertrophic, 1 | 192600 |
| | | Laing distal myopathy | 160500 |
| | | Left ventricular noncompaction 5 | 613426 |
| | | Myopathy, myosin storage | 608358 |
| | | Scapuloperoneal syndrome, myopathic type | 181430 |
| *MYOT* | 604103 | Muscular dystrophy, limb-girdle, type 1A | 159000 |
| | | Myopathy, myofibrillar, 3 | 609200 |
| | | Myopathy, spheroid body | 182920 |
| *NEB* | 161650 | Nemaline myopathy 2, autosomal recessive | 256030 |
| *PABPN1* | 602279 | Oculopharyngeal muscular dystrophy | 164300 |
| *PLEC1* | 601282 | Epidermolysis bullosa simplex with pyloric atresia | 612138 |
| | | Epidermolysis bullosa simplex, Ogna type | 131950 |
| | | Muscular dystrophy with epidermolysis bullosa simplex | 226670 |
| | | Muscular dystrophy, limb-girdle, type 2Q | 613723 |
| *POMGNT1* | 606822 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3 | 253280 |
| | | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3 | 613151 |
| | | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3 | 613157 |
| *POMGNT2* | 614828 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8 | 614830 |
| *POMK* | 615247 | ?Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12 | 615249 |
| *POMT1* | 607423 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1 | 236670 |
| | | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1 | 613155 |
| | | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1 | 609308 |
| *POMT2* | 607439 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2 | 613150 |
| | | Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2 | 613156 |
| | | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2 | 613158 |
| *PTPLA* | 610467 | / | / |
| *PTRF* | 603198 | Lipodystrophy, congenital generalized, type 4 | 613327 |
| *RYR1* | 180901 | Central core disease | 117000 |
| | | King-Denborough syndrome | 145600 |
| | | Minicore myopathy with external ophthalmoplegia | 255320 |
| | | Neuromuscular disease, congenital, with uniform type 1 fiber | 117000 |
| | | {Malignant hyperthermia susceptibility 1} | 145600 |
| *SEPN1* | 606210 | Muscular dystrophy, rigid spine, 1 | 602771 |
| | | Myopathy, congenital, with fiber-type disproportion | 255310 |
| *SGCA* | 600119 | Muscular dystrophy, limb-girdle, type 2D | 608099 |
| *SGCB* | 600900 | Muscular dystrophy, limb-girdle, type 2E | 604286 |
| *SGCD* | 601411 | Cardiomyopathy, dilated, 1L | 606685 |
| | | Muscular dystrophy, limb-girdle, type 2F | 601287 |
| *SGCG* | 608896 | Muscular dystrophy, limb-girdle, type 2C | 253700 |
| *SMCHD1* | 614982 | Fascioscapulohumeral muscular dystrophy 2, digenic | 158901 |
| *SYNE1* | 608441 | Emery-Dreifuss muscular dystrophy 4, autosomal dominant | 612998 |
| | | Spinocerebellar ataxia, autosomal recessive 8 | 610743 |
| *SYNE2* | 608442 | Emery-Dreifuss muscular dystrophy 5, autosomal dominant | 612999 |
| *TCAP* | 604488 | Cardiomyopathy, dilated, 1N | 607487 |
| | | Muscular dystrophy, limb-girdle, type 2G | 601954 |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *TIA1* | 603518 | Welander distal myopathy | 604454 |
| *TMEM43* | 612048 | Arrhythmogenic right ventricular dysplasia 5 | 604400 |
| | | Emery-Dreifuss muscular dystrophy 7, AD | 614302 |
| *TMEM5* | 605862 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10 | 615041 |
| *TNNT1* | 191041 | Nemaline myopathy 5, Amish type | 605355 |
| *TNPO3* | 610032 | / | / |
| *TPM2* | 190990 | Arthrogryposis multiplex congenita, distal, type 1 | 108120 |
| | | Arthrogryposis, distal, type 2B | 601680 |
| | | CAP myopathy 2 | 609285 |
| | | Nemaline myopathy 4, autosomal dominant | 609285 |
| *TPM3* | 191030 | CAP myopathy 1 | 609284 |
| | | Myopathy congenital, with fiber-type disproportion | 255310 |
| | | Nemaline myopathy 1, autosomal dominant or recessive | 609284 |
| *TRAPPC11* | 614138 | Muscular dystrophy, limb-girdle, type 2S | 615356 |
| *TRIM32* | 602290 | Bardet-Biedl syndrome 11 | 209900 |
| | | Muscular dystrophy, limb-girdle, type 2H | 254110 |
| *TTN* | 188840 | Cardiomyopathy, dilated, 1G | 604145 |
| | | Cardiomyopathy, familial hypertrophic, 9 | 613765 |
| | | Muscular dystrophy, limb-girdle, type 2J | 608807 |
| | | Myopathy, early-onset, with fatal cardiomyopathy | 611705 |
| | | Myopathy, proximal, with early respiratory muscle involvement | 603689 |
| | | Tibial muscular dystrophy, tardive | 600334 |
| *VCP* | 601023 | Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia | 613954 |
| | | Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1 | 167320 |

**Genes involved in hereditary motor and sensory neuropathies:** 55 genes, based on the disease group 'hereditary motor and sensory neuropathies' from the Gene Table of Neuromuscular Disorders; www.musclegenetable.fr (Kaplan and Hamroun 2013).

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *AARS* | 601065 | Charcot-Marie-Tooth disease, axonal, type 2N | 613287 |
| *AIFM1* | 300169 | Combined oxidative phosphorylation deficiency 6 | 300816 |
| | | Cowchock syndrome | 310490 |
| *ARHGEF10* | 608136 | Slowed nerve conduction velocity, AD | 608236 |
| *ATL1* | 606439 | Neuropathy, hereditary sensory, type ID | 613708 |
| | | Spastic paraplegia 3A, autosomal dominant | 182600 |
| *CTDP1* | 604927 | Congenital cataracts, facial dysmorphism, and neuropathy | 604168 |
| *DNM2* | 302378 | Charcot-Marie-Tooth disease, axonal, type 2M | 606482 |
| | | Charcot-Marie-Tooth disease, dominant intermediate B | 606482 |
| | | Lethal congenital contracture syndrome 5 | 615368 |
| | | Myopathy, centronuclear | 160150 |
| *DNMT1* | 126375 | Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant | 604121 |
| | | Neuropathy, hereditary sensory, type IE | 614116 |
| *DYNC1H1* | 600112 | Charcot-Marie-Tooth disease, axonal, type 20 | 614228 |
| | | Mental retardation, autosomal dominant 13 | 614563 |
| | | Spinal muscular atrophy, lower extremity-predominant, AD | 158600 |
| *EGR2* | 129010 | Charcot-Marie-Tooth disease, type 1D | 607678 |
| | | Dejerine-Sottas disease | 145900 |
| | | Neuropathy, congenital hypomyelinating, 1 | 605253 |
| *FBLN5* | 604580 | Cutis laxa, autosomal dominant 2 | 614434 |
| | | Cutis laxa, autosomal recessive, type IA | 219100 |
| | | Macular degeneration, age-related, 3 | 608895 |
| *FGD4* | 611104 | Charcot-Marie-Tooth disease, type 4H | 609311 |
| *FIG4* | 609390 | Amyotrophic lateral sclerosis 11 | 612577 |
| | | Charcot-Marie-Tooth disease, type 4J | 611228 |
| | | Yunis-Varon syndrome | 216340 |
| *GAN* | 605379 | Giant axonal neuropathy-1 | 256850 |
| *GARS* | 600287 | Charcot-Marie-Tooth disease, type 2D | 601472 |
| | | Neuropathy, distal hereditary motor, type VA | 600794 |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *GDAP1* | 606598 | Charcot-Marie-Tooth disease, axonal, type 2K | 607831 |
| | | Charcot-Marie-Tooth disease, axonal, with vocal cord paresis | 607706 |
| | | Charcot-Marie-Tooth disease, recessive intermediate, A | 608340 |
| | | Charcot-Marie-Tooth disease, type 4A | 214400 |
| *GJB1* | 304040 | Charcot-Marie-Tooth neuropathy, X-linked dominant, 1 | 302800 |
| *GNB4* | 610863 | Charcot-Marie-Tooth disease, dominant intermediate F | 615185 |
| *HINT1* | 601314 | Neuromyotonia and axonal neuropathy, autosomal recessive | 137200 |
| *HK1* | 142600 | Hemolytic anemia due to hexokinase deficiency | 235700 |
| | | Neuropathy, hereditary motor and sensory, Russe type | 605285 |
| *HOXD10* | 142984 | Charcot-Marie-Tooth disease, foot deformity of | 192950 |
| | | Vertical talus, congenital | 192950 |
| *HSPB1* | 602195 | Charcot-Marie-Tooth disease, axonal, type 2F | 606595 |
| | | Neuropathy, distal hereditary motor, type IIB | 608634 |
| *HSPB8* | 608014 | Charcot-Marie-Tooth disease, axonal, type 2L | 608673 |
| | | Neuropathy, distal hereditary motor, type IIA | 158590 |
| *IKBKAP* | 603722 | Dysautonomia, familial | 223900 |
| *INF2* | 610982 | Charcot-Marie-Tooth disease, dominant intermediate E | 614455 |
| | | Glomerulosclerosis, focal segmental, 5 | 613237 |
| *KARS* | 601421 | Charcot-Marie-Tooth disease, recessive intermediate, B | 613641 |
| | | Deafness, autosomal recessive 89 | 613916 |
| *KIF1A* | 601255 | Mental retardation, autosomal dominant 9 | 614255 |
| | | Neuropathy, hereditary sensory, type IIC | 614213 |
| | | Spastic paraplegia 30, autosomal recessive | 610357 |
| *KIF1B* | 605995 | Charcot-Marie-Tooth disease, type 2A1 | 118210 |
| | | Pheochromocytoma | 171300 |
| | | {Neuroblastoma, susceptibility to, 1} | 256700 |
| *LITAF* | 603795 | Charcot-Marie-Tooth disease, type 1C | 601098 |
| *LMNA* | 150330 | Cardiomyopathy, dilated, 1A | 115200 |
| | | Charcot-Marie-Tooth disease, type 2B1 | 605588 |
| | | Emery-Dreifuss muscular dystrophy 2, AD | 181350 |
| | | Emery-Dreifuss muscular dystrophy 3, AR | 181350 |
| | | Heart-hand syndrome, Slovenian type | 610140 |
| | | Hutchinson-Gilford progeria | 176670 |
| | | Lipodystrophy, familial partial, 2 | 151660 |
| | | Malouf syndrome | 212112 |
| | | Mandibuloacral dysplasia | 248370 |
| | | Muscular dystrophy, congenital | 613205 |
| | | Muscular dystrophy, limb-girdle, type 1B | 159001 |
| | | Restrictive dermopathy, lethal | 275210 |
| *LRSAM1* | 610933 | Charcot-Marie-Toothe disease, axonal, type 2P | 614436 |
| *MARS* | 156560 | ?Infantile liver failure syndrome 2 | 615486 |
| *MED25* | 610197 | Charcot-Marie-Tooth disease, type 2B2 | 605589 |
| *MFN2* | 608507 | Charcot-Marie-Tooth disease, type 2A2 | 609260 |
| | | Hereditary motor and sensory neuropathy VI | 601152 |
| *MPZ* | 159440 | Charcot-Marie-Tooth disease, dominant intermediate D | 607791 |
| | | Charcot-Marie-Tooth disease, type 1B | 118200 |
| | | Charcot-Marie-Tooth disease, type 2I | 607677 |
| | | Charcot-Marie-Tooth disease, type 2J | 607736 |
| | | Dejerine-Sottas disease | 145900 |
| | | Neuropathy, congenital hypomyelinating | 605253 |
| | | Roussy-Levy syndrome | 180800 |
| *MTMR2* | 603557 | Charcot-Marie-Tooth disease, type 4B1 | 601382 |
| *NDRG1* | 605262 | Charcot-Marie-Tooth disease, type 4D | 601455 |
| *NEFL* | 162280 | Charcot-Marie-Tooth disease, type 1F | 607734 |
| *PDK3* | 300906 | ?Charcot-Marie-Tooth disease, X-linked dominant, 6 | 300905 |
| *PMP22* | 601097 | Charcot-Marie-Tooth disease, type 1A | 118220 |
| | | Charcot-Marie-Tooth disease, type 1E | 118300 |
| | | Dejerine-Sottas disease | 145900 |
| | | Neuropathy, inflammatory demyelinating | 139393 |
| | | Neuropathy, recurrent, with pressure palsies | 162500 |
| | | Roussy-Levy syndrome | 180800 |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *PRPS1* | 311850 | Arts syndrome | 301835 |
| | | Charcot-Marie-Tooth disease, X-linked recessive, 5 | 311070 |
| | | Deafness, X-linked 1 | 304500 |
| | | Gout, PRPS-related | 300661 |
| | | Phosphoribosylpyrophosphate synthetase superactivity | 300661 |
| *PRX* | 605725 | Charcot-Marie-Tooth disease, type 4F | 614895 |
| | | Dejerine-Sottas disease, autosomal recessive | 145900 |
| *RAB7* | 602298 | Charcot-Marie-Tooth disease, type 2B | 600882 |
| *SBF1* | 603560 | ?Charcot-Marie-Tooth disease, type 4B3 | 615284 |
| *SBF2* | 607697 | Charcot-Marie-Tooth disease, type 4B2 | 604563 |
| *SEPT9* | 604061 | Amyotrophy, hereditary neuralgic | 162100 |
| | | Leukemia, acute myeloid, therapy-related | / |
| | | Ovarian carcinoma | / |
| *SH3TC2* | 608206 | Charcot-Marie-Tooth disease, type 4C | 601596 |
| | | Mononeuropathy of the median nerve, mild | 613353 |
| *SLC12A6* | 604878 | Agenesis of the corpus callosum with peripheral neuropathy | 218000 |
| *SPTLC1* | 605712 | Neuropathy, hereditary sensory and autonomic, type IA | 162400 |
| *SPTLC2* | 605713 | Neuropathy, hereditary sensory and autonomic, type IC | 613640 |
| *TFG* | 602498 | ?Spastic paraplegia 57, autosomal recessive | 615658 |
| | | Chondrosarcoma, extraskeletal myxoid | 612237 |
| | | Hereditary motor and sensory neuropathy, proximal type | 604484 |
| *TRPV4* | 605427 | Brachyolmia type 3 | 113500 |
| | | Digital arthropathy-brachydactyly, familial | 606835 |
| | | Hereditary motor and sensory neuropathy, type IIc | 606071 |
| | | Metatropic dysplasia | 156530 |
| | | Parastremmatic dwarfism | 168400 |
| | | Scapuloperoneal spinal muscular atrophy | 181405 |
| | | SED, Maroteaux type | 184095 |
| | | Spinal muscular atrophy, distal, congenital nonprogressive | 600175 |
| | | Spondylometaphyseal dysplasia, Kozlowski type | 184252 |
| | | [Sodium serum level QTL 1] | 613508 |
| *VRK1* | 602168 | Pontocerebellar hypoplasia type 1A | 607596 |
| *WNK1* | 605232 | Neuropathy, hereditary sensory and autonomic, type II | 201300 |
| | | Pseudohypoaldosteronism, type IIC | 614492 |
| *YARS* | | Charcot-Marie-Tooth disease, dominant intermediate C | 608323 |

**Genes involved in early onset epileptic encephalopathies:** 30 genes based on (Kodera *et al.* 2013).

| HUGO | OMIM number | Phenotype | MIM Phenotype number |
|---|---|---|---|
| *ARHGEF9* | 300429 | Epileptic encephalopathy, early infantile, 8 | 300607 |
| *ARX* | 300382 | Epileptic encephalopathy, early infantile, 1 | 308350 |
| | | Hydranencephaly with abnormal genitalia | 300215 |
| | | Lissencephaly, X-linked 2 | 300215 |
| | | Mental retardation, X-linked 29 and others | 300419 |
| | | Partington syndrome | 309510 |
| | | Proud syndrome | 300004 |
| *CASK* | 300172 | FG syndrome 4 | 300422 |
| | | Mental retardation and microcephaly with pontine and cerebellar hypoplasia | 300749 |
| | | Mental retardation, with or without nystagmus | 300422 |
| *CDKL5* | 300203 | Angelman syndrome-like | 105830 |
| | | Epileptic encephalopathy, early infantile, 2 | 300672 |
| *COL4A1* | 120130 | Angiopathy, hereditary, with nephropathy, aneurysms, and muscle | 611773 |
| | | Brain small vessel disease with Axenfeld-Rieger anomaly | 607595 |
| | | Brain small vessel disease with hemorrhage | 607595 |
| | | Porencephaly 1 | 175780 |
| | | {Hemorrhage, intracerebral, susceptibility to} | *614519* |
| *COL4A2* | 120090 | Porencephaly 2 | 614483 |
| | | {Hemorrhage, intracerebral, susceptibility to} | *614519* |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | MIM Phenotype number |
|---|---|---|---|
| *FOXG1* | 164874 | Rett syndrome, congenital variant | 613454 |
| *GABRG2* | 137164 | Epilepsy, generalized, with febrile seizures plus, type 3 | 611277 |
| | | Febrile seizures, familial, 8 | 611277 |
| | | {Epilepsy, childhood absence, susceptibility to, 2} | 607681 |
| *GRIN2A* | 138253 | Epilepsy, focal, with speech disorder and with or without mental retardation | 245570 |
| *KCNQ2* | 602235 | Epileptic encephalopathy, early infantile, 7 | 613720 |
| | | Myokymia | 121200 |
| | | Seizures, benign neonatal, 1 | 121200 |
| *MAGI2* | 606382 | / | / |
| *MAPK10* | 602897 | / | / |
| *MECP2* | 300005 | Angelman syndrome | 105830 |
| | | Encephalopathy, neonatal severe | 300673 |
| | | Mental retardation, X-linked syndromic, Lubs type | 300260 |
| | | Mental retardation, X-linked, syndromic 13 | 300055 |
| | | Rett syndrome | 312750 |
| | | Rett syndrome, preserved speech variant | 312750 |
| | | {Autism susceptibility, X-linked 3} | 300496 |
| *MEF2C* | 600662 | Chromosome 5q14.3 deletion syndrome | 613443 |
| | | Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations | 613443 |
| *NTNG1* | 608818 | / | / |
| *PCDH19* | 300460 | Epileptic encephalopathy, early infantile, 9 | 300088 |
| *PLCB1* | 607120 | Epileptic encephalopathy, early infantile, 12 | 613722 |
| *PNKP* | 605610 | Epileptic encephalopathy, early infantile, 10 | 613402 |
| *PNPO* | 603287 | Pyridoxamine 5'-phosphate oxidase deficiency | 610090 |
| *SCN1A* | 182389 | Dravet syndrome | 607208 |
| | | Epilepsy, generalized, with febrile seizures plus, type 2 | 604403 |
| | | Febrile seizures, familial, 3A | 604403 |
| | | Migraine, familial hemiplegic, 3 | 609634 |
| *SCN2A* | 182390 | Epileptic encephalopathy, early infantile, 11 | 613721 |
| | | Seizures, benign familial infantile, 3 | 607745 |
| *SLC25A22* | 609302 | Epileptic encephalopathy, early infantile, 3 | 609304 |
| *SLC2A1* | 138140 | Dystonia 9 | 601042 |
| | | GLUT1 deficiency syndrome 1 | 606777 |
| | | GLUT1 deficiency syndrome 2 | 612126 |
| | | {Epilepsy, idiopathic generalized, suscpetibility to, 12} | 614847 |
| *SNPH* | 604942 | / | / |
| *SPTAN1* | 182810 | Epileptic encephalopathy, early infantile, 5 | 613477 |
| *SRGAP2* | 606524 | / | / |
| *ST3GAL5* | 604402 | Amish infantile epilepsy syndrome | 609056 |
| *STXBP1* | 602926 | Epileptic encephalopathy, early infantile, 4 | 612164 |
| *SYN1* | 313440 | Epilepsy, X-linked, with variable learning disabilities and behavior disorders | 300491 |
| SYP | 313475 | Mental retardation, X-linked 96 | 300802 |

**Genes involved in isolated and combined dystonia:** 12 genes based on (Klein and Münchau 2003).

| HUGO | OMIM number | Phenotype | MIM phenotype number |
|---|---|---|---|
| *ATP1A3* | 182350 | Alternating hemiplegia of childhood 2 | 614820 |
| | | Dystonia-12 | 128235 |
| *GCH1* | 600225 | Dystonia, DOPA-responsive, with or without hyperphenylalaninemia | 128230 |
| | | Hyperphenylalaninemia, BH4-deficient, B | 233910 |
| *GNAL* | 139312 | Dystonia 25 | 615073 |
| *PNKD* | 609023 | Paroxysmal nonkinesigenic dyskinesia | 118800 |
| *PRRT2* | 614386 | Convulsions, familial infantile, with paroxysmal choreoathetosis | 602066 |
| | | Episodic kinesigenic dyskinesia 1 | 128200 |
| | | Seizures, benign familial infantile, 2 | 605751 |
| *SGCE* | 604149 | Dystonia-11, myoclonic | 159900 |
| *SLC2A1* | 138140 | Dystonia 9 | 601042 |
| | | GLUT1 deficiency syndrome 1 | 606777 |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | MIM phenotype number |
|---|---|---|---|
| | | GLUT1 deficiency syndrome 2 | 612126 |
| | | {Epilepsy, idiopathic generalized, suscpetibility to, 12} | 614847 |
| *SPR* | 182125 | Dystonia, dopa-responsive, due to sepiapterin reductase deficiency | 612716 |
| *TAF1* | 313650 | Dystonia-Parkinsonism, X-linked | 314250 |
| *TH* | 191290 | Segawa syndrome, recessive | 605407 |
| *THAP1* | 609520 | Dystonia 6, torsion | 602629 |
| *TOR1A* | 605204 | Dystonia, early-onset atypical, with myoclonic features | / |
| | | Dystonia-1, torsion | 128100 |
| | | {Dystonia-1, modifier of} | / |

**Genes involved in non syndromic deafness and hereditary hearing loss:** 60 genes based on (Smith *et al.* 1999).

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *ACTG1* | 102560 | Baraitser-Winter syndrome 2 | 614583 |
| | | Deafness, autosomal dominant 20/26 | 604717 |
| *BSND* | | Bartter syndrome, type 4a | 602522 |
| | | Sensorineural deafness with mild renal dysfunction | 602522 |
| *CCDC50* | 611051 | Deafness, autosomal dominant 44 | 607453 |
| *CDH23* | 605516 | Deafness, autosomal recessive 12 | 601386 |
| | | Usher syndrome, type 1D | 601067 |
| | | Usher syndrome, type 1D/F digenic | 601067 |
| *CLDN14* | 605608 | Deafness, autosomal recessive 29 | 614035 |
| *COCH* | 603196 | Deafness, autosomal dominant 9 | 601369 |
| *COL11A2* | 120290 | Deafness, autosomal dominant 13 | 601868 |
| | | Deafness, autosomal recessive 53 | 609706 |
| | | Fibrochondrogenesis 2 | 614524 |
| | | Otospondylomegaepiphyseal dysplasia | 215150 |
| | | Stickler syndrome, type III | 184840 |
| | | Weissenbacher-Zweymuller syndrome | 277610 |
| *DFNA5* | 608798 | Deafness, autosomal dominant 5 | 600994 |
| *DFNB31* | 607928 | Deafness, autosomal recessive 31 | 607084 |
| | | Usher syndrome, type 2D | 611383 |
| *DFNB59* | 610219 | Deafness, autosomal recessive 59 | 610220 |
| *DIAPH1* | 302121 | Deafness, autosomal dominant 1 | 124900 |
| *DSPP* | 125485 | Deafness, autosomal dominant 36, with dentinogenesis | 605594 |
| | | Dentin dysplasia, type II | 125420 |
| | | Dentinogenesis imperfecta, Shields type II | 125490 |
| | | Dentinogenesis imperfecta, Shields type III | 125500 |
| *ESPN* | 606351 | Deafness, autosomal recessive 36 | 609006 |
| | | Deafness, neurosensory, without vestibular involvement, autosomal dominant | / |
| *ESRRB* | 602167 | Deafness, autosomal recessive 35 | 608565 |
| *EYA4* | 603550 | Cardiomyopathy, dilated, 1J | 605362 |
| | | Deafness, autosomal dominant 10 | 601316 |
| *FAM189A2* | 607710 | / | / |
| *GJB2* | 121011 | Bart-Pumphrey syndrome | 149200 |
| | | Deafness, autosomal dominant 3A | 601544 |
| | | Deafness, autosomal recessive 1A | 220290 |
| | | Hystrix-like ichthyosis with deafness | 602540 |
| | | Keratitis-ichthyosis-deafness syndrome | 148210 |
| | | Keratoderma, palmoplantar, with deafness | 148350 |
| *GJB3* | 603324 | Vohwinkel syndrome | 124500 |
| | | Deafness, autosomal dominant 2B | 612644 |
| | | Deafness, autosomal dominant, with peripheral neuropathy | / |
| | | Deafness, autosomal recessive | / |
| | | Deafness, digenic, GJB2/GJB3 | 220290 |
| | | Erythrokeratodermia variabilis et progressiva | 133200 |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *GJB6* | 604418 | Deafness, autosomal dominant 3B | 612643 |
| | | Deafness, autosomal recessive 1B | 612645 |
| | | Deafness, digenic GJB2/GJB6 | 220290 |
| | | Ectodermal dysplasia 2, Clouston type | 129500 |
| *GPSM2* | 609245 | Chudley-McCullough syndrome | 604213 |
| *GRHL2* | 608576 | Deafness, autosomal dominant 28 | 608641 |
| *GRXCR1* | 613283 | Deafness, autosomal recessive 25 | 613285 |
| *HGF* | 142409 | Deafness, autosomal recessive 39 | 608265 |
| *KCNQ4* | 603537 | Deafness, autosomal dominant 2A | 600101 |
| *LHFPL5* | 609427 | Deafness, autosomal recessive 67 | 610265 |
| *LOXHD1* | 613072 | Deafness, autosomal recessive 77 | 613079 |
| *LRTOMT* | 612414 | Deafness, autosomal recessive 63 | 611451 |
| *MARVELD2* | 610572 | Deafness, autosomal recessive 49 | 610153 |
| *MYH14* | 608568 | Deafness, autosomal dominant 4A | 600652 |
| | | Peripheral neuropathy, myopathy, hoarseness, and hearing loss | 614369 |
| *MYH9* | 160775 | Deafness, autosomal dominant 17 | 603622 |
| | | Epstein syndrome | 153650 |
| | | Fechtner syndrome | 153640 |
| | | Macrothrombocytopenia and progressive sensorineural deafness | 600208 |
| | | May-Hegglin anomaly | 155100 |
| | | Sebastian syndrome | 605249 |
| *MYO15A* | 602666 | Deafness, autosomal recessive 3 | 600316 |
| *MYO1A* | 601478 | Deafness, autosomal dominant 48 | 607841 |
| *MYO3A* | 606808 | Deafness, autosomal recessive 30 | 607101 |
| *MYO6* | 600970 | Deafness, autosomal dominant 22 | 606346 |
| | | Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy | 606346 |
| | | Deafness, autosomal recessive 37 | 607821 |
| *MYO7A* | 276903 | Deafness, autosomal dominant 11 | 601317 |
| | | Deafness, autosomal recessive 2 | 600060 |
| | | Usher syndrome, type 1B | 276900 |
| *OTOA* | 607038 | Deafness, autosomal recessive 22 | 607039 |
| *OTOF* | 603681 | Auditory neuropathy, autosomal recessive, 1 | 601071 |
| | | Deafness, autosomal recessive 9 | 601071 |
| *P2RX2* | 600844 | Deafness, autosomal dominant 41 | 608224 |
| *PCDH15* | 605514 | Deafness, autosomal recessive 23 | 609533 |
| | | Usher syndrome, type 1D/F digenic | 601067 |
| | | Usher syndrome, type 1F | 602083 |
| *POU3F4* | 300039 | Deafness, X-linked 2 | 304400 |
| *POU4F3* | 602460 | Deafness, autosomal dominant 15 | 602459 |
| *PRPS1* | 311850 | Arts syndrome | 301835 |
| | | Charcot-Marie-Tooth disease, X-linked recessive, 5 | 311070 |
| | | Deafness, X-linked 1 | 304500 |
| | | Gout, PRPS-related | 300661 |
| | | Phosphoribosylpyrophosphate synthetase superactivity | 300661 |
| *PTPRQ* | 603317 | Deafness, autosomal recessive 84A | 613391 |
| *RDX* | 179410 | Deafness, autosomal recessive 24 | 611022 |
| *SIX1* | 601205 | Brachiootic syndrome 3 | 608389 |
| | | Deafness, autosomal dominant 23 | 605192 |
| *SLC17A8* | 607557 | Deafness, autosomal dominant 25 | 605583 |
| *SLC26A4* | 605646 | Deafness, autosomal recessive 4, with enlarged vestibular aqueduct | 600791 |
| | | Pendred syndrome | 274600 |
| *SLC26A5* | 604943 | Deafness, autosomal recessive 61 | 613865 |
| *SMPX* | 300226 | Deafness, X-linked 4 | 300066 |
| *STRC* | 606440 | Deafness, autosomal recessive 16 | 603720 |
| *SYNE4* | 615535 | Deafness, autosomal recessive 76 | 615540 |
| *TECTA* | 602574 | Deafness, autosomal dominant 8/12 | 601543 |
| | | Deafness, autosomal recessive 21 | 603629 |
| *TJP2* | 607709 | Hypercholanemia, familial | 607748 |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *TMC1* | 606706 | Deafness, autosomal dominant 36 | 606705 |
| | | Deafness, autosomal recessive 7 | 600974 |
| *TMIE* | 607237 | Deafness, autosomal recessive 6 | 600971 |
| *TMPRSS3* | 605511 | Deafness, autosomal recessive 8/10 | 601072 |
| *TPRN* | 613354 | Deafness, autosomal recessive 79 | 613307 |
| *TRIOBP* | 609761 | Deafness, autosomal recessive 28 | 609823 |
| *USH1C* | 605242 | Deafness, autosomal recessive 18A | 602092 |
| | | Usher syndrome, type 1C | 276904 |
| *WFS1* | 606201 | ?Cataract 41 | 116400 |
| | | Deafness, autosomal dominant 6/14/38 | 600965 |
| | | Wolfram syndrome | 222300 |
| | | Wolfram-like syndrome, autosomal dominant | 614296 |
| | | {Diabetes mellitus, noninsulin-dependent, association with} | 125853 |

**Genes involved in intellectual disability, X-linked:** 107 genes involved in X-linked intellectual disability based on (Lubs *et al.* 2012; Piton *et al.* 2013).

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *ABCD1* | 300371 | Adrenoleukodystrophy | 300100 |
| | | Adrenomyeloneuropathy, adult | 300100 |
| *ACSL4* | 300157 | Mental retardation, X-linked 63 | 300387 |
| *AFF2* | 300806 | Mental retardation, X-linked, FRAXE type | 309548 |
| *AGTR2* | 300034 | / | / |
| *AP1S2* | 300629 | Mental retardation, X-linked syndromic, Fried type | 300630 |
| *ARHGEF6* | 300267 | Mental retardation, X-linked 46 | 300436 |
| *ARHGEF9* | 300429 | Epileptic encephalopathy, early infantile, 8 | 300607 |
| *ARX* | 300382 | Epileptic encephalopathy, early infantile, 1 | 308350 |
| | | Hydranencephaly with abnormal genitalia | 300215 |
| | | Lissencephaly, X-linked 2 | 300215 |
| | | Mental retardation, X-linked 29 and others | 300419 |
| | | Partington syndrome | 309510 |
| | | Proud syndrome | 300004 |
| *ATP6AP2* | 300556 | ?Mental retardation, X-linked, syndromic, Hedera type | 300423 |
| | | ?Parkinsonism with spasticity, X-linked | 300911 |
| *ATP7A* | 300011 | Menkes disease | 309400 |
| | | Occipital horn syndrome | 304150 |
| | | Spinal muscular atrophy, distal, X-linked 3 | 300489 |
| *ATRX* | 300032 | Alpha-thalassemia myelodysplasia syndrome, somatic | 300448 |
| | | Alpha-thalassemia/mental retardation syndrome | 301040 |
| | | Mental retardation-hypotonic facies syndrome, X-linked | 309580 |
| *BCOR* | 300485 | Microphthalmia, syndromic 2 | 300166 |
| *BRWD3* | 300553 | Mental retardation, X-linked 93 | 300659 |
| *CACNA1F* | 300110 | Aland Island eye disease | 300600 |
| | | Cone-rod dystropy, X-linked, 3 | 300476 |
| | | Night blindness, congenital stationary (incomplete), 2A, X-linked | 300071 |
| *CASK* | 300172 | FG syndrome 4 | 300422 |
| | | Mental retardation and microcephaly with pontine and cerebellar hypoplasia | 300749 |
| | | Mental retardation, with or without nystagmus | 300422 |
| *CDKL5* | 300203 | Angelman syndrome-like | 105830 |
| | | Epileptic encephalopathy, early infantile, 2 | 300672 |
| *CLCN4* | 302910 | / | / |
| *CNKSR2* | 300724 | | |
| *CUL4B* | 300304 | Mental retardation, X-linked, syndromic 15 (Cabezas type) | 300354 |
| *DCX* | 300121 | Lissencephaly, X-linked | 300067 |
| | | Subcortical laminal heteropia, X-linked | 300067 |
| *DKC1* | 300126 | Dyskeratosis congenita, X-linked | 305000 |
| *DLG3* | 300189 | Mental retardation, X-linked 90 | 300850 |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *DMD* | 300377 | Becker muscular dystrophy | 300376 |
| | | Cardiomyopathy, dilated, 3B | 302045 |
| | | Duchenne muscular dystrophy | 310200 |
| *EIF2S3* | 300161 | / | / |
| *FANCB* | 300515 | Fanconi anemia, complementation group B | 300514 |
| *FGD1* | 300546 | Aarskog-Scott syndrome | 305400 |
| | | Mental retardation, X-linked syndromic 16 | 305400 |
| *FLNA* | 300017 | Cardiac valvular dysplasia, X-linked | 314400 |
| | | Congenital short bowel syndrome | 300048 |
| | | FG syndrome 2 | 300321 |
| | | Frontometaphyseal dysplasia | 305620 |
| | | Heterotopia, periventricular | 300049 |
| | | Heterotopia, periventricular, ED variant | 300537 |
| | | Intestinal pseudoobstruction, neuronal | 300048 |
| | | Melnick-Needles syndrome | 309350 |
| | | Otopalatodigital syndrome, type I | 311300 |
| | | Otopalatodigital syndrome, type II | 304120 |
| | | Terminal osseous dysplasia | 300244 |
| *FMR1* | 309550 | Fragile X syndrome | 300624 |
| | | Fragile X tremor/ataxia syndrome | 300623 |
| | | Premature ovarian failure 1 | 311360 |
| *FRMPD4* | 300838 | / | / |
| *FTSJ1* | 300499 | Mental retardation, X-linked 9 | 309549 |
| *GDI1* | 300104 | Mental retardation, X-linked 41 | 300849 |
| *GK* | 300474 | Glycerol kinase deficiency | 307030 |
| *GPC3* | 300037 | Simpson-Golabi-Behmel syndrome, type 1 | 312870 |
| | | Wilms tumor, somatic | 194070 |
| *GRIA3* | 305915 | Mental retardation, X-linked 94 | 300699 |
| *HCCS* | 300056 | Microphthalmia, syndromic 7 | 309801 |
| *HCFC1* | 300019 | Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type ) | 309541 |
| *HDAC8* | 300269 | Cornelia de Lange syndrome 5 | 300882 |
| | | Wilson-Turner syndrome | 309585 |
| *HPRT* | 308000 | HPRT-related gout | 300323 |
| | | Lesch-Nyhan syndrome | 300322 |
| *HSD17B10* | 300256 | / | / |
| *HUWE1* | 300697 | Mental retardation, X-linked syndromic, Turner type | 300706 |
| *IDS* | 300823 | Mucopolysaccharidosis II | 309900 |
| *IGBP1* | 300139 | Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia | 300472 |
| *IKBKG* | 300248 | Ectodermal dysplasia, hypohidrotic, with immune deficiency | 300291 |
| | | Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency | 300301 |
| | | Immunodeficiency, isolated | 300584 |
| | | Incontinentia pigmenti, type II | 308300 |
| | | Invasive pneumococcal disease, recurrent isolated, 2 | 300640 |
| | | {Atypical mycobacteriosis, familial} | 300636 |
| *IL1RAPL* | 300206 | Mental retardation, X-linked 21/34 | 300143 |
| *IQSEC2* | 300522 | Mental retardation, X-linked 1 | 309530 |
| *KDM5C* | 314690 | Mental retardation, X-linked, syndromic, Claes-Jensen type | 300534 |
| *KIAA2022* | 300524 | Mental retardation, X-linked 98 | 300912 |
| *KLF8* | 300286 | / | / |
| *L1CAM* | 308840 | Corpus callosum, partial agenesis of | 304100 |
| | | CRASH syndrome | 303350 |
| | | Hydrocephalus due to aqueductal stenosis | 307000 |
| | | Hydrocephalus with congenital idiopathic intestinal pseudoobstruction | 307000 |
| | | Hydrocephalus with Hirschsprung disease | 307000 |
| | | MASA syndrome | 303350 |
| *LAMP2* | 309060 | Danon disease | 300257 |
| *LAS1L* | – | | |
| *MAGT1* | 300715 | Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia | 300853 |
| *MAOA* | 309850 | Brunner syndrome | 300615 |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *MBTPS2* | 300294 | ?Olmsted syndrome, X-linked | 300918 |
| | | IFAP syndrome with or without BRESHECK syndrome | 308205 |
| | | Keratosis follicularis spinulosa decalvans, X-linked | 308800 |
| *MECP2* | 300005 | Angelman syndrome | 105830 |
| | | Encephalopathy, neonatal severe | 300673 |
| | | Mental retardation, X-linked syndromic, Lubs type | 300260 |
| | | Mental retardation, X-linked, syndromic 13 | 300055 |
| | | Rett syndrome | 312750 |
| | | Rett syndrome, preserved speech variant | 312750 |
| | | {Autism susceptibility, X-linked 3} | 300496 |
| *MED12* | 300188 | Lujan-Fryns syndrome | 309520 |
| | | Ohdo syndrome, X-linked | 300895 |
| | | Opitz-Kaveggia syndrome | 305450 |
| *MID1* | 300552 | Opitz GBBB syndrome, type I | 300000 |
| *MTM1* | 300415 | Myotubular myopathy, X-linked | 310400 |
| *NAA10* | 300013 | ?Microphthalmia, syndrome 1 | 309800 |
| | | N-terminal acetyltransferase deficiency | 300855 |
| *NDP* | 300658 | Exudative vitreoretinopathy, X-linked | 305390 |
| | | Norrie disease | 310600 |
| *NDUFA1* | 300078 | Mitochondrial complex I deficiency | 252010 |
| *NHS* | 300457 | Cataract 40, X-linked | 302200 |
| | | Nance-Horan syndrome | 302350 |
| *NLGN3* | 300336 | {Asperger syndrome susceptibility, X-linked 1} | 300494 |
| | | {Autism susceptibility, X-linked 1} | 300425 |
| *NLGN4* | 300427 | Mental retardation, X-linked | 300495 |
| | | {Asperger syndrome susceptibility, X-linked 2} | 300497 |
| | | {Autism susceptibility, X-linked 2} | 300495 |
| *NSDHL* | 300275 | CHILD syndrome | 308050 |
| | | CK syndrome | 300831 |
| *NXF5* | 300319 | / | / |
| *OCRL1* | 300535 | Dent disease 2 | 300555 |
| | | Lowe syndrome | 309000 |
| *OFD1* | 300170 | ?Retinitis pigmentosa 23 | 300424 |
| | | Joubert syndrome 10 | 300804 |
| | | Oral-facial-digital syndrome 1 | 311200 |
| | | Simpson-Golabi-Behmel syndrome, type 2 | 300209 |
| *OPHN1* | 300127 | Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance | 300486 |
| *OTC* | 300461 | Ornithine transcarbamylase deficiency 311250 3 | 311250 |
| *PAK3* | 300142 | Mental retardation, X-linked 30/47 300558 3 | 300558 |
| *PCDH19* | 300460 | Epileptic encephalopathy, early infantile, 9 | 300088 |
| *PDHA1* | 300502 | Leigh syndrome, X-linked | 308930 |
| | | Pyruvate dehydrogenase E1-alpha deficiency | |
| *PGK1* | 311800 | Phosphoglycerate kinase 1 deficiency | 300653 |
| *PHF6* | 300414 | Borjeson-Forssman-Lehmann syndrome | 301900 |
| *PHF8* | 300560 | Mental retardation syndrome, X-linked, Siderius type | 300263 |
| *PLP* | 300401 | Pelizaeus-Merzbacher disease | 312080 |
| | | Spastic paraplegia 2, X-linked | 312920 |
| *PORCN* | 300651 | Focal dermal hypoplasia | 305600 |
| *PQBP1* | 300463 | Renpenning syndrome | 309500 |
| *PRPS1* | 311850 | Arts syndrome | 301835 |
| | | Charcot-Marie-Tooth disease, X-linked recessive, 5 | 311070 |
| | | Deafness, X-linked 1 | 304500 |
| | | Gout, PRPS-related | 300661 |
| | | Phosphoribosylpyrophosphate synthetase superactivity | 300661 |
| *PTCHD1* | 300828 | / | / |
| *RAB39B* | 300774 | Mental retardation, X-linked 72 | 300271 |
| *RAB40AL* | 300405 | Mental retardation, X-linked, syndromic, Martin-Probst type | 300519 |
| *RBM10* | 300080 | TARP syndrome | 311900 |
| *RPL10* | 312173 | {Autism, susceptibility to, X-linked 5} | 300847 |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *RPS6KA3* | 300075 | Coffin-Lowry syndrome | 303600 |
| | | Mental retardation, X-linked 19 | 300844 |
| *SHROOM4* | 300579 | Stocco dos Santos X-linked mental retardation syndrome | 300434 |
| *SLC16A2* | 300095 | Allan-Herndon-Dudley syndrome | 300523 |
| *SLC6A8* | 300036 | Cerebral creatine deficiency syndrome 1 | 300352 |
| *SLC9A6* | 300231 | Mental retardation, X-linked syndromic, Christianson type | 300243 |
| *SMC1A* | 300040 | Cornelia de Lange syndrome 2 | 300590 |
| *SMS* | 300105 | Mental retardation, X-linked, Snyder-Robinson type | 309583 |
| *SOX3* | 313430 | Mental retardation, X-linked, with isolated growth hormone deficiency | 300123 |
| | | Panhypopituitarism, X-linked | 312000 |
| *SRPX2* | 300642 | Rolandic epilepsy, mental retardation, and speech dyspraxia | 300643 |
| *SYN1* | 313440 | Epilepsy, X-linked, with variable learning disabilities and behavior disorders | 300491 |
| *SYP* | 313475 | Mental retardation, X-linked 96 | 300802 |
| *THOC2* | 300395 | / | / |
| *TIMM8A* | 300356 | Deafness, X-linked 1, progressive | |
| | | Jensen syndrome | 311150 |
| | | Mohr-Tranebjaerg syndrome | 304700 |
| *TSPAN7* | 300096 | Mental retardation, X-linked 58 | 300210 |
| *UBE2A* | 312180 | Mental retardation, X-linked syndromic, Nascimento-type | 300860 |
| *UPF3B* | 300298 | Mental retardation, X-linked, syndromic 14 | 300676 |
| *ZDHHC15* | 300576 | ?Mental retardation, X-linked 91 | 300577 |
| *ZDHHC9* | 300646 | Mental retardation, X-linked syndromic, Raymond type | 300799 |
| *ZNF41* | 314995 | / | / |
| *ZNF674* | 300573 | / | / |
| *ZNF711* | 314990 | Mental retardation, X-linked 97 | 300803 |
| *ZNF81* | 314998 | Mental retardation, X-linked 45 | 300498 |

**Genes involved in intellectual disability, autosomal recessive:** 39 genes involved in autosomal recessive intellectual disability (Musante and Ropers 2014).

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *ADK* | 102750 | Hypermethioninemia due to adenosine kinase deficiency | 614300 |
| *ADRA2B* | 104260 | / | / |
| *ASCC3* | 614217 | / | / |
| *ASCL1* | 100790 | Central hypoventilation syndrome, congenital | 209880 |
| | | Haddad syndrome | 209880 |
| *ARL14EP* | 612295 | / | / |
| *CASP2* | 600639 | / | / |
| *CC2D1A* | 610055 | Mental retardation, autosomal recessive 3 | 608443 |
| *CCNA2* | 123835 | / | / |
| *COQ5* | – | | |
| *CRADD* | | Mental retardation, autosomal recessive 34 | 614499 |
| *CRBN* | 609262 | Mental retardation, autosomal recessive 2 | 607417 |
| *EEF1B2* | 600655 | / | / |
| *ELP2* | – | | |
| *ENTPD1* | 601752 | Spastic paraplegia 64 | 615683 |
| *FASN* | 600212 | / | / |
| *GRIK2* | 138244 | Mental retardation, autosomal recessive, 6 | 611092 |
| *HIST3H3* | 602820 | / | / |
| *INPP4A* | 600916 | / | / |
| *KIAA1033* | 6015748 | / | / |
| *MAN1B1* | 604346 | Mental retardation, autosomal recessive 15 | 614202 |
| *MED23* | 605042 | Mental retardation, autosomal recessive 18 | 614249 |
| *NDST1* | 600853 | / | / |
| *PECR* | 605843 | / | / |
| *PRMT9* | – | | |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *PRRT2* | 614386 | Convulsions, familial infantile, with paroxysmal choreoathetosis | 602066 |
| | | Episodic kinesigenic dyskinesia 1 | 128200 |
| | | Seizures, benign familial infantile, 2 | 605751 |
| *PRSS12* | 606709 | Mental retardation, autosomal recessive 1 | 249500 |
| *RABL6* | 610615 | / | / |
| *RALGDS* | 601619 | / | / |
| *RGS7* | 602517 | / | / |
| *SCAPER* | 611611 | / | / |
| *ST3GAL3* | 606494 | Epileptic encephalopathy, early infantile, 15 | 615006 |
| | | Mental retardation, autosomal recessive 12 | 611090 |
| *TECR* | 610057 | Mental retardation, autosomal recessive 14 | 614020 |
| *TRAPPC9* | 611966 | Mental retardation, autosomal recessive 13 | 613192 |
| *TRMT1* | 611669 | / | / |
| *TTI2* | 614426 | Mental retardation, autosomal recessive 39 | 615541 |
| *TUSC3* | 601385 | Mental retardation, autosomal recessive 7 | 611093 |
| *UBR7* | 613816 | / | / |
| *ZC3H14* | 613279 | / | / |
| *ZNF526* | 614387 | / | / |

**Genes involved in intellectual disability, autosomal dominant:** 37 genes involved in autosomal dominant intellectual disability based on an internal literature survey as no consensual review is currently available.

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| *ANKRD11* | 611192 | KBG syndrome | 148050 |
| *ARID1B* | 614556 | Mental retardation, autosomal dominant 12 | 614562 |
| *CACNG2* | 602911 | Mental retardation, autosomal dominant 10 | 614256 |
| *CDH15* | 114019 | Mental retardation, autosomal dominant 3 | 612580 |
| *CREBBP* | 600140 | Rubinstein-Taybi syndrome | 180849 |
| *DOCK8* | 611432 | Hyper-IgE recurrent infection syndrome, autosomal recessive | 243700 |
| | | Mental retardation, autosomal dominant 2 | 614113 |
| *DYRK1A* | 600855 | Mental retardation, autosomal dominant 7 | 614104 |
| *EPB41L1* | 602879 | ?Mental retardation, autosomal dominant 11 | 614257 |
| *FOLR1* | 136430 | Neurodegeneration due to cerebral folate transport deficiency | 613068 |
| *FOXG1* | 164874 | Rett syndrome, congenital variant | 613454 |
| *FOXP1* | 605515 | Mental retardation with language impairment and autistic features | 613670 |
| *GRIN1* | 611239 | / | / |
| *GRIN2A* | 138253 | Epilepsy, focal, with speech disorder and with or without mental retardation | 245570 |
| *GRIN2B* | 138252 | Mental retardation, autosomal dominant 6 | 613970 |
| *HDAC4* | 605314 | Brachydactyly-mental retardation syndrome | 600430 |
| *HRAS* | 190020 | Congenital myopathy with excess of muscle spindles | 218040 |
| | | Costello syndrome | 218040 |
| | | Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic | 163200 |
| | | {Bladder cancer, somatic} | 109800 |
| | | {Nevus sebaceous, somatic} | 162900 |
| | | {Thyroid carcinoma, follicular, somatic} | 188470 |
| *KCNK9* | 605874 | Birk-Barel mental retardation dysmorphism syndrome | 612292 |
| *KCNQ2* | 602235 | Epileptic encephalopathy, early infantile, 7 | 613720 |
| | | Myokymia | 121200 |
| | | Seizures, benign neonatal, 1 | 121200 |
| *KIF1A* | 601255 | Mental retardation, autosomal dominant 9 | 614255 |
| | | Neuropathy, hereditary sensory, type IIC | 614213 |
| | | Spastic paraplegia 30, autosomal recessive | 610357 |
| *KIRREL3* | 607761 | Mental retardation, autosomal dominant 4 | 612581 |
| *MBD5* | 611472 | Mental retardation, autosomal dominant 1 | 156200 |
| *MEF2C* | 600662 | Chromosome 5q14.3 deletion syndrome | 613443 |
| | | Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations | 613443 |
| *NRXN1* | 600565 | Pitt-Hopkins-like syndrome 2 | 614325 |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | Phenotype MIM number |
|---|---|---|---|
| | | {Schizophrenia, susceptibility to, 17} | 614332 |
| *NRXN2* | 600566 | / | / |
| *PACS1* | 607492 | Mental retardation, autosomal dominant 17 | 615009 |
| *PAFAH1B1* | 601545 | Lissencephaly 1 | 607432 |
| | | Subcortical laminar heterotopia | 607432 |
| *PRODH* | 606810 | Hyperprolinemia, type I | 239500 |
| | | {Schizophrenia, susceptibility to, 4} | 600850 |
| *RAI1* | 607642 | Smith-Magenis syndrome | 182290 |
| *SCN1A* | 182389 | Dravet syndrome | 607208 |
| | | Epilepsy, generalized, with febrile seizures plus, type 2 | 604403 |
| | | Febrile seizures, familial, 3A | 604403 |
| | | Migraine, familial hemiplegic, 3 | 609634 |
| *SCN2A* | 182390 | Epileptic encephalopathy, early infantile, 11 | 613721 |
| | | Seizures, benign familial infantile, 3 | 607745 |
| *SHANK2* | 603290 | {Autism susceptibility 17} | 613436 |
| *SHANK3* | 606230 | Phelan-McDermid syndrome | 606232 |
| | | {Schizophrenia 15} | 613950 |
| *SLC2A1* | 138140 | Dystonia 9 | 601042 |
| | | GLUT1 deficiency syndrome 1 | 606777 |
| | | GLUT1 deficiency syndrome 2 | 612126 |
| | | {Epilepsy, idiopathic generalized, suscpetibility to, 12} | 614847 |
| *STXBP1* | 602926 | Epileptic encephalopathy, early infantile, 4 | 612164 |
| *SYNGAP1* | 603384 | Mental retardation, autosomal dominant 5 | 612621 |
| *TCF4* | 602272 | Pitt-Hopkins syndrome | 610954 |
| *ZEB2* | 605802 | Mowat-Wilson syndrome | 235730 |

**Fifty seven genes for which the American College of Medical Genetics and Genomics recommends to report incidental findings to patients:** as referenced in (Green *et al.* 2013).

| HUGO | OMIM number | Phenotype | MIM phenotype number |
|---|---|---|---|
| *BRCA1* | 113705 | Hereditary Breast and | 604370, |
| *BRCA2* | 600185 | Ovarian Cancer | 612555 |
| *TP53* | 191170 | Li-Fraumeni Syndrome | 151623 |
| *STK11* | 602216 | Peutz-Jeghers Syndrome | 175200 |
| *MLH1* | 120436 | | |
| *MSH2* | 609309 | Lynch Syndrome | 120435 |
| *MSH6* | 600678 | | |
| *PMS2* | 600259 | | |
| *APC* | 611731 | Familial adenomatous polyposis | 175100 |
| *MUTYH* | 604933 | MYH-Associated Polyposis; Adenomas, multiple colorectal, FAP type 2; Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas | 608456, 132600 |
| *VHL* | 608537 | Von Hippel Lindau syndrome | 193300 |
| *MEN1* | 613733 | Multiple Endocrine Neoplasia Type 1 | 131100 |
| *RET* | 164761 | Multiple Endocrine Neoplasia Type 2; Familial Medullary Thyroid Cancer (FMTC) | 171400, 162300, 1552401 |
| *NTRK1* | 191315 | Familial Medullary Thyroid Cancer (FMTC) | 1552401 |
| *PTEN* | 601728 | PTEN Hamartoma Tumor Syndrome | 153480 |
| *RB1* | 614041 | Retinoblastoma | 180200 |
| *SDHD* | 602690 | Hereditary | 168000 (PGL1) |
| *SDHAF2* | 613019 | Paraganglioma- | 601650 (PGL2) |
| *SDHC* | 602413 | Pheochromocytoma | 605373 (PGL3) |
| *SDHB* | 185470 | Syndrome | 115310 (PGL4) |
| *TSC1* | 605284 | Tuberous Sclerosis | 191100, |
| *TSC2* | 191092 | Complex | 613254 |
| *WT1* | 607102 | WT1-related Wilms | 194070 |
| *NF2* | 607379 | Neurofibromatosis type 2 | 101100 |
| *COL3A1* | 120180 | EDS - vascular type | 130050 |

**Table 1** (*contd*)

| HUGO | OMIM number | Phenotype | MIM phenotype number |
|---|---|---|---|
| *FBN1* | 134797 | | 154700, |
| *TGFBR1* | 190181 | Marfan Syndrome, | 609192, |
| *TGFBR2* | 190182 | Loeys-Dietz Syndromes, | 608967, |
| *SMAD3* | 603109 | and Familial Thoracic Aortic Aneurysms and | 610168, |
| *ACTA2* | 102620 | Dissections | 610380, |
| *MYLK* | 600922 | | 613795, |
| *MYH11* | 160745 | | 611788 |
| *MYBPC3* | 600958 | | 115197, |
| *MYH7* | 160760 | | 192600, |
| *TNNT2* | 191045 | | 601494, |
| *TNNI3* | 191044 | Hypertrophic | 613690, |
| *TPM1* | 191010 | cardiomyopathy, | 115196, |
| *MYL3* | 160790 | Dilated cardiomyopathy | 608751, |
| *ACTC1* | 102540 | | 612098, |
| *PRKAG2* | 602743 | | 600858, |
| *GLA* | 300644 | | 301500, |
| *MYL2* | 160781 | | 608758, |
| *LMNA* | 150330 | | 115200 |
| *RYR2* | 180902 | Catecholaminergic polymorphic ventricular tachycardia | 604772 |
| *PKP2* | 602861 | Arrhythmogenic right | 609040, |
| *DSP* | 125647 | ventricular cardiomyopathy | 604400, |
| *DSC2* | 125645 | | 610476, |
| *TMEM43* | 612048 | | 607450, |
| *DSG2* | 125671 | | 610193 |
| *KCNQ1* | 607542 | Romano-Ward Long QT | 192500, |
| *KCNH2* | 152427 | Syndromes Types 1, 2, | 613688, |
| *SCN5A* | 600163 | and 3, Brugada Syndrome | 603830, 601144 |
| *LDLR* | 606945 | Familial | 143890, |
| *APOB* | 107730 | hypercholesterolemia | 603776 |
| *PCSK9* | 607786 | | |
| *RYR1* | 180901 | Malignant hyperthermia | 145600 |
| *CACNA1S* | 114208 | susceptibility | |

**Table 2.** Detailed mean sequence coverage at a depth of ≥20× among 31 simplex exome runs for the analyzed gene lists.

**Genes involved in myopathies:**

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| 100% mean sequence coverage at ≥20× | | |
| *CAV3* | NM_001234 | 100 |
| *ISCU* | NM_014301 | 100 |
| *DPM3* | NM_153741 | 100 |
| <100% and ≥90% mean sequence coverage at ≥20× | | |
| *SGCA* | NM_000023 | 94.98 |
| *CAPN3* | NM_000070 | 96.08 |
| *EMD* | NM_000117 | 90.12 |
| *SGCG* | NM_000231 | 99.49 |
| *MTM1* | NM_000252 | 94.01 |
| *MYH7* | NM_000257 | 98.16 |
| *SGCD* | NM_000337 | 98.45 |
| *LAMA2* | NM_000426 | 98.92 |
| *DNM2* | NM_001005360 | 95 |
| *FKTN* | NM_001079802 | 98.04 |
| *ALG13* | NM_001099922 | 90.95 |
| *ACVR1* | NM_001111067 | 99.61 |
| *GNE* | NM_001128227 | 98.45 |

**Table 2** (*contd*)

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| *DYSF* | NM_001130978 | 92.56 |
| *ITGA7* | NM_001144997 | 95.76 |
| *FHL1* | NM_001159702 | 98.16 |
| *NEB* | NM_001164507 | 91.53 |
| *DAG1* | NM_001165928 | 99.33 |
| *LDB3* | NM_001171610 | 94.17 |
| *POMGNT1* | NM_001243766 | 99.58 |
| *CNTN1* | NM_001843 | 98.84 |
| *CRYAB* | NM_001885 | 99.79 |
| *DES* | NM_001927 | 94.1 |
| *LAMP2* | NM_002294 | 97.34 |
| *TCAP* | NM_003673 | 97.08 |
| *DPM1* | NM_003859 | 91.63 |
| *DMD* | NM_004006 | 96.26 |
| *COL6A3* | NM_004369 | 99.25 |
| *LARGE* | NM_004737 | 99.85 |
| *CHKB* | NM_005198 | 95.28 |
| *MSTN* | NM_005259 | 99.63 |
| *MYOT* | NM_006790 | 99.6 |
| *B3GNT1* | NM_006876 | 98.19 |
| *POMT1* | NM_007171 | 98.85 |
| *TRIM32* | NM_012210 | 95.34 |
| *TNPO3* | NM_012470 | 98.67 |
| *POMT2* | NM_013382 | 93.41 |
| *SMCHD1* | NM_015295 | 95.01 |
| *MYH2* | NM_017534 | 99.15 |
| *KLHL9* | NM_018847 | 99.4 |
| *GMPPB* | NM_021971 | 97.48 |
| *TIA1* | NM_022173 | 97.52 |
| *POMK* | NM_032237 | 99.96 |
| *MEGF10* | NM_032446 | 96.3 |
| *POMGNT2* | NM_032806 | 99.92 |
| *DNAJB6* | NM_058246 | 97.11 |
| *BIN1* | NM_139343 | 92.86 |
| *TPM3* | NM_152263 | 97.69 |
| *KLHL40* | NM_152393 | 93.31 |
| *SYNE2* | NM_182914 | 98.51 |
| *SYNE1* | NM_182961 | 97.08 |
| *TRAPPC11* | NM_199053 | 96.8 |
| *MATR3* | NM_199189 | 97.42 |
| *ANO5* | NM_213599 | 98.95 |
| *TPM2* | NM_213674 | 99.23 |
| <90% mean sequence coverage at ≥20× | | |
| *SGCB* | NM_000232 | 76.69 |
| *MYBPC3* | NM_000256 | 88.66 |
| *PLEC* | NM_000445 | 77.51 |
| *RYR1* | NM_000540 | 89.55 |
| *ACTA1* | NM_001100 | 76.12 |
| *KBTBD13* | NM_001101362 | 54.34 |
| *LMNA* | NM_001257374 | 81.4 |
| *TTN* | NM_001267550 | 85.99 |
| *FLNC* | NM_001458 | 89.87 |
| *COL6A1* | NM_001848 | 86.07 |
| *COL6A2* | NM_001849 | 76.85 |
| *TNNT1* | NM_003283 | 71.84 |
| *DPM2* | NM_003863 | 75.23 |
| *BAG3* | NM_004281 | 74.91 |
| *PABPN1* | NM_004643 | 85.71 |
| *VCP* | NM_007126 | 89.3 |
| *PTRF* | NM_012232 | 74.2 |
| *PTPLA* | NM_014241 | 69.39 |

**Table 2** (*contd*)

| | | |
|---|---|---|
| *TMEM5* | NM_014254 | 82.77 |
| *SEPN1* | NM_020451 | 84.91 |
| *FKRP* | NM_024301 | 63.67 |
| *TMEM43* | NM_024334 | 86.36 |
| *CFL2* | NM_138638 | 75 |
| *B3GALNT2* | NM_152490 | 85.97 |

**Genes involved in hereditary motor and sensory neuropathies:**

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| 100% mean sequence coverage at ≥20× | | |
| *PMP22* | NM_000304 | 100 |
| *HOXD10* | NM_002148 | 100 |
| *NGF* | NM_002506 | 100 |
| *PRPS1* | NM_002764 | 100 |
| <100% and ≥90% mean sequence coverage at ≥20× | | |
| *MPZ* | NM_000530 | 98.18 |
| *DNM2* | NM_001005360 | 95 |
| *TFG* | NM_001007565 | 97.21 |
| *SLC12A6* | NM_001042495 | 99.23 |
| *GJB1* | NM_001097642 | 97.03 |
| *ATL1* | NM_001127713 | 97.49 |
| *KARS* | NM_001130089 | 99.01 |
| *DNMT1* | NM_001130823 | 91.88 |
| *AIFM1* | NM_001130847 | 96.66 |
| *EGR2* | NM_001136178 | 97.07 |
| *PDK3* | NM_001142386 | 96.48 |
| *MTMR2* | NM_001243571 | 99.84 |
| *KIF1A* | NM_001244008 | 94.05 |
| *DYNC1H1* | NM_001376 | 98.1 |
| *AARS* | NM_001605 | 99.93 |
| *GARS* | NM_002047 | 91.24 |
| *VRK1* | NM_003384 | 99.19 |
| *IKBKAP* | NM_003640 | 96.93 |
| *YARS* | NM_003680 | 98.87 |
| *RAB7A* | NM_004637 | 99.93 |
| *CTDP1* | NM_004715 | 91.1 |
| *SPTLC2* | NM_004863 | 93.07 |
| *MARS* | NM_004990 | 96.86 |
| *NDRG1* | NM_006096 | 99.94 |
| *NEFL* | NM_006158 | 97.21 |
| *FBLN5* | NM_006329 | 95.22 |
| *SPTLC1* | NM_006415 | 99.26 |
| *HSPB8* | NM_014365 | 97.86 |
| *ARHGEF10* | NM_014629 | 97.22 |
| *FIG4* | NM_014845 | 97.42 |
| *MFN2* | NM_014874 | 97.04 |
| *KIF1B* | NM_015074 | 99.17 |
| *GDAP1* | NM_018972 | 99.81 |
| *WNK1* | NM_018979 | 97.61 |
| *TRPV4* | NM_021625 | 95.56 |
| *GNB4* | NM_021629 | 97.94 |
| *GAN* | NM_022041 | 92.14 |
| *SH3TC2* | NM_024577 | 99.13 |
| *SBF2* | NM_030962 | 96.1 |
| *HK1* | NM_033500 | 95.9 |
| *FGD4* | NM_139241 | 91.15 |
| <90% mean sequence coverage at ≥20× | | |
| *LRSAM1* | NM_001005373 | 85.7 |
| *SEPT9* | NM_001113491 | 84.36 |
| *LITAF* | NM_001136473 | 87.8 |

**Table 2** (*contd*)

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| *LMNA* | NM_001257374 | 81.4 |
| *HSPB1* | NM_001540 | 69.59 |
| *SBF1* | NM_002972 | 89.35 |
| *HINT1* | NM_005340 | 89.09 |
| *INF2* | NM_022489 | 80.92 |
| *MED25* | NM_030973 | 77.19 |
| *PRX* | NM_181882 | 72.65 |

**Genes involved in early onset epileptic encephalopathies:**

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| 100% mean sequence coverage at ≥20× | | |
| *MAPK10* | NM_002753 | 100 |
| <100% and ≥90% mean sequence coverage at ≥20× | | |
| *GRIN2A* | NM_000833 | 99.58 |
| *SCN2A* | NM_001040142 | 97.05 |
| *NTNG1* | NM_001113226 | 98.38 |
| *SPTAN1* | NM_001130438 | 97.64 |
| *PCDH19* | NM_001184880 | 96.51 |
| *MEF2C* | NM_001193347 | 99.35 |
| *SCN1A* | NM_001202435 | 95.56 |
| *COL4A1* | NM_001845 | 97.23 |
| *COL4A2* | NM_001846 | 98.91 |
| *STXBP1* | NM_003165 | 91.43 |
| *CASK* | NM_003688 | 92.09 |
| *MECP2* | NM_004992 | 91.86 |
| *MAGI2* | NM_012301 | 96.48 |
| *SRGAP2* | NM_015326 | 94.86 |
| *PNPO* | NM_018129 | 96.88 |
| *PLCB1* | NM_182734 | 95.55 |
| *GABRG2* | NM_198903 | 99.58 |
| <90% mean sequence coverage at ≥20× | | |
| *CDKL5* | NM_001037343 | 88.46 |
| *SLC25A22* | NM_001191061 | 82.36 |
| *SYP* | NM_003179 | 82.02 |
| *ST3GAL5* | NM_003896 | 85.17 |
| *FOXG1* | NM_005249 | 60.78 |
| *SLC2A1* | NM_006516 | 80.19 |
| *SYN1* | NM_006950 | 80.47 |
| *PNKP* | NM_007254 | 89.83 |
| *SNPH* | NM_014723 | 88.13 |
| ARHGEF9 | NM_015185 | 89.82 |
| *ARX* | NM_139058 | 42.93 |
| *KCNQ2* | NM_172107 | 84.8 |

**Genes involved in isolated and combined dystonia:**

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| 100% mean sequence coverage at ≥20× | | |
| (–) | | |
| <100% and ≥90% mean sequence coverage at ≥20× | | |
| *TOR1A* | NM_000113 | 90.5 |
| *GCH1* | NM_001024024 | 90.28 |
| *GNAL* | NM_001142339 | 99.11 |
| *TAF1* | NM_004606 | 94.79 |

**Table 2** (*contd*)

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| *PNKD* | NM_015488 | 99.29 |
| *THAP1* | NM_018105 | 98.57 |
| <90% mean sequence coverage at ≥20× | | |
| *SGCE* | NM_001099401 | 87.97 |
| *SPR* | NM_003124 | 66.89 |
| *SLC2A1* | NM_006516 | 80.19 |
| *PRRT2* | NM_145239 | 85.68 |
| *ATP1A3* | NM_152296 | 87.84 |
| *TH* | NM_199292 | 78.81 |

**Genes involved in non syndromic deafness and hereditary hearing loss:**

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| 100% mean sequence coverage at ≥20× | | |
| *GRXCR1* | NM_001080476 | 100 |
| *PRPS1* | NM_002764 | 100 |
| *GJB2* | NM_004004 | 100 |
| <100% and ≥90% mean sequence coverage at ≥20× | | |
| *MYO7A* | NM_000260 | 95.92 |
| *POU3F4* | NM_000307 | 99.14 |
| *SLC26A4* | NM_000441 | 95.04 |
| *HGF* | NM_000601 | 94.93 |
| *MARVELD2* | NM_001038603 | 97.95 |
| *TRIOBP* | NM_001039141 | 90.07 |
| *SYNE4* | NM_001039876 | 96.31 |
| *DFNB59* | NM_001042702 | 92.47 |
| *GJB6* | NM_001110219 | 98.99 |
| *FAM189A2* | NM_001127608 | 96.64 |
| *PCDH15* | NM_001142770 | 96.62 |
| *PTPRQ* | NM_001145026 | 97.03 |
| *LRTOMT* | NM_001145308 | 91.56 |
| *LOXHD1* | NM_001145472 | 96.85 |
| *CLDN14* | NM_001146077 | 98.95 |
| *CDH23* | NM_001171933 | 96 |
| *MYO1A* | NM_001256041 | 97.51 |
| *MYH9* | NM_002473 | 92.47 |
| *POU4F3* | NM_002700 | 99.51 |
| *COCH* | NM_004086 | 96.14 |
| *EYA4* | NM_004100 | 99.54 |
| *ESRRB* | NM_004452 | 98.99 |
| *TJP2* | NM_004817 | 95.77 |
| *MYO6* | NM_004999 | 95.72 |
| *TECTA* | NM_005422 | 99.56 |
| *WFS1* | NM_006005 | 93.88 |
| *GPSM2* | NM_013296 | 98.44 |
| *SMPX* | NM_014332 | 99.99 |
| *DFNB31* | NM_015404 | 97.43 |
| *MYO15A* | NM_016239 | 94.51 |
| *MYO3A* | NM_017433 | 96.62 |
| *GJB3* | NM_024009 | 99.23 |
| *TMPRSS3* | NM_024022 | 99.04 |
| *GRHL2* | NM_024915 | 97.61 |
| *BSND* | NM_057176 | 93.96 |
| *TMC1* | NM_138691 | 94.48 |
| *SLC17A8* | NM_139319 | 96.75 |
| *USH1C* | NM_153676 | 96.38 |
| *CCDC50* | NM_178335 | 94.66 |
| *LHFPL5* | NM_182548 | 98.24 |
| *OTOF* | NM_194248 | 92.98 |

**Table 2** (*contd*)

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| <90% mean sequence coverage at ≥20× | | |
| *SLC26A5* | NM_206883 | 99.4 |
| *TPRN* | NM_001128228 | 81.75 |
| *MYH14* | NM_001145809 | 85.97 |
| *RDX* | NM_001260493 | 86.42 |
| *ACTG1* | NM_001614 | 79.53 |
| *DFNA5* | NM_004403 | 88.5 |
| *KCNQ4* | NM_004700 | 83.78 |
| *DIAPH1* | NM_005219 | 89.13 |
| *SIX1* | NM_005982 | 89.69 |
| *DSPP* | NM_014208 | 83.17 |
| *ESPN* | NM_031475 | 62.63 |
| *COL11A2* | NM_080681 | 88.29 |
| *OTOA* | NM_144672 | 82.79 |
| *TMIE* | NM_147196 | 78.83 |
| *STRC* | NM_153700 | 57 |
| *P2RX2* | NM_174873 | 69.74 |

**Genes involved in intellectual disability, X-linked:**

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| 100% mean sequence coverage at ≥20× | | |
| *AGTR2* | NM_000686 | 100 |
| *PRPS1* | NM_002764 | 100 |
| <100% and ≥90% mean sequence coverage at ≥20× | | |
| *ATP7A* | NM_000052 | 96.39 |
| *IDS* | NM_000202 | 95.46 |
| *MTM1* | NM_000252 | 94.01 |
| *NDP* | NM_000266 | 99.94 |
| *OCRL* | NM_000276 | 94.68 |
| *PGK1* | NM_000291 | 92.93 |
| *OTC* | NM_000531 | 92.13 |
| *GRIA3* | NM_000828 | 96.36 |
| *KIAA2022* | NM_001008537 | 98.09 |
| *PHF6* | NM_001015877 | 97.18 |
| *FANCB* | NM_001018113 | 99.59 |
| *RAB40AL* | NM_001031834 | 99.39 |
| *PQBP1* | NM_001032381 | 99.96 |
| *THOC2* | NM_001081550 | 90.96 |
| *FLNA* | NM_001110556 | 91.75 |
| *BCOR* | NM_001123383 | 97.79 |
| *PLP1* | NM_001128834 | 96.74 |
| *NSDHL* | NM_001129765 | 93.62 |
| *NHS* | NM_001136024 | 98.51 |
| *GPC3* | NM_001164617 | 93.56 |
| *SLC9A6* | NM_001177651 | 96.85 |
| *PCDH19* | NM_001184880 | 96.51 |
| *PHF8* | NM_001184896 | 96.72 |
| *MAOA* | NM_001270458 | 98.19 |
| *L1CAM* | NM_001278116 | 92.57 |
| *DKC1* | NM_001363 | 92.73 |
| *EIF2S3* | NM_001415 | 95.01 |
| *IGBP1* | NM_001551 | 99.76 |
| *CLCN4* | NM_001830 | 94.37 |
| *FMR1* | NM_002024 | 93.79 |
| *AFF2* | NM_002025 | 90.1 |
| *LAMP2* | NM_002294 | 97.34 |
| *OPHN1* | NM_002547 | 95.1 |
| *PAK3* | NM_002578 | 92.06 |

**Table 2** (*contd*)

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| *CUL4B* | NM_003588 | 95.63 |
| *OFD1* | NM_003611 | 92.78 |
| *CASK* | NM_003688 | 92.09 |
| *AP1S2* | NM_003916 | 94.87 |
| *DMD* | NM_004006 | 96.26 |
| *TIMM8A* | NM_004085 | 98.98 |
| *KDM5C* | NM_004187 | 97.21 |
| *FGD1* | NM_004463 | 93.7 |
| *NDUFA1* | NM_004541 | 99.94 |
| *RPS6KA3* | NM_004586 | 90.96 |
| *SMS* | NM_004595 | 90.34 |
| *TSPAN7* | NM_004615 | 93.33 |
| *ARHGEF6* | NM_004840 | 95.94 |
| *MECP2* | NM_004992 | 91.86 |
| *MED12* | NM_005120 | 94 |
| *CACNA1F* | NM_005183 | 90.53 |
| *HCCS* | NM_005333 | 97.34 |
| *HCFC1* | NM_005334 | 93.39 |
| *RPL10* | NM_006013 | 98.21 |
| *ZNF41* | NM_007130 | 99.66 |
| *ZNF81* | NM_007137 | 99.86 |
| *IL1RAPL1* | NM_014271 | 94.34 |
| *FRMPD4* | NM_014728 | 91.48 |
| *CNKSR2* | NM_014927 | 94.93 |
| *MBTPS2* | NM_015884 | 96.29 |
| *ZDHHC9* | NM_016032 | 97.94 |
| *SHROOM4* | NM_020717 | 96.01 |
| *DLG3* | NM_021120 | 91.63 |
| *ACSL4* | NM_022977 | 97.99 |
| *HUWE1* | NM_031407 | 95.24 |
| *MAGT1* | NM_032121 | 95.79 |
| *NXF5* | NM_032946 | 92.35 |
| *ZDHHC15* | NM_144969 | 97.65 |
| *BRWD3* | NM_153252 | 92.02 |
| *RAB39B* | NM_171998 | 91.62 |
| *PTCHD1* | NM_173495 | 95.46 |
| *FTSJ1* | NM_177439 | 96.96 |
| *DCX* | NM_178152 | 96.34 |
| *NLGN3* | NM_181303 | 92.1 |
| *NLGN4X* | NM_181332 | 99.73 |
| *PORCN* | NM_203475 | 92.93 |
| <90% mean sequence coverage at ≥20× | | |
| *ABCD1* | NM_000033 | 79.45 |
| *HPRT1* | NM_000194 | 85.56 |
| *ATRX* | NM_000489 | 88.41 |
| *CDKL5* | NM_001037343 | 88.46 |
| *ZNF674* | NM_001039891 | 84.35 |
| *IKBKG* | NM_001099856 | 29.41 |
| *IQSEC2* | NM_001111125 | 81.54 |
| *PDHA1* | NM_001173454 | 79.61 |
| *GDI1* | NM_001493 | 88.02 |
| *SYP* | NM_003179 | 82.02 |
| *UBE2A* | NM_003336 | 80.03 |
| *NAA10* | NM_003491 | 87.5 |
| *HSD17B10* | NM_004493 | 87.48 |
| *SLC6A8* | NM_005629 | 76.22 |
| *SOX3* | NM_005634 | 21.28 |
| *RBM10* | NM_005676 | 87.08 |
| *ATP6AP2* | NM_005765 | 85.5 |
| *SMC1A* | NM_006306 | 86.37 |
| *SLC16A2* | NM_006517 | 83.32 |
| *SYN1* | NM_006950 | 80.47 |

**Table 2** (*contd*)

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| *KLF8* | NM_007250 | 82.29 |
| *SRPX2* | NM_014467 | 89.83 |
| *ARHGEF9* | NM_015185 | 89.82 |
| *HDAC8* | NM_018486 | 84.25 |
| *ZNF711* | NM_021998 | 89.13 |
| *LAS1L* | NM_031206 | 84.92 |
| *MID1* | NM_033290 | 88.14 |
| *UPF3B* | NM_080632 | 83.85 |
| *ARX* | NM_139058 | 42.93 |
| *GK* | NM_203391 | 84.67 |

**Genes involved in intellectual disability, autosomal recessive:**

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| 100% mean sequence coverage at ≥20× | | |
| *HIST3H3* | NM_003493 | 100 |
| *CRADD* | NM_003805 | 100 |
| *ARL14EP* | NM_152316 | 100 |
| <100% and ≥90% mean sequence coverage at ≥20× | | |
| *RALGDS* | NM_001042368 | 92 |
| *TTI2* | NM_001102401 | 99.91 |
| *TRMT1* | NM_001136035 | 91.48 |
| *SCAPER* | NM_001145923 | 98.05 |
| *CCNA2* | NM_001237 | 99.99 |
| *ELP2* | NM_001242875 | 97.87 |
| *NDST1* | NM_001543 | 97.34 |
| *RGS7* | NM_002924 | 98.13 |
| *PRSS12* | NM_003619 | 95.23 |
| *INPP4A* | NM_004027 | 99.1 |
| *ADK* | NM_006721 | 99 |
| *ASCC3* | NM_006828 | 96.95 |
| *KIAA1033* | NM_015275 | 95.92 |
| *MED23* | NM_015979 | 98.9 |
| *MAN1B1* | NM_016219 | 96.48 |
| *CRBN* | NM_016302 | 90.47 |
| *PECR* | NM_018441 | 99.38 |
| *EEF1B2* | NM_021121 | 99.78 |
| *ZC3H14* | NM_024824 | 93.45 |
| *TRAPPC9* | NM_031466 | 98.98 |
| *COQ5* | NM_032314 | 93.93 |
| *CASP2* | NM_032982 | 99.6 |
| *PRMT9* | NM_138364 | 99.49 |
| *ST3GAL3* | NM_174963 | 98.02 |
| *UBR7* | NM_175748 | 97.48 |
| *GRIK2* | NM_175768 | 97.96 |
| <90% mean sequence coverage at ≥20× | | |
| *ADRA2B* | NM_000682 | 85.09 |
| *ENTPD1* | NM_001098175 | 87.65 |
| *FASN* | NM_004104 | 80.1 |
| *ASCL1* | NM_004316 | 51.77 |
| *TUSC3* | NM_006765 | 87.15 |
| *CC2D1A* | NM_017721 | 81.44 |
| *RABL6* | NM_024718 | 86.6 |
| *ZNF526* | NM_133444 | 85.6 |
| *TECR* | NM_138501 | 89.57 |
| *PRRT2* | NM_145239 | 85.68 |

**Table 2** (*contd*)

**Genes involved in intellectual disability, autosomal dominant:**

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| 100% mean sequence coverage at ≥20× | | |
| (–) | | |
| <100% and ≥90% mean sequence coverage at ≥20× | | |
| *GRIN2A* | NM_000833 | 99.58 |
| *GRIN2B* | NM_000834 | 98.18 |
| *SCN2A* | NM_001040142 | 97.05 |
| *NRXN1* | NM_001135659 | 97.93 |
| *MEF2C* | NM_001193347 | 99.35 |
| *SCN1A* | NM_001202435 | 95.56 |
| *TCF4* | NM_001243226 | 96.43 |
| *KIF1A* | NM_001244008 | 94.05 |
| *ANKRD11* | NM_001256182 | 93.41 |
| *STXBP1* | NM_003165 | 91.43 |
| *CREBBP* | NM_004380 | 93.67 |
| *HRAS* | NM_005343 | 99.19 |
| *HDAC4* | NM_006037 | 96.7 |
| *SHANK2* | NM_012309 | 90.47 |
| *ZEB2* | NM_014795 | 99.68 |
| *FOLR1* | NM_016724 | 98.39 |
| *PACS1* | NM_018026 | 95.34 |
| *MBD5* | NM_018328 | 96.12 |
| *ARID1B* | NM_020732 | 91.33 |
| *RAI1* | NM_030665 | 95.06 |
| *KIRREL3* | NM_032531 | 97.91 |
| *FOXP1* | NM_032682 | 96.02 |
| *DYRK1A* | NM_101395 | 98.45 |
| *DOCK8* | NM_203447 | 97.63 |
| <90% mean sequence coverage at ≥20× | | |
| *PAFAH1B1* | NM_000430 | 88.39 |
| *GRIN1* | NM_001185090 | 68.52 |
| *EPB41L1* | NM_001258329 | 85.1 |
| *KCNK9* | NM_001282534 | 75.99 |
| *CDH15* | NM_004933 | 87.78 |
| *FOXG1* | NM_005249 | 60.78 |
| *CACNG2* | NM_006078 | 89.34 |
| *SLC2A1* | NM_006516 | 80.19 |
| *SYNGAP1* | NM_006772 | 76.19 |
| *NRXN2* | NM_015080 | 76.82 |
| *PRODH* | NM_016335 | 85.01 |
| *SHANK3* | NM_033517 | 73.42 |
| *KCNQ2* | NM_172107 | 84.8 |

**57 genes for which the American College of Medical Genetics and Genomics recommends to report incidental findings to patients:**

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| 100% mean sequence coverage at ≥20× | | |
| *ACTA2* | NM_001141945 | 100 |
| <100% and ≥90% mean sequence coverage at ≥20× | | |
| *BRCA2* | NM_000059 | 92.19 |
| *CACNA1S* | NM_000069 | 94.51 |
| *COL3A1* | NM_000090 | 96.67 |
| *FBN1* | NM_000138 | 98.91 |
| *GLA* | NM_000169 | 99.36 |
| *MYH7* | NM_000257 | 98.16 |
| *MYL3* | NM_000258 | 99.46 |
| *PTEN* | NM_000314 | 98.95 |

**Table 2** (*contd*)

| Gene symbol | Ref. Seq. ID | Mean coverage (%) |
|---|---|---|
| *RB1* | NM_000321 | 91.78 |
| *TSC1* | NM_000368 | 97.99 |
| *APOB* | NM_000384 | 95.48 |
| *MYL2* | NM_000432 | 95.06 |
| *LDLR* | NM_000527 | 95.6 |
| *TSC2* | NM_000548 | 94.17 |
| *VHL* | NM_000551 | 94.89 |
| *TNNT2* | NM_001001430 | 90.29 |
| *NTRK1* | NM_001007792 | 97.69 |
| *TGFBR2* | NM_001024847 | 93.57 |
| *RYR2* | NM_001035 | 97.41 |
| *MYH11* | NM_001040113 | 94.6 |
| *APC* | NM_001127510 | 96.15 |
| *SCN5A* | NM_001160160 | 97.65 |
| *MLH1* | NM_001258274 | 99.53 |
| *MSH2* | NM_001258281 | 97.08 |
| *SDHB* | NM_003000 | 91.91 |
| *SDHC* | NM_003001 | 96.8 |
| *DSP* | NM_004415 | 94.98 |
| *DSC2* | NM_004949 | 91.4 |
| *ACTC1* | NM_005159 | 96.51 |
| *BRCA1* | NM_007300 | 96.01 |
| *MUTYH* | NM_012222 | 96.61 |
| *NF2* | NM_016418 | 96.91 |
| *SDHAF2* | NM_017841 | 98.77 |
| *RET* | NM_020975 | 91.37 |
| *WT1* | NM_024424 | 95.17 |
| *MYLK* | NM_053025 | 92.44 |
| *PCSK9* | NM_174936 | 93.01 |
| <90% mean sequence coverage at ≥20× | | |
| *MSH6* | NM_000179 | 87.72 |
| *KCNQ1* | NM_000218 | 88.73 |
| *KCNH2* | NM_000238 | 59.03 |
| *MYBPC3* | NM_000256 | 88.66 |
| *TNNI3* | NM_000363 | 85.84 |
| *TPM1* | NM_000366 | 81.23 |
| *STK11* | NM_000455 | 70.65 |
| *PMS2* | NM_000535 | 77.96 |
| *RYR1* | NM_000540 | 89.55 |
| *TP53* | NM_001126113 | 89.06 |
| *LMNA* | NM_001257374 | 81.4 |
| *SDHD* | NM_001276506 | 74.4 |
| *DSG2* | NM_001943 | 89.38 |
| *PKP2* | NM_004572 | 89.29 |
| *TGFBR1* | NM_004612 | 88.17 |
| *SMAD3* | NM_005902 | 86.33 |
| *PRKAG2* | NM_016203 | 89.23 |
| *TMEM43* | NM_024334 | 86.36 |
| *MEN1* | NM_130804 | 86.62 |

## References

Amberger J., Bocchini C. A., Scott A. F. and Hamosh A. 2009 McKusick's Online Mendelian Inheritance in Man (OMIM). *Nucleic Acids Res.* **37**, D793–796.

Kaplan J. C. and Hamroun D. 2013 The 2014 version of the gene table of monogenic neuromuscular disorders (nuclear genome). *Neuromuscul Disord.* **23**, 1081–1111.

Kodera H., Kato M., Nord A. S., Walsh T., Lee M., Yamanaka G. *et al.* 2013 Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. *Epilepsia* **54**, 1262–1269.

Klein C. M. C. and Münchau A. 2003 *Dystonia overview*. In *Gene reviews* (ed. R. A. Pagon, M. P. Adam, H. H. Ardinger *et al.*). University of Washington, Seattle (http://www.ncbi.nlm.nih.gov/books/NBK1155/).

Smith R. J. H., Hildebrand M. S. *et al.* 1999. Deafness and hereditary hearing loss overview. In *Gene Reviews* (ed. R. A. Pagon, M. P. Adam, H. H. Ardinger *et al.*). University of Washington, Seattle (http://www.ncbi.nlm.nih.gov/books/NBK1434/).

Lubs H. A., Stevenson R. E. and Schwartz C. E. 2012 Fragile X and X-linked intellectual disability: four decades of discovery. *Am. J. Hum. Genet.* **90**, 579–590.

Piton A., Redin C. and Mandel J. L. 2013 XLID-causing mutations and associated genes challenged in light of data from large-scale human exome sequencing. *Am. J. Hum. Genet.* **93**, 368–383.

Musante L. and Ropers H. H. 2014 Genetics of recessive cognitive disorders. *Trends Genet.* **30**, 32–39.

Green R. C., Berg J. S., Grody W. W., Kalia S. S., Korf B. R., Martin C. L. *et al.* 2013 ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. *Genet. Med.* **15**, 565–574.