

Tabella 1: PrenatalScreen® - Elenco dei geni analizzati e della malattie genetiche investigate

| | MALATTIA | PhenoMIM | GENE |
|----|--------------------------------------------------------------------------------|----------|-------------------|
| 1 | 17-alpha-hydroxylase/17,20-lyase deficiency | 202110 | CYP17A1 |
| 2 | 17-beta-hydroxysteroid dehydrogenase X deficiency | 300438 | HSD17B10 |
| 3 | 3-beta-hydroxysteroid dehydrogenase, type II, deficiency | 201810 | HSD3B2 |
| 4 | 3-hydroxy-3-methylglutaric aciduria | 246450 | HMGCL |
| 5 | 3-methylglutaconic aciduria type 1 | 250950 | AUH |
| 6 | 3-methylglutaconic aciduria type 3 | 258501 | OPA3 |
| 7 | 46XY sex reversal 3 | 612965 | NR5A1 |
| 8 | 4-hydroxybutyric aciduria | 271980 | ALDH5A1 |
| 9 | Aarskog-Scott syndrome | 305400 | FGD1 |
| 10 | ABCD syndrome | 600501 | EDNRB |
| 11 | Acampomelic campomelic dysplasia | 114290 | SOX9 |
| 12 | Achalasia-addisonianism-alacrimia syndrome | 231550 | AAAS |
| 13 | Achondrogenesis type 1B | 600972 | SLC26A2 |
| 14 | Achondrogenesis, type IA | 200600 | TRIP11 |
| 15 | Achondrogenesis, type II or hypochondrogenesis | 200610 | COL2A1 |
| 16 | Achondroplasia | 100800 | FGFR3 |
| 17 | Acromicric dysplasia | 102370 | FBN1 |
| 18 | Acyl-CoA dehydrogenase 9 deficiency | 611126 | ACAD9 |
| 19 | Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency | 202010 | CYP11B1 |
| 20 | Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete | 613743 | CYP11A1 |
| 21 | Adrenocortical insufficiency | 612965 | NR5A1 |
| 22 | Adrenoleukodystrophy | 300100 | ABCD1 |
| 23 | Adult neuronal ceroid lipofuscinosis | 256730 | PPT1 |
| 24 | Adult neuronal ceroid lipofuscinosis 10 | 610127 | CTSD |
| 25 | Adult neuronal ceroid lipofuscinosis 4A | 204300 | CLN6 |
| 26 | Aicardi-Goutières syndrome | 225750 | TREX1 |
| 27 | Aicardi-Goutieres syndrome 2 | 610181 | RNASEH2B |
| 28 | Aicardi-Goutieres syndrome 3 | 610329 | RNASEH2C |
| 29 | Aicardi-Goutieres syndrome 4 | 610333 | RNASEH2A |
| 30 | Aicardi-Goutieres syndrome 5 | 612952 | SAMHD1 |
| 31 | Aicardi-Goutieres syndrome 6 | 615010 | ADAR |
| 32 | Aldosteronism, glucocorticoid-remediable | 103900 | CYP11B1 |
| 33 | Allan-Herndon-Dudley syndrome | 300523 | SLC16A2 |
| 34 | Alpers syndrome | 203700 | POLG |
| 35 | Alpha-methylacyl-Coa Racemase deficiency | 614307 | AMACR |
| 36 | Alpha-thalassemia | 604131 | HBA1- HBA2 |

| | | | |
|----|--------------------------------------------------------------------------------|--------|----------------------------|
| 37 | Alpha-thalassemia myelodysplasia syndrome, somatic | 300448 | ATRX |
| 38 | Alpha-thalassemia/mental retardation syndrome | 301040 | ATRX |
| 39 | Alport syndrome X-Linked | 301050 | COL4A5 |
| 40 | Alport syndrome autosomal recessive | 203780 | COL4A3 - COL4A4 |
| 41 | Alström syndrome | 203800 | ALMS1 |
| 42 | Amish infantile epilepsy syndrome | 609056 | ST3GAL5 |
| 43 | Amyotrophic lateral sclerosis 11 | 612577 | FIG4 |
| 44 | Amyotrophic lateral sclerosis 2, juvenile | 205100 | ALS2 |
| 45 | Anauxetic dysplasia | 607095 | RMRP |
| 46 | Andersen syndrome | 170390 | KCNJ2 |
| 47 | Angelman syndrome | 105830 | UBE3A |
| 48 | Antenatal Bartter syndrome | 241200 | KCNJ1 |
| 49 | Antenatal Bartter syndrome type 1 | 601678 | SLC12A1 |
| 50 | Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis | 201750 | POR |
| 51 | Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis | 207410 | FGFR2 |
| 52 | Apert syndrome | 101200 | FGFR2 |
| 53 | Aplasia/hypoplasia of limbs and pelvis | 276820 | WNT7A |
| 54 | Aplastic anemia | 609135 | NBN |
| 55 | Apparent mineralocorticoid excess | 218030 | HSD11B2 |
| 56 | Argininosuccinic aciduria | 207900 | ASL |
| 57 | Aromatic L-amino acid decarboxylase deficiency | 608643 | DDC |
| 58 | Arrhythmogenic right ventricular dysplasia 2 | 600996 | RYR2 |
| 59 | Arthrogyrosis - renal dysfunction - cholestasis | 208085 | VPS33B |
| 60 | Arthrogyrosis multiplex congenita, distal, type 1 | 108120 | TPM2 |
| 61 | Arthrogyrosis multiplex congenita, distal, type 2B | 601680 | TNNI2 |
| 62 | Arthrogyrosis, distal, type 2B | 601680 | TPM2 |
| 63 | Arthrogyrosis, renal dysfunction, and cholestasis 2 | 613404 | VIPAR |
| 64 | Asperger syndrome susceptibility, X-linked 1 | 300494 | NLGN3 |
| 65 | Ataxia - oculomotor apraxia type 1 | 208920 | APTX |
| 66 | Ataxia with vitamin E deficiency | 277460 | TTPA |
| 67 | Ataxia-telangiectasia | 208900 | ATM |
| 68 | Atelosteogenesis type II | 256050 | SLC26A2 |
| 69 | Atrial fibrillation, familial, 16 | 613120 | SCN3B |
| 70 | Atrial fibrillation, familial, 17 | 611819 | SCN4B |
| 71 | Atrial fibrillation, familial, 3 | 607554 | KCNQ1 |
| 72 | Atrial fibrillation, familial, 4 | 611493 | KCNE2 |
| 73 | Atrial fibrillation, familial, 9 | 613980 | KCNJ2 |
| 74 | Autism, susceptibility to, X-linked 5 | 300847 | RPL10 |

| | | | |
|-----|---------------------------------------------------------------------|--------------------------------|-----------------------------------------|
| 75 | Autoimmune lymphoproliferative syndrome type IV | 614470 | NRAS |
| 76 | Autoimmune lymphoproliferative syndrome, type IA | 601859 | FAS |
| 77 | Autoimmune lymphoproliferative syndrome, type IB | 601859 | FASLG |
| 78 | Autoimmune lymphoproliferative syndrome, type II | 603909 | CASP10 |
| 79 | Autoimmune polyendocrinopathy syndrome , type I | 240300 | AIRE |
| 80 | Autosomal dominant Charcot-Marie-Tooth disease type 2K | 607831 | GDAP1 |
| 81 | Autosomal recessive ataxia due to ubiquinone deficiency | 612016 | ADCK3 |
| 82 | Autosomal recessive Charcot-Marie-Tooth disease with hoarseness | 607706 | GDAP1 |
| 83 | Autosomal recessive distal spinal muscular atrophy type 4 | 611067 | PLEKHG5 |
| 84 | Autosomal recessive dopa-responsive dystonia | 605407 | TH |
| 85 | Autosomal recessive hypophosphatemic rickets 1 | 241520 | DMP1 |
| 86 | Autosomal recessive hypophosphatemic rickets 2 | 613312 | ENPP1 |
| 87 | Autosomal recessive intermediate Charcot-Marie-Tooth disease type A | 608340 | GDAP1 |
| 88 | Autosomal recessive limb-girdle muscular dystrophy type 2I | 607155 | FKRP |
| 89 | Autosomal recessive limb-girdle muscular dystrophy type C | 609308 - 613157 - 613158 | POMT1 - POMGNT1 - POMT2 |
| 90 | Autosomal recessive malignant osteopetrosis 1 | 259700 | TCIRG1 |
| 91 | Autosomal recessive malignant osteopetrosis 4 | 611490 | CLCN7 |
| 92 | Autosomal recessive nonsyndromic sensorineural deafness type DFNB12 | 601386 | CDH23 |
| 93 | Autosomal recessive nonsyndromic sensorineural deafness type DFNB18 | 602092 | USH1C |
| 94 | Autosomal recessive nonsyndromic sensorineural deafness type DFNB1A | 220290 | GJB2 |
| 95 | Autosomal recessive nonsyndromic sensorineural deafness type DFNB2 | 600060 | MYO7A |
| 96 | Autosomal recessive polycystic kidney disease | 263200 | PKHD1 |
| 97 | Autosomal recessive progressive external ophthalmoplegia | 258450 | POLG |
| 98 | Autosomal recessive spastic ataxia of Charlevoix-Saguenay | 270550 | SACS |
| 99 | Autosomal recessive spondylocostal dysostosis 1 | 277300 | DLL3 |
| 100 | Avascular necrosis of the femoral head | 608805 | COL2A1 |
| 101 | Bannayan-Riley-Ruvalcaba syndrome | 153480 | PTEN |
| 102 | Bardet-Biedl syndrome 11 | 615988 | TRIM32 |
| 103 | Basal cell carcinoma, somatic | 605462 | PTCH1 |
| 104 | Basal cell nevus syndrome | 109400 | PTCH1 |
| 105 | Beare-Stevenson cutis gyrata syndrome | 123790 | FGFR2 |
| 106 | Becker muscular dystrophy | 300376 | DMD |
| 107 | Beckwith-Wiedemann syndrome | 130650 | CDKN1C - H19 - NSD1 |
| 108 | Bent bone dysplasia syndrome | 614592 | FGFR2 |
| 109 | Beta-thalassemia | 613985 | HBB |
| 110 | Bethlem myopathy | 158810 | COL6A1 - COL6A2 - COL6A3 |

| | | | |
|-----|------------------------------------------------------------------------------------|--------|-----------------|
| 111 | Bifunctional enzyme deficiency | 261515 | HSD17B4 |
| 112 | Biotinidase deficiency | 253260 | BTD |
| 113 | Björnstad syndrome | 262000 | BCS1L |
| 114 | Bloom syndrome | 210900 | BLM |
| 115 | Borjeson-Forssman-Lehmann syndrome | 301900 | PHF6 |
| 116 | Brachyolmia type 3 | 113500 | TRPV4 |
| 117 | Brachytelephalangi chondrodysplasia punctata | 302950 | ARSE |
| 118 | Brittle cornea syndrome | 229200 | ZNF469 |
| 119 | Brugada syndrome 2 | 611777 | GPD1L |
| 120 | Brugada syndrome 7 | 613120 | SCN3B |
| 121 | Brunner syndrome | 300615 | MAOA |
| 122 | Buschke-Ollendorff syndrome | 166700 | LEMD3 |
| 123 | Caffey disease | 114000 | COL1A1 |
| 124 | Campomelic dysplasia with autosomal sex reversal | 114290 | SOX9 |
| 125 | Canavan disease | 271900 | ASPA |
| 126 | CAP myopathy 1 | 609284 | TPM3 |
| 127 | CAP myopathy 2 | 609285 | TPM2 |
| 128 | Carbamoylphosphate synthetase deficiency | 237300 | CPS1 |
| 129 | Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1 | 604377 | SCO2 |
| 130 | Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2 | 615119 | COX15 |
| 131 | Cardiofaciocutaneous syndrome | 115150 | BRAF |
| 132 | Cardiofaciocutaneous syndrome 2 | 615278 | KRAS |
| 133 | Cardiofaciocutaneous syndrome 3 | 615279 | MAP2K1 |
| 134 | Cardiofaciocutaneous syndrome 4 | 615280 | MAP2K2 |
| 135 | Cardiomyopathy, familial hypertrophic | 192600 | CAV3 |
| 136 | Carnitine deficiency, systemic primary | 212140 | SLC22A5 |
| 137 | Carnitine palmitoyl transferase 1A deficiency | 255120 | CPT1A |
| 138 | Carnitine palmitoyl transferase II deficiency, infantile form | 600649 | CPT2 |
| 139 | Carnitine palmitoyl transferase II deficiency, neonatal form | 608836 | CPT2 |
| 140 | Carnitine-acylcarnitine translocase deficiency | 212138 | SLC25A20 |
| 141 | Carpenter syndrome | 201000 | RAB23 |
| 142 | Cartilage-hair hypoplasia | 250250 | RMRP |
| 143 | Cataract - intellectual deficit - hypogonadism | 212720 | RAB3GAP2 |
| 144 | Cataract 40, X-linked | 302200 | NHS |
| 145 | CATSHL syndrome | 610474 | FGFR3 |
| 146 | Central core disease | 117000 | RYR1 |
| 147 | Central hypoventilation syndrome, congenital | 209880 | RET |
| 148 | Cerebellar ataxia - intellectual deficit - dysequilibrium syndrome | 224050 | VLDLR |

| | | | |
|-----|-----------------------------------------------------------------------------|--------|----------------|
| 149 | Cerebral dysgenesis-neuropathy-ichthyosis-palmoplantar keratoderma syndrome | 609528 | SNAP29 |
| 150 | Cerebrotendinous xanthomatosis | 213700 | CYP27A1 |
| 151 | Charcot-Marie-Tooth disease axonal type 2B1 | 605588 | LMNA |
| 152 | Charcot-Marie-Tooth disease type 4A | 214400 | GDAP1 |
| 153 | Charcot-Marie-Tooth disease type 4E | 605253 | EGR2 |
| 154 | Charcot-Marie-Tooth disease type 4F | 614895 | PRX |
| 155 | Charcot-Marie-Tooth disease type 4H | 609311 | FGD4 |
| 156 | Charcot-Marie-Tooth disease, axonal, type 2M | 606482 | DNM2 |
| 157 | Charcot-Marie-Tooth disease, dominant intermediate B | 606482 | DNM2 |
| 158 | Charcot-Marie-Tooth disease, type 1A | 118220 | PMP22 |
| 159 | Charcot-Marie-Tooth disease, type 1B | 118200 | MPZ |
| 160 | Charcot-Marie-Tooth disease, type 1C | 601098 | LITAF |
| 161 | Charcot-Marie-Tooth disease, type 1E | 118300 | PMP22 |
| 162 | Charcot-Marie-Tooth disease, type 1F | 607734 | NEFL |
| 163 | Charcot-Marie-Tooth disease, type 2A1 | 118210 | KIF1B |
| 164 | Charcot-Marie-Tooth disease, type 2A2 | 609260 | MFN2 |
| 165 | Charcot-Marie-Tooth disease, type 2B | 600882 | RAB7A |
| 166 | Charcot-Marie-Tooth disease, type 2D | 601472 | GARS |
| 167 | Charcot-Marie-Tooth disease, type 2E | 607684 | NEFL |
| 168 | Charcot-Marie-Tooth disease, type 2I | 607677 | MPZ |
| 169 | Charcot-Marie-Tooth disease, type 2J | 607736 | MPZ |
| 170 | Charcot-Marie-Tooth disease, type 4B1 | 601382 | MTMR2 |
| 171 | Charcot-Marie-Tooth disease, type 4B2 | 604563 | SBF2 |
| 172 | Charcot-Marie-Tooth disease, type 4C | 601596 | SH3TC2 |
| 173 | Charcot-Marie-Tooth disease, type 4D | 601455 | NDRG1 |
| 174 | Charcot-Marie-Tooth disease, type 4J | 611228 | FIG4 |
| 175 | Charcot-Marie-Tooth neuropathy, X-linked dominant, 1 | 302800 | GJB1 |
| 176 | CHARGE syndrome | 214800 | CHD7 |
| 177 | Chediak-Higashi syndrome | 214500 | LYST |
| 178 | Chilblain lupus 2 | 614415 | SAMHD1 |
| 179 | CHILD syndrome | 308050 | NSDHL |
| 180 | Childhood-onset hypophosphatasia | 241510 | ALPL |
| 181 | Cholestasis, benign recurrent intrahepatic | 243300 | ATP8B1 |
| 182 | Cholestasis, benign recurrent intrahepatic, 2 | 605479 | ABCB11 |
| 183 | Cholestasis, intrahepatic, of pregnancy, 1 | 147480 | ATP8B1 |
| 184 | Cholestasis, intrahepatic, of pregnancy, 3 | 614972 | ABCB4 |
| 185 | Cholestasis, progressive familial intrahepatic 1 | 211600 | ATP8B1 |
| 186 | Cholestasis, progressive familial intrahepatic 2 | 601847 | ABCB11 |
| 187 | Cholestasis, progressive familial intrahepatic 3 | 602347 | ABCB4 |

| | | | |
|-----|-------------------------------------------------------------------------|--------------------------------|---------------------------------------|
| 188 | Chondrodysplasia, Blomstrand type | 215045 | PTH1R |
| 189 | Ciliary dyskinesia, primary, 1 | 244400 | DNAI1 |
| 190 | Ciliary dyskinesia, primary, 3 | 608644 | DNAH5 |
| 191 | Citrullinemia type I | 215700 | ASS1 |
| 192 | CK syndrome | 300831 | NSDHL |
| 193 | Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency | 201910 | CYP21A2 |
| 194 | Classic galactosemia | 230400 | GALT |
| 195 | Classic maple syrup urine disease | 248600 | DBT |
| 196 | Classical homocystinuria | 236200 | CBS |
| 197 | COACH syndrome | 216360 | TMEM67 |
| 198 | Cockayne syndrome type A | 216400 | ERCC8 |
| 199 | Cockayne syndrome type B | 133540 | ERCC6 |
| 200 | Coenzyme Q10 deficiency, primary, 5 | 614654 | COQ9 |
| 201 | Coffin-Lowry syndrome | 303600 | RPS6KA3 |
| 202 | COFS syndrome 1 | 214150 | ERCC6 |
| 203 | Cohen Syndrome type 1 | 216550 | VPS13B |
| 204 | Combined immunodeficiency with skin granulomas | 233650 | RAG1 - RAG2 |
| 205 | Combined oxidative phosphorylation defect type 2 | 610498 | MRPS16 |
| 206 | Combined oxidative phosphorylation defect type 5 | 611719 | MRPS22 |
| 207 | Combined oxidative phosphorylation deficiency 4 | 610678 | TUFM |
| 208 | Combined pituitary hormone deficiencies, genetic forms | 182230 - 262600 - 613038 | HESX1 - PROP1 - POU1F1 |
| 209 | Combined pituitary hormone deficiency with spine abnormalities | 221750 | LHX3 |
| 210 | Complete androgen insensitivity syndrome | 300068 | AR |
| 211 | Complex I, mitochondrial respiratory chain, deficiency of | 252010 | NDUFS6 |
| 212 | Congenital bile acid synthesis defect type 4 | 214950 | AMACR |
| 213 | Congenital disorder of glycosylation type 1a | 212065 | PMM2 |
| 214 | Congenital disorder of glycosylation type 1b | 602579 | MPI |
| 215 | Congenital disorder of glycosylation type 1e | 608799 | DPM1 |
| 216 | Congenital disorder of glycosylation type 1j | 608093 | DPAGT1 |
| 217 | Congenital disorder of glycosylation type 2a | 212066 | MGAT2 |
| 218 | Congenital disorder of glycosylation type 2c | 266265 | SLC35C1 |
| 219 | Congenital disorder of glycosylation type 2d | 607091 | B4GALT1 |
| 220 | Congenital disorder of glycosylation type 2f | 603585 | SLC35A1 |
| 221 | Congenital disorder of glycosylation type 1c | 603147 | ALG6 |
| 222 | Congenital disorder of glycosylation type 1k | 608540 | ALG1 |
| 223 | Congenital disorder of glycosylation, type 1d | 601110 | ALG3 |
| 224 | Congenital disorder of glycosylation, type 1f | 609180 | MPDU1 |
| 225 | Congenital disorder of glycosylation, type 1g | 607143 | ALG12 |

| | | | |
|-----|------------------------------------------------------------------------------------------|--------------------------------|----------------------------------------|
| 226 | Congenital disorder of glycosylation, type Ih | 608104 | ALG8 |
| 227 | Congenital disorder of glycosylation, type Ii | 607906 | ALG2 |
| 228 | Congenital disorder of glycosylation, type IIb | 606056 | MOGS |
| 229 | Congenital disorder of glycosylation, type Iie | 608779 | COG7 |
| 230 | Congenital disorder of glycosylation, type IIg | 611209 | COG1 |
| 231 | Congenital disorder of glycosylation, type IIh | 611182 | COG8 |
| 232 | Congenital disorder of glycosylation, type II | 608776 | ALG9 |
| 233 | Congenital disorder of glycosylation, type Im | 610768 | DOLK |
| 234 | Congenital disorder of glycosylation, type In | 612015 | RFT1 |
| 235 | Congenital disorder of glycosylation, type Iq | 612379 | SRD5A3 |
| 236 | Congenital fibrinogen deficiency | 202400 | FGA |
| 237 | Congenital heart defects, nonsyndromic, 1, X-linked | 306955 | ZIC3 |
| 238 | Congenital hereditary endothelial dystrophy type II | 217700 | SLC4A11 |
| 239 | Congenital lipoid adrenal hyperplasia | 201710 | STAR |
| 240 | Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells | 610370 | NEUROG3 |
| 241 | Congenital muscular dystrophy type 1A | 607855 | LAMA2 |
| 242 | Congenital muscular dystrophy type 1D | 608840 | LARGE |
| 243 | Congenital muscular dystrophy type 5B | 606612 | FKRP |
| 244 | Congenital muscular dystrophy with cerebellar involvement | 613151 - 613155 - 613156 | POMGNT1 - POMT1 - POMT2 |
| 245 | Congenital myopathy with excess of muscle spindles | 218040 | HRAS |
| 246 | Corneal dystrophy - perceptive deafness | 217400 | SLC4A11 |
| 247 | Cornelia de Lange syndrome 1 | 122470 | NIPBL |
| 248 | Cornelia de Lange syndrome 2 | 300590 | SMC1A |
| 249 | Cornelia de Lange syndrome 3 | 610759 | SMC3 |
| 250 | Corpus callosum agenesis - neuronopathy | 218000 | SLC12A6 |
| 251 | Corpus callosum hypoplasia-retardation-adducted thumbs-spasticity-hydrocephalus syndrome | 307000 | L1CAM |
| 252 | Corpus callosum, agenesis of, with mental retardation | 300472 | IGBP1 |
| 253 | Costello syndrome | 218040 | HRAS |
| 254 | Cowden syndrome 1 | 158350 | PTEN |
| 255 | Craniofacial-deafness-hand syndrome | 122880 | PAX3 |
| 256 | Craniofacial-skeletal-dermatologic dysplasia | 101600 | FGFR2 |
| 257 | Craniofrontonasal dysplasia | 304110 | EFNB1 |
| 258 | Craniosynostosis, type 1 | 123100 | TWIST1 |
| 259 | Creatine phosphokinase, elevated serum | 123320 | CAV3 |
| 260 | Crouzon syndrome | 123500 | FGFR2 |
| 261 | Crouzon syndrome with acanthosis nigricans | 612247 | FGFR3 |
| 262 | Culler-Jones syndrome | 615849 | GLI2 |

| | | | |
|-----|---------------------------------------------------------|--------|--------------------|
| 263 | Cutis laxa, autosomal dominant 2 | 614434 | FBLN5 |
| 264 | Cutis laxa, autosomal recessive, type IA | 219100 | FBLN5 |
| 265 | Cutis laxa, autosomal recessive, type IB | 614437 | EFEMP2 |
| 266 | Cutis laxa, autosomal recessive, type IIA | 219200 | ATP6V0A2 |
| 267 | Cystic fibrosis; mucoviscidosis | 219700 | CFTR |
| 268 | Cystinosis | 219800 | CTNS |
| 269 | Czech dysplasia | 609162 | COL2A1 |
| 270 | Danon disease | 300257 | LAMP2 |
| 271 | Deafness - encephaloneuropathy - obesity - valvulopathy | 614651 | PDSS1 |
| 272 | Deafness, autosomal dominant 13 | 601868 | COL11A2 |
| 273 | Deafness, autosomal dominant 3B | 612643 | GJB6 |
| 274 | Deafness, autosomal dominant type 1 | 124900 | DIAPH1 |
| 275 | Deafness, autosomal dominant type 10 | 601316 | EYA4 |
| 276 | Deafness, autosomal dominant type 12 | 601543 | TECTA |
| 277 | Deafness, autosomal dominant type 15 | 602459 | POU4F3 |
| 278 | Deafness, autosomal dominant type 17 | 603622 | MYH9 |
| 279 | Deafness, autosomal dominant type 20 | 604717 | ACTG1 |
| 280 | Deafness, autosomal dominant type 22 | 606346 | MYO6 |
| 281 | Deafness, autosomal dominant type 23 | 605192 | SIX1 |
| 282 | Deafness, autosomal dominant type 25 | 605583 | SLC17A8 |
| 283 | Deafness, autosomal dominant type 28 | 608641 | GRHL2 |
| 284 | Deafness, autosomal dominant type 2A | 600101 | KCNQ4 |
| 285 | Deafness, autosomal dominant type 2B | 612644 | GJB3 |
| 286 | Deafness, autosomal dominant type 36 | 606705 | TMC1 |
| 287 | Deafness, autosomal dominant type 4 | 600652 | MYH14 |
| 288 | Deafness, autosomal dominant type 40 | 123740 | CRYM |
| 289 | Deafness, autosomal dominant type 48 | 607841 | MYO1A |
| 290 | Deafness, autosomal dominant type 5 | 600994 | DFNA5 |
| 291 | Deafness, autosomal dominant type 50 | 613074 | MIR96 |
| 292 | Deafness, autosomal dominant type 6 | 600965 | WFS1 |
| 293 | Deafness, autosomal dominant type 64 | 614152 | DIABLO |
| 294 | Deafness, autosomal dominant type 9 | 601369 | COCH |
| 295 | Deafness, autosomal recessive 1B | 612645 | GJB6 |
| 296 | Deafness, autosomal recessive 53 | 609706 | COL11A2 |
| 297 | Deafness, digenic GJB2/GJB6 | 220290 | GJB6 |
| 298 | Dejerine-Sottas disease | 145900 | MPZ - PMP22 |
| 299 | Dent disease | 300009 | CLCN5 |
| 300 | Dent disease 2 | 300555 | OCRL |
| 301 | Denys-Drash syndrome | 194080 | WT1 |

| | | | |
|-----|--------------------------------------------------------------------|--------------------|-----------------------------------------|
| 302 | Dermatopathia pigmentosa reticularis | 125595 | KRT14 |
| 303 | Desmosterolosis | 602398 | DHCR24 |
| 304 | Diabetes mellitus, noninsulin-dependent | 125853 | ABCC8 |
| 305 | Diabetes mellitus, permanent neonatal | 606176 | ABCC8 |
| 306 | Diabetes mellitus, transient neonatal 2 | 610374 | ABCC8 |
| 307 | Diastrophic dwarfism | 222600 | SLC26A2 |
| 308 | Digital arthropathy-brachydactyly, familial | 606835 | TRPV4 |
| 309 | Dihydropyrimidine dehydrogenase deficiency | 274270 | DPYD |
| 310 | Dilated cardiomyopathy with ataxia | 610198 | DNAJC19 |
| 311 | Donnai-Barrow syndrome | 222448 | LRP2 |
| 312 | Dowling-Degos disease 1 | 179850 | KRT5 |
| 313 | Duchenne muscular dystrophy | 310200 | DMD |
| 314 | Dyschromatosis symmetrica hereditaria | 127400 | ADAR |
| 315 | Dyskeratosis congenita X-linked | 305000 | DKC1 |
| 316 | Dystonia-1, torsion | 128100 | TOR1A |
| 317 | Dystonia-11, myoclonic | 159900 | SGCE |
| 318 | Dystrophic epidermolysis bullosa pruriginosa | 604129 | COL7A1 |
| 319 | Early infantile epileptic encephalopathy | 308350 - 609304 | ARX - SLC25A22 |
| 320 | Ectodermal dysplasia 1, hypohidrotic, X-linked | 305100 | EDA |
| 321 | Ectodermal dysplasia 2, Clouston type | 129500 | GJB6 |
| 322 | Ectodermal dysplasia, hypohidrotic, with immune deficiency | 300291 | IKBK |
| 323 | Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency | 300301 | IKBK |
| 324 | Ectopia lentis, familial | 129600 | FBN1 |
| 325 | Ehlers-Danlos syndrome type 6 | 225400 | PLOD1 |
| 326 | Ehlers-Danlos syndrome, cardiac valvular type | 225320 | COL1A2 |
| 327 | Ehlers-Danlos syndrome, type I | 130000 | COL5A1 - COL5A2 - COL1A1 |
| 328 | Ehlers-Danlos syndrome, type II | 130010 | COL5A1 |
| 329 | Ehlers-Danlos syndrome, type III | 130020 | COL3A1 |
| 330 | Ehlers-Danlos syndrome, type IV | 130050 | COL3A1 |
| 331 | Ehlers-Danlos syndrome, type VIIA | 130060 | COL1A1 |
| 332 | Eiken syndrome | 600002 | PTH1R |
| 333 | Elliptocytosis-1 | 611804 | EPB41 |
| 334 | Ellis-van Creveld syndrome | 225500 | EVC2 |
| 335 | Emery-Dreifuss muscular dystrophy 1, X-linked | 310300 | EMD |
| 336 | Encephalopathy due to prosaposin deficiency | 611721 | PSAP |
| 337 | Encephalopathy, progressive, with or without lipodystrophy | 615924 | BSC12 |
| 338 | Epidermolysis bullosa simplex with muscular dystrophy | 226670 | PLEC |

| | | | |
|-----|------------------------------------------------------------------------------------|--------|---------------------|
| 339 | Epidermolysis bullosa simplex with pyloric atresia | 612138 | PLEC |
| 340 | Epidermolysis bullosa simplex, Dowling-Meara type | 131760 | KRT14 - KRT5 |
| 341 | Epidermolysis bullosa simplex, Koebner type | 131900 | KRT14 - KRT5 |
| 342 | Epidermolysis bullosa simplex, recessive 1 | 601001 | KRT14 - KRT5 |
| 343 | Epidermolysis bullosa simplex, Weber-Cockayne type | 131800 | KRT14 - KRT5 |
| 344 | Epidermolysis bullosa simplex-MP | 131960 | KRT5 |
| 345 | Epidermylysis bullosa simplex-MCR | 609352 | KRT5 |
| 346 | Epilepsy, familial temporal lobe, 1 | 600512 | LGI1 |
| 347 | Epilepsy, progressive myoclonic 2A (Lafora) | 254780 | EPM2A |
| 348 | Epilepsy, progressive myoclonic 2B (Lafora) | 254780 | NHLRC1 |
| 349 | Epilepsy, pyridoxine-dependent | 266100 | ALDH7A1 |
| 350 | Epilepsy, X-linked, with variable learning disabilities and behavior disorders | 300491 | SYN1 |
| 351 | Epileptic encephalopathy, early infantile, 15 | 615006 | ST3GAL3 |
| 352 | Epileptic encephalopathy, early infantile, 2 | 300672 | CDKL5 |
| 353 | Epileptic encephalopathy, early infantile, 8 | 300607 | ARHGEF9 |
| 354 | Epileptic encephalopathy, early infantile, 9 | 300088 | PCDH19 |
| 355 | Epiphyseal dysplasia, multiple 1 | 132400 | COMP |
| 356 | Epiphyseal dysplasia, multiple, with myopia and deafness | 132450 | COL2A1 |
| 357 | Erythrocytosis, familial, 2 | 263400 | VHL |
| 358 | Escobar syndrome | 265000 | CHRNA3 |
| 359 | Ethylmalonic encephalopathy | 602473 | ETHE1 |
| 360 | Exostoses, multiple, type 1 | 133700 | EXT1 |
| 361 | Exostoses, multiple, type 2 | 133701 | EXT2 |
| 362 | Exudative vitreoretinopathy 2, X-linked | 305390 | NDP |
| 363 | Fabry disease | 301500 | GLA |
| 364 | Failure of tooth eruption, primary | 125350 | PTH1R |
| 365 | Familial dysautonomia | 223900 | IKBKAP |
| 366 | Familial hypomagnesemia - hypercalciuria - nephrocalcinosis - severe ocular | 248190 | CLDN19 |
| 367 | Familial Mediterranean fever | 249100 | MEFV |
| 368 | Fanconi anemia complementation group C | 227645 | FANCC |
| 369 | Fanconi anemia, complementation group A | 227650 | FANCA |
| 370 | Fanconi anemia, complementation group B | 300514 | FANCB |
| 371 | Fanconi anemia, complementation group G | 614082 | FANCG |
| 372 | Fatal infantile lactic acidosis with methylmalonic aciduria | 245400 | SUCLG1 |
| 373 | Fatal mitochondrial disease due to combined oxidative phosphorylation deficiency 3 | 610505 | TSM1 |
| 374 | Favism | 134700 | G6PD |
| 375 | Fertile eunuch syndrome | 228300 | GNRHR |

| | | | |
|-----|---------------------------------------------------------------------------------------------------------------------|--------|--------------------------------|
| 376 | Fetal akinesia deformation sequence | 208150 | RAPSN DOK7 |
| 377 | Fetal Gaucher disease | 608013 | GBA |
| 378 | FG syndrome 4 | 300422 | CASK |
| 379 | Fibrochondrogenesis | 228520 | COL11A1 |
| 380 | Fibrochondrogenesis 2 | 614524 | COL11A2 |
| 381 | Fibromatosis, gingival | 135300 | SOS1 |
| 382 | Fibular hypoplasia or aplasia - femoral bowing - oligodactyly | 228930 | WNT7A |
| 383 | Focal cortical dysplasia, Taylor balloon cell type | 607341 | TSC1 |
| 384 | Focal dermal hypoplasia | 305600 | PORCN |
| 385 | Fraser syndrome (gene FRAS1) | 219000 | FRAS1 - FREM2 |
| 386 | Frasier syndrome | 136680 | WT1 |
| 387 | Free sialic acid storage disease, infantile form | 269920 | SLC17A5 |
| 388 | French-Canadian type Leigh syndrome | 220111 | LRPPRC |
| 389 | Fucosidosis | 230000 | FUCA1 |
| 390 | Fumaric aciduria | 606812 | FH |
| 391 | Galactokinase deficiency with cataracts | 230200 | GALK1 |
| 392 | Gallbladder disease 1 | 600803 | ABCB4 |
| 393 | Gaucher disease type 2 | 230900 | GBA |
| 394 | Gaucher disease type 3 | 231000 | GBA |
| 395 | Gaucher disease type 3C | 231005 | GBA |
| 396 | Geleophysic dysplasia 1 | 231050 | ADAMTSL2 |
| 397 | Geleophysic dysplasia 2 | 614185 | FBN1 |
| 398 | Generalized junctional epidermolysis bullosa, non-Herlitz type | 226650 | COL17A1 |
| 399 | Glutaric acidemia type 2 | 231680 | ETFA - ETFB - ETFDH |
| 400 | Glutaryl-CoA dehydrogenase deficiency | 231670 | GCDH |
| 401 | Glutathione synthetase deficiency with 5-oxoprolinuria | 266130 | GSS |
| 402 | Glycerol kinase deficiency | 307030 | GK |
| 403 | Glycine encephalopathy | 605899 | AMT - GCSH - GLDC |
| 404 | Glycogen storage disease due to acid maltase deficiency | 232300 | GAA |
| 405 | Glycogen storage disease due to glucose-6-phosphatase deficiency type 1a | 232200 | G6PC |
| 406 | Glycogen storage disease due to glucose-6-phosphatase deficiency type b | 232220 | SLC37A4 |
| 407 | Glycogen storage disease due to glucose-6-phosphatase deficiency type c | 232240 | SLC37A4 |
| 408 | Glycogen storage disease due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form | 232500 | GBE1 |
| 409 | Glycogen storage disease due to glycogen debranching enzyme deficiency | 232400 | AGL |
| 410 | Glycogen storage disease due to muscle glycogen phosphorylase deficiency | 232600 | PYGM |
| 411 | GM1 gangliosidosis type 1 | 230500 | GLB1 |
| 412 | GM1 gangliosidosis type 2 | 230600 | GLB1 |

| | | | |
|-----|----------------------------------------------------------------------------------|--------|---------------|
| 413 | GM1 gangliosidosis type 3 | 230650 | GLB1 |
| 414 | Gnathodiaphyseal dysplasia | 166260 | ANO5 |
| 415 | GRACILE syndrome | 603358 | BCS1L |
| 416 | Greenberg dysplasia | 215140 | LBR |
| 417 | GrisCELLI disease type 1 | 214450 | MYO5A |
| 418 | GrisCELLI disease type 2 | 607624 | RAB27A |
| 419 | Guanidinoacetate methyltransferase deficiency | 612736 | GAMT |
| 420 | Heinz body anemia | 140700 | HBA2 |
| 421 | Hemochromatosis | 235200 | HFE |
| 422 | Hemochromatosis, type 2A | 602390 | HFE2 |
| 423 | Hemoglobin H disease, nondeletional | 613978 | HBA2 |
| 424 | Hemolytic anemia due to G6PD deficiency | 300908 | G6PD |
| 425 | Hemolytic anemia due to red cell pyruvate kinase deficiency | 266200 | PKLR |
| 426 | Hemophagocytic lymphohistiocytosis, familial, 2 | 603553 | PRF1 |
| 427 | Hemophagocytic lymphohistiocytosis, familial, 3 | 608898 | UNC13D |
| 428 | Hemophagocytic lymphohistiocytosis, familial, 4 | 603552 | STX11 |
| 429 | Hemophagocytic lymphohistiocytosis, familial, 5 | 613101 | STXBP2 |
| 430 | Hemophilia A | 306700 | F8 |
| 431 | Hemophilia B | 306900 | F9 |
| 432 | Hepatic venoocclusive disease with immunodeficiency | 235550 | SP110 |
| 433 | Hepatoencephalopathy due to combined oxidative phosphorylation deficiency type 1 | 609060 | GFM1 |
| 434 | Hereditary fructose intolerance | 229600 | ALDOB |
| 435 | Hereditary motor and sensory neuropathy VI | 601152 | MFN2 |
| 436 | Hereditary motor and sensory neuropathy, type IIc | 606071 | TRPV4 |
| 437 | Hereditary sensory and autonomic neuropathy type 4 | 256800 | NTRK1 |
| 438 | Hermansky-Pudlak syndrome 2 | 608233 | AP3B1 |
| 439 | Hermansky-pudlak syndrome 9 | 614171 | PLDN |
| 440 | Heterotaxy, visceral, 1, X-linked | 306955 | ZIC3 |
| 441 | Heterotopia, periventricular | 300049 | FLNA |
| 442 | Histidinemia | 235800 | HAMP |
| 443 | Holocarboxylase synthetase deficiency | 253270 | HLCS |
| 444 | Holoprosencephaly | 607502 | DISP1 |
| 445 | Holoprosencephaly-2 | 157170 | SIX3 |
| 446 | Holoprosencephaly-3 | 142945 | SHH |
| 447 | Holoprosencephaly-5 | 609637 | ZIC2 |
| 448 | Holoprosencephaly-7 | 610828 | PTCH1 |
| 449 | Holoprosencephaly-9 | 610829 | GLI2 |
| 450 | Homocystinuria due to MTHFR deficiency | 236250 | MTHFR |
| 451 | Hoyeraal-Hreidarsson syndrome | 300240 | DKC1 |

| | | | |
|-----|-------------------------------------------------------------------------------|--------|-----------------|
| 452 | Hyaline fibromatosis syndrome | 228600 | ANTXR2 |
| 453 | Hyperammonemia due to N-acetylglutamate synthetase deficiency | 237310 | NAGS |
| 454 | Hyper-IgE recurrent infection syndrome, autosomal recessive | 243700 | DOCK8 |
| 455 | Hyperinsulinemic hypoglycemia, familial, 1 | 256450 | ABCC8 |
| 456 | Hyperornithinemia-hyperammonemia-homocitrullinuria | 238970 | SLC25A15 |
| 457 | Hyperthyroidism, nonautoimmune | 609152 | TSHR |
| 458 | Hypochondroplasia | 146000 | FGFR3 |
| 459 | Hypoglycemia of infancy, leucine-sensitive | 240800 | ABCC8 |
| 460 | Hypogonadotropic hypogonadism 5 | 612370 | CHD7 |
| 461 | Hypogonadotropic hypogonadism 7 without anosmia | 146110 | GNRHR |
| 462 | Hypomyelination - congenital cataract | 610532 | FAM126A |
| 463 | Hypoparathyroidism - intellectual deficit - dysmorphism syndrome | 241410 | TBCE |
| 464 | Hypophosphatemic rickets | 300554 | CLCN5 |
| 465 | Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia | 218700 | PAX8 |
| 466 | Hypothyroidism, congenital, nongoitrous, 1 | 275200 | TSHR |
| 467 | Ichthyosis follicularis - alopecia - photophobia | 308205 | MBTPS2 |
| 468 | Ichthyosis, autosomal recessive 4B (harlequin) | 242500 | ABCA12 |
| 469 | Ichthyosis, congenital, autosomal recessive 1 | 242300 | TGM1 |
| 470 | Ichthyosis, congenital, autosomal recessive 4A | 601277 | ABCA12 |
| 471 | Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis | 607626 | CLDN1 |
| 472 | IMAGE syndrome | 614732 | CDKN1C |
| 473 | Immunodeficiency 10 | 612783 | STIM1 |
| 474 | Immunodeficiency 17, CD3 gamma deficient | 615607 | CD3G |
| 475 | Immunodeficiency 18, SCID variant | 615615 | CD3E |
| 476 | Immunodeficiency 19 | 615617 | CD3D |
| 477 | Immunodeficiency 27A, mycobacteriosis, AR | 209950 | IFNGR1 |
| 478 | Immunodeficiency 28, mycobacteriosis | 614889 | IFNGR2 |
| 479 | Immunodeficiency 29, mycobacteriosis | 614890 | IL12B |
| 480 | Immunodeficiency 30 | 614891 | IL12RB1 |
| 481 | Immunodeficiency 31A, mycobacteriosis, autosomal dominant | 614892 | STAT1 |
| 482 | Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive | 613796 | STAT1 |
| 483 | Immunodeficiency 31C, autosomal dominant | 614162 | STAT1 |
| 484 | Immunodeficiency 33 | 300636 | IKBKKG |
| 485 | Immunodeficiency 35 | 611521 | TYK2 |
| 486 | Immunodeficiency 9 | 612782 | ORAI1 |
| 487 | Immunodeficiency, common variable, 1 | 607594 | ICOS |
| 488 | Immunodeficiency, common variable, 3 | 613493 | CD19 |
| 489 | Immunodeficiency, X-linked | 300853 | MAGT1 |
| 490 | Immunodeficiency-centromeric instability-facial anomalies syndrome 1 | 242860 | DNMT3B |

| | | | |
|-----|--------------------------------------------------------------------|----------|------------------------------------------------|
| 491 | Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked | 304790 | FOXP3 |
| 492 | Incontinentia pigmenti, type II | 308300 | IKBKKG |
| 493 | Infantile bilateral striatal necrosis | 271930 | NUP62 |
| 494 | Infantile hypophosphatasia | 241500 | ALPL |
| 495 | Infantile neuroaxonal dystrophy 2A | 256600 | PLA2G6 |
| 496 | Infantile neuroaxonal dystrophy 2B | 610217 | PLA2G6 |
| 497 | Infantile onset spinocerebellar ataxia | 271245 | C10orf2 |
| 498 | Interleukin 1 receptor antagonist deficiency | 612852 | IL1RN |
| 499 | Isolated CoQ-cytochrome C reductase deficiency | 124000 | BCS1L |
| 500 | Isolated growth hormone deficiency type III | 307200 | BTK |
| 501 | Isolated thyroid-stimulating hormone deficiency | 275100 | TSHB |
| 502 | Isovaleric acidemia | 243500 | IVD |
| 503 | Jackson-Weiss syndrome | 123150 | FGFR2 |
| 504 | Jervell and Lange-Nielsen syndrome | 220400 | KCNQ1 |
| 505 | Jervell and Lange-Nielsen syndrome 2 | 612347 | KCNE1 |
| 506 | Jeune syndrome | 611263 | IFT80 |
| 507 | Johanson-Blizzard syndrome | 243800 | UBR1 |
| 508 | Joubert syndrome 4 | 609583 | NPHP1 |
| 509 | Joubert syndrome 6 | 610688 | TMEM67 |
| 510 | Joubert syndrome with hepatic defect | 216360 | RPGRIP1L |
| 511 | Joubert syndrome with ocular defect | 608629 | AHI1 |
| 512 | Joubert syndrome with oculorenal defect 5 | 610188 | CEP290 |
| 513 | Junctional epidermolysis bullosa - pyloric atresia | 226730 | ITGA6 - ITGB4 |
| 514 | Junctional epidermolysis bullosa, Herlitz type | 226700 | LAMA3- LAMB3 - LAMC2 |
| 515 | Junctional epidermolysis bullosa, non-Herlitz type | 226650 | ITGB4 - LAMA3- LAMB3- LAMC2 |
| 516 | Juvenile neuronal ceroid lipofuscinosis 3 | 204200 | CLN3 |
| 517 | Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome | 175050 | SMAD4 |
| 518 | Kabuki syndrome 1 | 147920 | KMT2D (MLL2) |
| 519 | Kabuki syndrome 2 | 300867 | KDM6A |
| 520 | Kahrizi syndrome | 612713 | SRD5A3 |
| 521 | Kelley-Seegmiller syndrome | 300323 | HPRT1 |
| 522 | Ketoacidosis due to beta-ketothiolase deficiency | 203750 | ACAT1 |
| 523 | King-Denborough syndrome | 145600 | RYR1 |
| 524 | Kniest dysplasia | 156550 | COL2A1 |
| 525 | Krabbe disease | 245200 - | GALC - PSAP |

| | | | |
|-----|------------------------------------------------------------|--------------------|--------------------------------------------------------------------------------------|
| | | 611722 | |
| 526 | Lacticacidemia due to PDX1 deficiency | 245349 | PDHX |
| 527 | LADD syndrome | 149730 | FGFR2 - FGFR3 |
| 528 | Late infantile neuronal ceroid lipofuscinosis | 610951 | MFSD8 |
| 529 | Late infantile neuronal ceroid lipofuscinosis 5 | 256731 | CLN5 |
| 530 | Late infantile neuronal ceroid lipofuscinosis 6 | 601780 | CLN6 |
| 531 | Late infantile neuronal ceroid lipofuscinosis 8 | 600143 | CLN8 |
| 532 | Lathosterolosis | 607330 | SC5DL |
| 533 | Legg-Calve-Perthes disease | 150600 | COL2A1 |
| 534 | Leigh syndrome | 256000 | BCS1L - DLD - NDUFAF2 - NDUFS4 - NDUFS7 - NDUFS3 - NDUFS8 |
| 535 | Leigh syndrome due to cytochrome c oxidase deficiency | 256000 | COX15 |
| 536 | Leigh syndrome due to mitochondrial COX4 deficiency | 256000 | COX10 |
| 537 | Leigh syndrome with nephrotic syndrome | 607426 - 614652 | COQ2 - PDSS2 |
| 538 | Leigh syndrome, due to COX deficiency | 256000 | SURF1 |
| 539 | Leigh syndrome, X-linked | 308930 | PDHA1 |
| 540 | LEOPARD syndrome 1 | 151100 | PTPN11 |
| 541 | LEOPARD syndrome 2 | 611554 | RAF1 |
| 542 | LEOPARD syndrome 3 | 613707 | BRAF |
| 543 | Leprechaunism | 246200 | INSR |
| 544 | Lesch-Nyhan syndrome | 300322 | HPRT1 |
| 545 | Lethal acantholytic epidermolysis bullosa | 609638 | DSP |
| 546 | Lethal ataxia with deafness and optic atrophy | 301835 | PRPS1 |
| 547 | Lethal congenital contractural syndrome 2 | 607598 | ERBB3 |
| 548 | Lethal congenital contracture syndrome 5 | 615368 | DNM2 |
| 549 | Lethal congenital contracture syndrome type 1 | 253310 | GLE1 |
| 550 | Lethal osteosclerotic bone dysplasia | 259775 | FAM20C |
| 551 | Lethal restrictive dermopathy | 275210 | LMNA - ZMPSTE24 |
| 552 | Leukemia, juvenile myelomonocytic | 607785 | PTPN11 |
| 553 | Leukocyte adhesion deficiency, type III | 612840 | FERMT3 |
| 554 | Leydig cell adenoma, somatic, with precocious puberty | 176410 | LHCGR |
| 555 | Leydig cell hypoplasia with hypergonadotropic hypogonadism | 238320 | LHCGR |
| 556 | Leydig cell hypoplasia with pseudohermaphroditism | 238320 | LHCGR |
| 557 | Lhermitte-Duclos syndrome | 158350 | PTEN |
| 558 | Limb girdle dystrophy with epidermolysis bullosa simplex | 613723 | PLEC |

| | | | |
|-----|---------------------------------------------------------------|--------|--------------------------------------|
| 559 | Lipodystrophy, congenital generalized, type 2 | 269700 | BSCL2 |
| 560 | Lissencephaly 3 | 611603 | TUBA1A |
| 561 | Lissencephaly syndrome, Norman-Roberts type | 257320 | RELN |
| 562 | Lissencephaly, X-linked | 300067 | DCX |
| 563 | Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency | 609016 | HADH |
| 564 | Long QT syndrome 1 | 192500 | KCNQ1 |
| 565 | Long QT syndrome 12 | 612955 | SNTA1 |
| 566 | Long QT syndrome 2 | 613688 | KCNH2 |
| 567 | Long QT syndrome 4 | 600919 | ANK2 |
| 568 | Long QT syndrome 5 | 613695 | KCNE1 |
| 569 | Long QT syndrome 6 | 613693 | KCNE2 |
| 570 | Long QT syndrome 9 | 611818 | CAV3 |
| 571 | Long QT syndrome-10 | 611819 | SCN4B |
| 572 | Long QT syndrome-11 | 611820 | AKAP9 |
| 573 | Luteinizing hormone resistance, female | 238320 | LHCGR |
| 574 | Lymphangiomyomatosis | 606690 | TSC1 |
| 575 | Lymphoproliferative syndrome, X-linked, 2 | 300635 | XIAP |
| 576 | Macrocephaly/autism syndrome | 605309 | PTEN |
| 577 | Macroglobulinemia, Waldenstrom | 153600 | MYD88 |
| 578 | Macular degeneration, age-related, 3 | 608895 | FBLN5 |
| 579 | Mandibuloacral dysplasia with type A lipodystrophy | 248370 | LMNA |
| 580 | Mandibuloacral dysplasia with type B lipodystrophy | 608612 | ZMPSTE24 |
| 581 | Mannosidosis, alpha-, types I and II | 248500 | MAN2B1 |
| 582 | Maple syrup urine disease | 248600 | DLD - BCKDHA - BCKDHB |
| 583 | Marfan syndrome | 154700 | FBN1 |
| 584 | Marinesco-Sjögren syndrome | 248800 | SIL1 |
| 585 | Marshall syndrome | 154780 | COL11A1 |
| 586 | Masa syndrome | 303350 | L1CAM |
| 587 | MASS syndrome | 604308 | FBN1 |
| 588 | Meacham syndrome | 608978 | WT1 |
| 589 | Meckel syndrome type 1 | 249000 | MKS1 |
| 590 | Meckel syndrome, type 5 | 611561 | RPGRIP1L |
| 591 | Medium chain acyl-CoA dehydrogenase deficiency | 201450 | ACADM |
| 592 | Medullary thyroid carcinoma | 155240 | RET |
| 593 | Megalencephalic leukoencephalopathy with subcortical cysts | 604004 | MLC1 |
| 594 | Melorheostosis with osteopoikilosis | 155950 | LEMD3 |
| 595 | Menkes disease | 309400 | ATP7A |
| 596 | Mental retardation and microcephaly and cerebellar hypoplasia | 300749 | CASK |

| | | | |
|-----|-----------------------------------------------------------|--------|-----------------|
| 597 | Mental retardation syndrome, X-linked, Siderius type | 300263 | PHF8 |
| 598 | Mental retardation, autosomal dominant type 1 | 156200 | MBD5 |
| 599 | Mental retardation, autosomal dominant type 12 | 614562 | ARID1B |
| 600 | Mental retardation, autosomal dominant type 14 | 614607 | ARID1A |
| 601 | Mental retardation, autosomal dominant type 15 | 614608 | SMARCB1 |
| 602 | Mental retardation, autosomal dominant type 16 | 614609 | SMARCA4 |
| 603 | Mental retardation, autosomal dominant type 20 | 613443 | MEF2C |
| 604 | Mental retardation, autosomal dominant type 5 | 612621 | SYNGAP1 |
| 605 | Mental retardation, autosomal dominant type 6 | 613970 | GRIN2B |
| 606 | Mental retardation, autosomal dominant type 9 | 614255 | KIF1A |
| 607 | Mental retardation, autosomal recessive 1 | 249500 | PRSS12 |
| 608 | Mental retardation, autosomal recessive 12 | 611090 | ST3GAL3 |
| 609 | Mental retardation, autosomal recessive 13 | 613192 | TRAPPC9 |
| 610 | Mental retardation, autosomal recessive 5 | 611091 | NSUN2 |
| 611 | Mental retardation, autosomal recessive 7 | 611093 | TUSC3 |
| 612 | Mental retardation, autosomal recessive, 6 | 611092 | GRIK2 |
| 613 | Mental retardation, with or without nystagmus | 300422 | CASK |
| 614 | Mental retardation, X-linked | 300034 | AGTR2 |
| 615 | Mental retardation, X-linked | 311040 | ELK1 |
| 616 | Mental retardation, X-linked | 300495 | NLGN4X |
| 617 | Mental retardation, X-linked 19 | 300844 | RPS6KA3 |
| 618 | Mental retardation, X-linked 21/34 | 300143 | IL1RAPL1 |
| 619 | Mental retardation, X-linked 30/47 | 300558 | PAK3 |
| 620 | Mental retardation, X-linked 41 | 300849 | GDI1 |
| 621 | Mental retardation, X-linked 45 | 300498 | ZNF81 |
| 622 | Mental retardation, X-linked 46 | 300436 | ARHGEF6 |
| 623 | Mental retardation, X-linked 58 | 300210 | TSPAN7 |
| 624 | Mental retardation, X-linked 63 | 300387 | ACSL4 |
| 625 | Mental retardation, X-linked 72 | 300271 | RAB39B |
| 626 | Mental retardation, X-linked 9 | 309549 | FTSJ1 |
| 627 | Mental retardation, X-linked 91 | 300577 | ZDHHC15 |
| 628 | Mental retardation, X-linked 93 | 300659 | BRWD3 |
| 629 | Mental retardation, X-linked 94 | 300699 | GRIA3 |
| 630 | Mental retardation, X-linked 96 | 300802 | SYP |
| 631 | Mental retardation, X-linked 97 | 300803 | ZNF711 |
| 632 | Mental retardation, X-linked 98 | 300912 | KIAA2022 |
| 633 | Mental retardation, X-linked syndromic 16 | 305400 | FGD1 |
| 634 | Mental retardation, X-linked syndromic 5 | 304340 | AP1S2 |
| 635 | Mental retardation, X-linked syndromic, Christianson type | 300243 | SLC9A6 |

| | | | |
|-----|----------------------------------------------------------------------------------------------------|--------------------|----------------------------------------------------------------------|
| 636 | Mental retardation, X-linked syndromic, Nascimento-type | 300860 | UBE2A |
| 637 | Mental retardation, X-linked syndromic, Raymond type | 300799 | ZDHHC9 |
| 638 | Mental retardation, X-linked syndromic, Turner type | 300706 | HUWE1 |
| 639 | Mental retardation, X-linked, Snyder-Robinson type | 309583 | SMS |
| 640 | Mental retardation, X-linked, syndromic 14 | 300676 | UPF3B |
| 641 | Mental retardation, X-linked, syndromic 15 (Cabezas type) | 300354 | CUL4B |
| 642 | Mental retardation, X-linked, syndromic, Claes-Jensen type | 300534 | KDM5C |
| 643 | Mental retardation, X-linked, syndromic, Hedera type | 300423 | ATP6AP2 |
| 644 | Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance | 300486 | OPHN1 |
| 645 | Mental retardation, X-linked, with isolated growth hormone deficiency | 300123 | SOX3 |
| 646 | Mental retardation-hypotonic facies syndrome, X-linked | 309580 | ATRX |
| 647 | Mesothelioma, somatic | 156240 | WT1 |
| 648 | Metachondromatosis | 156250 | PTPN11 |
| 649 | Metachromatic leukodystrophy | 249900 - 250100 | PSAP - ARSA |
| 650 | Metaphyseal chondrodysplasia, Murk Jansen type | 156400 | PTH1R |
| 651 | Metaphyseal dysplasia without hypotrichosis | 250460 | RMRP |
| 652 | Metatropic dysplasia | 156530 | TRPV4 |
| 653 | Methylmalonic acidemia with homocystinuria, type cb1C | 277400 | MMACHC |
| 654 | Methylmalonic acidemia with homocystinuria, type cb1D | 277410 | MMACHC |
| 655 | Mevalonic aciduria | 610377 | MVK |
| 656 | Micro syndrome | 600118 | RAB3GAP1 |
| 657 | Microcephaly 5, primary, autosomal recessive | 608716 | ASPM |
| 658 | Microphthalmia with coloboma 5 | 611638 | SHH |
| 659 | Microphthalmia, syndromic 2 | 300166 | BCOR |
| 660 | Microphthalmia, syndromic 7 | 309801 | HCCS |
| 661 | Minicore myopathy with external ophthalmoplegia | 255320 | RYR1 |
| 662 | Mitochondrial complex I deficiency | 252010 | NDUFA1 - NDUFAF2 - NDUFAF4 - NDUFS3 - NDUFS4 - NDUFV1 |
| 663 | Mitochondrial complex IV deficiency | 220110 - 603644 | COX10 - COX6B1 - FASTKD2 - SCO1 |
| 664 | Mitochondrial DNA depletion syndrome 1 (MNGIE type) | 603041 | TYMP |
| 665 | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria) | 612073 | SUCLA2 |
| 666 | Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy) | 612075 | RRM2B |
| 667 | Mitochondrial DNA depletion syndrome 8B (MNGIE type) | 612075 | RRM2B |

| | | | |
|-----|-------------------------------------------------------------------------------------|--------------------|-------------------------------|
| 668 | Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency 3 | 251880 | DGUOK |
| 669 | Mitochondrial DNA depletion syndrome, myopathic form | 609560 | TK2 |
| 670 | Mitochondrial neurogastrointestinal encephalomyopathy | 613662 | POLG |
| 671 | Mitochondrial respiratory chain complex III deficiency | 124000 | UQCRB - UQCRQ |
| 672 | Mitochondrial trifunctional protein deficiency | 609015 | HADHA - HADHB |
| 673 | Miyoshi muscular dystrophy 1 | 254130 | DYSF |
| 674 | Miyoshi muscular dystrophy 3 | 613319 | ANO5 |
| 675 | Mohr-Tranebjaerg syndrome | 304700 | TIMM8A |
| 676 | Mononeuropathy of the median nerve, mild | 613353 | SH3TC2 |
| 677 | Mowat-Wilson syndrome | 235730 | ZEB2 |
| 678 | Mucopolipidosis type 2 | 252500 | GNPTAB |
| 679 | Mucopolipidosis type 3 | 252600 | GNPTAB |
| 680 | Mucopolipidosis type 4 | 252650 | MCOLN1 |
| 681 | Mucopolysaccharidosis Ih | 607014 | IDUA |
| 682 | Mucopolysaccharidosis Ih/s | 607015 | IDUA |
| 683 | Mucopolysaccharidosis Is | 607016 | IDUA |
| 684 | Mucopolysaccharidosis type 2 | 309900 | IDS |
| 685 | Mucopolysaccharidosis type 3A (Sanfilippo syndrome type A) | 252900 | SGSH |
| 686 | Mucopolysaccharidosis type 4B | 253010 | GLB1 |
| 687 | Mucopolysaccharidosis type 6 | 253200 | ARSB |
| 688 | Mucopolysaccharidosis type 7 | 253220 | GUSB |
| 689 | Mucopolysaccharidosis type IIIB (Sanfilippo B) | 252920 | NAGLU |
| 690 | Muenke syndrome | 602849 | FGFR3 |
| 691 | MULIBREY nanism | 253250 | TRIM37 |
| 692 | Mullerian aplasia and hyperandrogenism | 158330 | WNT4 |
| 693 | Multiple endocrine neoplasia 1 | 131100 | MEN1 |
| 694 | Multiple endocrine neoplasia IIA | 171400 | RET |
| 695 | Multiple endocrine neoplasia IIB | 162300 | RET |
| 696 | Multiple endocrine neoplasia, type IV | 610755 | CDKN1B |
| 697 | Multiple epiphyseal dysplasia type 4 | 226900 | SLC26A2 |
| 698 | Multiple pterygium syndrome, lethal type | 253290 | CHRNA1 - CHRND - CHRNA3 |
| 699 | Muscle-eye-brain disease | 613153 - 613154 | FKRP - LARGE |
| 700 | Muscular dystrophy, limb-girdle, type 1A | 159000 | MYOT |
| 701 | Muscular dystrophy, limb-girdle, type 1E | 603511 | DNAJB6 |
| 702 | Muscular dystrophy, limb-girdle, type 2A | 253600 | CAPN3 |
| 703 | Muscular dystrophy, limb-girdle, type 2B | 253601 | DYSF |

| | | | |
|-----|-------------------------------------------------------|--------|--------------------------------|
| 704 | Muscular dystrophy, limb-girdle, type 2C | 253700 | SGCG |
| 705 | Muscular dystrophy, limb-girdle, type 2D | 608099 | SGCA |
| 706 | Muscular dystrophy, limb-girdle, type 2E | 604286 | SGCB |
| 707 | Muscular dystrophy, limb-girdle, type 2H | 254110 | TRIM32 |
| 708 | Muscular dystrophy, limb-girdle, type 2L | 611307 | ANO5 |
| 709 | Muscular dystrophy, limb-girdle, type IC | 607801 | CAV3 |
| 710 | Myasthenia gravis, neonatal transient | 100730 | CHRNA1 - CHRND |
| 711 | Myasthenia, limb-girdle, familial | 254300 | DOK7 |
| 712 | Myasthenic syndrome, fast-channel congenital | 608930 | CHRNA1 - CHRND |
| 713 | Myasthenic syndrome, slow-channel congenital | 601462 | CHRNA1 - CHRNB1 - CHRND |
| 714 | Myhre syndrome | 139210 | SMAD4 |
| 715 | Myopathy due to myoadenylate deaminase deficiency | 615511 | AMPD1 |
| 716 | Myopathy, actin, congenital | 161800 | ACTA1 |
| 717 | Myopathy, centronuclear | 160150 | DNM2 |
| 718 | Myopathy, congenital, with fiber-type disproportion | 255310 | TPM3 |
| 719 | Myopathy, congenital, with fiber-type disproportion 1 | 255310 | ACTA1 |
| 720 | Myopathy, distal, Tateyama type | 614321 | CAV3 |
| 721 | Myopathy, distal, with anterior tibial onset | 606768 | DYSF |
| 722 | Myopathy, myofibrillar, 3 | 609200 | MYOT |
| 723 | Myopathy, spheroid body | 182920 | MYOT |
| 724 | Myopathy, tubular aggregate, 1 | 160565 | STIM1 |
| 725 | Myopathy, tubular aggregate, 2 | 615883 | ORAI1 |
| 726 | Naegeli-Franceschetti-Jadassohn syndrome | 161000 | KRT14 |
| 727 | Nail-patella syndrome | 161200 | LMX1B |
| 728 | Nance-Horan syndrome | 302350 | NHS |
| 729 | Navajo neurohepatopathy | 256810 | MPV17 |
| 730 | Nemaline myopathy 1, autosomal dominant or recessive | 609284 | TPM3 |
| 731 | Nemaline myopathy 2 | 256030 | NEB |
| 732 | Nemaline myopathy 3, autosomal dominant or recessive | 161800 | ACTA1 |
| 733 | Nemaline myopathy 4, autosomal dominant | 609285 | TPM2 |
| 734 | Nemaline myopathy 5, Amish type | 605355 | TNNT1 |
| 735 | Neonatal adrenoleukodystrophy (gene PEX12) | 266510 | PEX12 |
| 736 | Neonatal adrenoleukodystrophy (gene PEX26) | 614873 | PEX26 |
| 737 | Neonatal adrenoleukodystrophy (gene PEX5) | 202370 | PEX5 |
| 738 | Nephrolithiasis, type I | 310468 | CLCN5 |
| 739 | Nephronophthisis 2, infantile | 602088 | INVS |
| 740 | Nephrotic syndrome, tupe 3 | 610725 | PLCE1 |

| | | | |
|-----|-----------------------------------------------------------------------|--------|-------------------------|
| 741 | Nephrotic syndrome, type 1 | 256300 | NPFS1 |
| 742 | Nephrotic syndrome, type 2 | 600995 | NPFS2 |
| 743 | Nephrotic syndrome, type 4 | 256370 | WT1 |
| 744 | Nephrotic syndrome, type 5, with or without ocular abnormalities | 614199 | LAMB2 |
| 745 | Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency | 250620 | HIBCH |
| 746 | Neurodegeneration due to cerebral folate transport deficiency | 613068 | FOLR1 |
| 747 | Neurofibromatosis, type 1 | 162200 | NF1 |
| 748 | Neurofibromatosis, type 2 | 101000 | NF2 |
| 749 | Neuromuscular disease, congenital, with uniform type 1 fiber | 117000 | RYR1 |
| 750 | Neuronal ceroid lipofuscinosis 2 | 204500 | TPP1 |
| 751 | Neuropathy, congenital hypomyelinating | 605253 | MPZ |
| 752 | Neuropathy, distal hereditary motor, type VA | 600794 | BSCL2 - GARS |
| 753 | Neutropenia, severe congenital 3, autosomal recessive | 610738 | HAX1 |
| 754 | Niemann-Pick disease type A | 257200 | SMPD1 |
| 755 | Niemann-Pick disease type B | 607616 | SMPD1 |
| 756 | Niemann-Pick disease type C1 | 257220 | NPC1 |
| 757 | Niemann-Pick disease type C2 | 607625 | NPC2 |
| 758 | Night blindness, autosomal dominant type 2 | 163500 | PDE6B |
| 759 | Nijmegen breakage syndrome | 251260 | NBN |
| 760 | Noonan syndrome 1 | 163950 | PTPN11 |
| 761 | Noonan syndrome 3 | 609942 | KRAS |
| 762 | Noonan syndrome 4 | 610733 | SOS1 |
| 763 | Noonan syndrome 5 | 611553 | RAF1 |
| 764 | Noonan syndrome 6 | 613224 | NRAS |
| 765 | Noonan syndrome 7 | 613706 | BRAF |
| 766 | Noonan syndrome-like disorder | 613563 | CBL |
| 767 | Noonan-like syndrome with loose anagen hair | 607721 | SHOC2 |
| 768 | Norrie disease | 310600 | NDP |
| 769 | Occipital horn syndrome | 304150 | ATP7A |
| 770 | Oculocerebrorenal syndrome | 309000 | OCRL |
| 771 | Omenn syndrome | 603554 | DCLRE1C |
| 772 | Omenn syndrome (gene RAG1) | 603554 | RAG1 - RAG2 |
| 773 | Opitz GBBB syndrome, type I | 300000 | MID1 |
| 774 | Ornithine transcarbamylase deficiency | 311250 | OTC |
| 775 | Osteoarthritis with mild chondrodysplasia | 604864 | COL2A1 |
| 776 | Osteochondritis dissecans | 165800 | ACAN |
| 777 | Osteogenesis imperfecta type 8 | 610915 | LEPRE1 |
| 778 | Osteogenesis imperfecta type VII | 610682 | CRTAP |
| 779 | Osteogenesis imperfecta, type I | 166200 | COL1A1 |

| | | | |
|-----|--------------------------------------------------|--------|------------------|
| 780 | Osteogenesis imperfecta, type II | 166210 | COL1A1 |
| 781 | Osteogenesis imperfecta, type III | 259420 | COL1A1 |
| 782 | Osteogenesis imperfecta, type IV | 166220 | COL1A1 |
| 783 | Osteopetrosis with renal tubular acidosis | 259730 | CA2 |
| 784 | Osteopetrosis, autosomal recessive 5 | 259720 | OSTM1 |
| 785 | Osteopoikilosis | 166700 | LEMD3 |
| 786 | Otospondylomegapiphyseal dysplasia | 215150 | COL11A2 - COL2A1 |
| 787 | Paget disease, juvenile | 239000 | TNFRSF11B |
| 788 | Pancreatic cancer, somatic | 260350 | SMAD4 |
| 789 | Panhypopituitarism, X-linked | 312000 | SOX3 |
| 790 | Pantothenate kinase-associated neurodegeneration | 234200 | PANK2 |
| 791 | Parastremmatic dwarfism | 168400 | TRPV4 |
| 792 | Partial androgen insensitivity syndrome | 312300 | AR |
| 793 | PCWH syndrome | 609136 | SOX10 |
| 794 | Pelizaesus-Merzbacher-like due to GJC2 mutation | 608804 | GJC2 |
| 795 | Peroxisomal acyl-CoA oxidase deficiency | 264470 | ACOX1 |
| 796 | Peroxisome biogenesis disorder 11A (Zellweger) | 614883 | PEX13 |
| 797 | Peroxisome biogenesis disorder 11B | 614885 | PEX13 |
| 798 | Peroxisome biogenesis disorder 13A (Zellweger) | 614887 | PEX14 |
| 799 | Peroxisome biogenesis disorder 4A (Zellweger) | 614862 | PEX6 |
| 800 | Peroxisome biogenesis disorder 4B | 614863 | PEX6 |
| 801 | Peroxisome biogenesis disorder 5A (Zellweger) | 614866 | PEX2 |
| 802 | Peroxisome biogenesis disorder 5B | 614867 | PEX2 |
| 803 | Peroxisome biogenesis disorder 6A (Zellweger) | 614870 | PEX10 |
| 804 | Peroxisome biogenesis disorder 6B | 614871 | PEX10 |
| 805 | Perrault syndrome | 233400 | HSD17B4 |
| 806 | Phenylketonuria | 261600 | PAH |
| 807 | Pheochromocytoma | 171300 | KIF1B - RET -VHL |
| 808 | Phosphoglycerate kinase 1 deficiency | 300653 | PGK1 |
| 809 | Pierson syndrome | 609049 | LAMB2 |
| 810 | Pitt-Hopkins syndrome | 610954 | TCF4 |
| 811 | Plasminogen deficiency type 1 | 217090 | PLG |
| 812 | Platyspondylic skeletal dysplasia, Torrance type | 151210 | COL2A1 |
| 813 | Polycystic kidney disease 2 | 613095 | PKD2 |
| 814 | Polyposis, juvenile intestinal | 174900 | SMAD4 |
| 815 | Porphyria, congenital erythropoietic | 263700 | UROS |
| 816 | Precocious puberty, male | 176410 | LHCGR |
| 817 | Primary lateral sclerosis, juvenile | 606353 | ALS2 |

| | | | |
|-----|-------------------------------------------------------------------------|--------|-----------------------------------------|
| 818 | Progressive epilepsy - intellectual deficit, Finnish type | 610003 | CLN8 |
| 819 | Properdin deficiency, X-linked | 312060 | CFP |
| 820 | Propionic acidemia (gene PCCA) | 606054 | PCCA - PCCB |
| 821 | Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis | 308990 | CLCN5 |
| 822 | Proximal spinal muscular atrophy type 1 | 253300 | SMN1 |
| 823 | Proximal spinal muscular atrophy type 2 | 253550 | SMN1 |
| 824 | Proximal spinal muscular atrophy type 3 | 253400 | SMN1 |
| 825 | Proximal spinal muscular atrophy type 4 | 271150 | SMN1 |
| 826 | Pseudoachondroplasia | 177170 | COMP |
| 827 | Pseudohermaphroditism, male, with gynecomastia | 264300 | HSD17B3 |
| 828 | Pseudohypoaldosteronism type 1, autosomal recessive | 264350 | SCNN1A - SCNN1B - SCNN1G |
| 829 | Pseudovaginal perineoscrotal hypospadias | 264600 | SRD5A2 |
| 830 | Pycnodysostosis | 265800 | CTSK |
| 831 | Pyogenic bacterial infections, recurrent, due to MYD88 deficiency | 612260 | MYD88 |
| 832 | Pyridoxal phosphate-responsive seizures | 610090 | PNPO |
| 833 | Pyruvate carboxylase deficiency | 266150 | PC |
| 834 | Pyruvate dehydrogenase phosphatase deficiency | 608782 | PDP1 |
| 835 | Renal-hepatic-pancreatic dysplasia | 208540 | NPHP3 |
| 836 | Renpenning syndrome | 309500 | PQBP1 |
| 837 | Retinitis pigmentosa type 1, autosomal dominant | 180100 | RP1 |
| 838 | Retinitis pigmentosa type 10, autosomal dominant | 180105 | IMPDH1 |
| 839 | Retinitis pigmentosa type 11, autosomal dominant | 600138 | PRPF31 |
| 840 | Retinitis pigmentosa type 13, autosomal dominant | 600059 | PRPF8 |
| 841 | Retinitis pigmentosa type 17, autosomal dominant | 600852 | CA4 |
| 842 | Retinitis pigmentosa type 18, autosomal dominant | 601414 | PRPF3 |
| 843 | Retinitis pigmentosa type 19, autosomal dominant | 601718 | ABCA4 |
| 844 | Retinitis pigmentosa type 27, autosomal dominant | 613750 | NRL |
| 845 | Retinitis pigmentosa type 30, autosomal dominant | 607921 | FSCN2 |
| 846 | Retinitis pigmentosa type 31, autosomal dominant | 609923 | TOPORS |
| 847 | Retinitis pigmentosa type 33, autosomal dominant | 610359 | SNRNP200 |
| 848 | Retinitis pigmentosa type 35, autosomal dominant | 610282 | SEMA4A |
| 849 | Retinitis pigmentosa type 4, autosomal dominant | 613731 | RHO |
| 850 | Retinitis pigmentosa type 42, autosomal dominant | 612943 | KLHL7 |
| 851 | Retinitis pigmentosa type 48, autosomal dominant | 613827 | GUCA1B |
| 852 | Retinitis pigmentosa type 50, autosomal dominant | 613194 | BEST1 |
| 853 | Retinitis pigmentosa type 7, autosomal dominant | 608133 | PRPH2 |
| 854 | Retinitis pigmentosa type 9, autosomal dominant | 180104 | RP9 |
| 855 | Rett syndrome, congenital variant | 613454 | FOXP1 |

| | | | |
|-----|---------------------------------------------------------------------------------------------------------------|--------|-----------------------|
| 856 | Rhabdomyosarcoma 2, alveolar | 268220 | PAX3 |
| 857 | Rhizomelic chondrodysplasia punctata type 1 | 215100 | PEX7 |
| 858 | Rhizomelic chondrodysplasia punctata type 3 | 600121 | AGPS |
| 859 | Rippling muscle disease | 606072 | CAV3 |
| 860 | Roberts syndrome | 269000 | ESCO2 |
| 861 | Robinow-Sorauf syndrome | 180750 | TWIST1 |
| 862 | Rolandic epilepsy, mental retardation, and speech dyspraxia | 300643 | SRPX2 |
| 863 | Roussy-Levy syndrome | 180800 | MPZ - PMP22 |
| 864 | Saethre-Chotzen syndrome | 101400 | FGFR2 - TWIST1 |
| 865 | Sandhoff disease | 268800 | HEXB |
| 866 | Sanfilippo syndrome type C | 252930 | HGSNAT |
| 867 | Scaphocephaly, maxillary retrusion, and mental retardation | 609579 | FGFR2 |
| 868 | Scapuloperoneal spinal muscular atrophy | 181405 | TRPV4 |
| 869 | Schizencephaly | 269160 | SHH - SIX3 |
| 870 | Schneckenbecken dysplasia | 269250 | SLC35D1 |
| 871 | Schwannomatosis | 162091 | NF2 |
| 872 | Schwartz-Jampel syndrome | 255800 | HSPG2 |
| 873 | Seckel syndrome | 210600 | ATR |
| 874 | SED congenita | 183900 | COL2A1 |
| 875 | SED, Maroteaux type | 184095 | TRPV4 |
| 876 | Senior-Loken syndrome | 606996 | NPHP4 |
| 877 | Senior-Loken syndrome | 610189 | CEP290 |
| 878 | Senior-Loken syndrome 1 | 266900 | NPHP1 |
| 879 | Senior-Loken syndrome 5 | 609254 | IQCB1 |
| 880 | Sensory ataxic neuropathy - dysarthria - ophthalmoparesis | 607459 | POLG |
| 881 | SERKAL syndrome | 611812 | WNT4 |
| 882 | Severe combined immunodeficiency due to complete RAG1/2 deficiency | 601457 | RAG1 - RAG2 |
| 883 | Severe combined immunodeficiency due to DCLRE1C deficiency | 602450 | DCLRE1C |
| 884 | Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation | 611291 | NHEJ1 |
| 885 | Severe combined immunodeficiency with sensitivity to ionizing radiation | 602450 | LIG4 |
| 886 | Severe combined immunodeficiency. deaminase deficiency | 102700 | ADA |
| 887 | Severe generalized recessive dystrophic epidermolysis bullosa | 226600 | COL7A1 |
| 888 | Severe neonatal-onset encephalopathy with microcephaly | 300673 | MECP2 |
| 889 | Severe T-cell immunodeficiency - congenital alopecia - nail dystrophy | 601705 | FOXN1 |
| 890 | Short QT syndrome 1 | 609620 | KCNH2 |
| 891 | Short QT syndrome 2 | 609621 | KCNQ1 |
| 892 | Short QT syndrome 3 | 609622 | KCNJ2 |
| 893 | Short-rib thoracic dysplasia 3 with or without polydactyly | 613091 | DYNC2H1 |

| | | | |
|-----|-------------------------------------------------------------------------|--------|--------------------------|
| 894 | Shwachman-Diamond syndrome | 260400 | SBDS |
| 895 | Sialidosis, type I | 256550 | NEU1 |
| 896 | Sialidosis, type II | 256550 | NEU1 |
| 897 | Sickle cell anemia | 603903 | HBB |
| 898 | Silver spastic paraplegia syndrome | 270685 | BSCL2 |
| 899 | Silver-Russell syndrome | 180860 | H19 |
| 900 | Simpson-Golabi-Behmel syndrome type 2 | 300209 | OFD1 |
| 901 | Simpson-Golabi-Behmel syndrome, type 1 | 312870 | GPC3 |
| 902 | Síndrome de Dursun | 612541 | G6PC3 |
| 903 | Single median maxillary central incisor | 147250 | SHH |
| 904 | Sjogren-Larsson syndrome | 270200 | ALDH3A2 |
| 905 | Slowed nerve conduction velocity, AD | 608236 | ARHGEF10 |
| 906 | SMED Strudwick type | 184250 | COL2A1 |
| 907 | Smith-Lemli-Opitz syndrome | 270400 | DHCR7 |
| 908 | Sotos syndrome 1 | 117550 | NSD1 |
| 909 | Spastic paralysis, infantile onset ascending | 607225 | ALS2 |
| 910 | Spastic paraplegia type 2, X-linked | 312920 | PLP1 |
| 911 | Spherocytosis, type 1 | 182900 | ANK1 |
| 912 | Spinal muscular atrophy with respiratory distress | 604320 | IGHMBP2 |
| 913 | Spinal muscular atrophy, distal, congenital nonprogressive | 600175 | TRPV4 |
| 914 | Spondyloepimetaphyseal dysplasia | 612813 | ACAN |
| 915 | Spondyloepiphyseal dysplasia | 608361 | ACAN |
| 916 | Spondylometaphyseal dysplasia, Kozlowski type | 184252 | TRPV4 |
| 917 | Spondyloperipheral dysplasia | 271700 | COL2A1 |
| 918 | Stickler syndrome, type I, nonsyndromic ocular | 609508 | COL2A1 |
| 919 | Stickler syndrome, type I | 108300 | COL2A1 |
| 920 | Stickler syndrome, type II | 604841 | COL11A1 |
| 921 | Stickler syndrome, type III | 184840 | COL11A2 |
| 922 | Stickler syndrome, type IV | 614134 | COL9A1 |
| 923 | Stiff skin syndrome | 184900 | FBN1 |
| 924 | Stocco dos Santos X-linked mental retardation syndrome | 300434 | SHROOM4 |
| 925 | Stormorken syndrome | 185070 | STIM1 |
| 926 | Stüve-Wiedemann syndrome | 601559 | LIFR |
| 927 | Subcortical laminar heteropia, X-linked | 300067 | DCX |
| 928 | Succinyl CoA:3-oxoacid CoA transferase deficiency | 245050 | OXCT1 |
| 929 | Sudden infant death with dysgenesis of the testes syndrome | 608800 | TSPYL1 |
| 930 | Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A | 252150 | MOCS1 - MOCS2 |
| 931 | Sulfocysteinuria | 272300 | SUOX |
| 932 | Surfactant metabolism dysfunction, pulmonary, 1 | 265120 | SFTPB |

| | | | |
|-----|---------------------------------------------------------------------|--------|-----------------------------------------|
| 933 | Surfactant metabolism dysfunction, pulmonary, 2 | 610913 | SFTPC |
| 934 | Surfactant metabolism dysfunction, pulmonary, 3 | 610921 | ABCA3 |
| 935 | Syndromic microphthalmia type 9 | 601186 | STRA6 |
| 936 | Tay-Sachs disease | 272800 | HEXA |
| 937 | T-B+ severe combined immunodeficiency due to gamma chain deficiency | 300400 | IL2RG |
| 938 | T-B+ severe combined immunodeficiency due to JAK3 deficiency | 600802 | JAK3 |
| 939 | T-B+ severe combined immunodeficiency, X-linked | 312863 | IL2RG |
| 940 | Telangiectasia, hereditary hemorrhagic, type 1 | 187300 | ENG |
| 941 | Telangiectasia, hereditary hemorrhagic, type 2 | 600376 | ACVRL1 |
| 942 | Tetra-amelia, autosomal recessive | 273395 | WNT3 |
| 943 | Thanatophoric dysplasia, type I | 187600 | FGFR3 |
| 944 | Thanatophoric dysplasia, type II | 187601 | FGFR3 |
| 945 | Thrombocythemia 2 | 601977 | MPL |
| 946 | Thrombocytopenia, congenital amegakaryocytic | 604498 | MPL |
| 947 | Thrombocytopenia-absent radius syndrome | 274000 | RBM8A |
| 948 | Thrombotic thrombocytopenic purpura, familial | 274150 | ADAMTS13 |
| 949 | Thyroid dysmorphogenesis 6 | 607200 | DUOX2 |
| 950 | Thyroid dysmorphogenesis 1 | 274400 | SLC5A5 |
| 951 | Thyroid dysmorphogenesis 2A | 274500 | TPO |
| 952 | Thyroid dysmorphogenesis 3 | 274700 | TG |
| 953 | Thyroxine-binding globulin deficiency | 314200 | SERPINA7 |
| 954 | Tietz albinism-deafness syndrome | 103500 | MITF |
| 955 | Tooth agenesis, selective, X-linked 1 | 313500 | EDA |
| 956 | Treacher Collins syndrome 1 | 154500 | TCOF1 |
| 957 | Treacher Collins syndrome 3 | 248390 | POLR1C |
| 958 | Trichothiodystrophy, complementation group A | 601675 | GTF2H5 |
| 959 | Tuberous sclerosis-1 | 191100 | TSC1 |
| 960 | Tuberous sclerosis-2 | 613254 | TSC2 |
| 961 | Tyrosinemia type 1 | 276700 | FAH |
| 962 | Tyrosinemia type 2 | 276600 | TAT |
| 963 | Tyrosinemia type 3 | 276710 | HPD |
| 964 | Ullrich congenital muscular dystrophy | 254090 | COL6A1 - COL6A2 - COL6A3 |
| 965 | Unverricht-Lundborg disease | 254800 | CSTB |
| 966 | Usher syndrome type 1 | 276900 | MYO7A |
| 967 | Usher syndrome type 1C | 276904 | USH1C |
| 968 | Usher syndrome type 1G | 606943 | USH1G |
| 969 | Usher syndrome type 2A | 276901 | USH2A |
| 970 | Usher syndrome type 2C | 605472 | GPR98 |

| | | | |
|------|-------------------------------------------------------------------------|--------|-----------------|
| 971 | Usher syndrome type 3A | 276902 | CLRN1 |
| 972 | Very long chain acyl-CoA dehydrogenase deficiency | 201475 | ACADVL |
| 973 | Vitamin B12-responsive methylmalonic acidemia type cblA | 251100 | MMAA |
| 974 | Vitamin B12-responsive methylmalonic acidemia type cblB | 251110 | MMAB |
| 975 | Vitamin B12-unresponsive methylmalonic acidemia type mut- | 251000 | MUT |
| 976 | Vitamin D-dependent rickets type 2A | 277440 | VDR |
| 977 | Vitamin D-dependent rickets, type I | 264700 | CYP27B1 |
| 978 | von Hippel-Lindau syndrome | 193300 | VHL |
| 979 | von Willebrand disease, type 1 | 193400 | VWF |
| 980 | von Willebrand disease, types 2A, 2B, 2M, and 2N | 613554 | VWF |
| 981 | von Willibrand disease, type 3 | 277480 | VWF |
| 982 | Waardenburg syndrome, type 1 | 193500 | PAX3 |
| 983 | Waardenburg syndrome, type 2A | 193510 | MITF |
| 984 | Waardenburg syndrome, type 2E, with or without neurologic involvement | 611584 | SOX10 |
| 985 | Waardenburg syndrome, type 3 | 148820 | PAX3 |
| 986 | Waardenburg syndrome, type 4C | 613266 | SOX10 |
| 987 | Waardenburg syndrome/ocular albinism, digenic | 103470 | MITF |
| 988 | Waardenburg-Shah syndrome 4A | 277580 | EDNRB |
| 989 | Waardenburg-Shah syndrome 4B | 613265 | EDN3 |
| 990 | Walker-Warburg syndrome (gene POMGNT1) | 253280 | POMGNT1 |
| 991 | Walker-Warburg syndrome (gene POMT1) | 236670 | POMT1 |
| 992 | Walker-Warburg syndrome (gene POMT2) | 613150 | POMT2 |
| 993 | Weill-Marchesani syndrome 2, dominant | 608328 | FBN1 |
| 994 | Weissenbacher-Zweymuller syndrome | 277610 | COL11A2 |
| 995 | Wilms tumor 2 | 194071 | H19 |
| 996 | Wilms tumor, type 1 | 194070 | WT1 |
| 997 | Wilson disease | 277900 | ATP7B |
| 998 | Wiskott-Aldrich syndrome | 301000 | WAS |
| 999 | Wolcott-Rallison syndrome | 226980 | EIF2AK3 |
| 1000 | Wrinkly skin syndrome | 278250 | ATP6V0A2 |
| 1001 | Xeroderma pigmentosum complementation group A | 278700 | XPA |
| 1002 | Xeroderma pigmentosum complementation group E | 278740 | DDB2 |
| 1003 | Xeroderma pigmentosum, group C | 278720 | XPC |
| 1004 | Xeroderma pigmentosum/Cockayne syndrome complex complementation group B | 610651 | ERCC3 |
| 1005 | Xeroderma pigmentosum/Cockayne syndrome complex complementation group D | 278730 | ERCC2 |
| 1006 | Xeroderma pigmentosum/Cockayne syndrome complex complementation group F | 278760 | ERCC4 |
| 1007 | Xeroderma pigmentosum/Cockayne syndrome complex complementation group G | 278780 | ERCC5 |

| | | | |
|------|------------------------------------------------------|--------|---------------|
| 1008 | X-linked agammaglobulinemia | 300755 | BTK |
| 1009 | X-linked centronuclear myopathy | 310400 | MTM1 |
| 1010 | X-linked Charcot-Marie-Tooth disease type 5 | 311070 | PRPS1 |
| 1011 | X-linked creatine transporter deficiency | 300352 | SLC6A8 |
| 1012 | X-linked distal spinal muscular atrophy | 300489 | ATP7A |
| 1013 | X-linked hyper-IgM syndrome | 308230 | CD40LG |
| 1014 | X-linked intellectual deficit with marfanoid habitus | 309520 | MED12 |
| 1015 | X-linked lymphoproliferative disease | 308240 | SH2D1A |
| 1016 | X-linked severe congenital neutropenia | 300299 | WNT10A |
| 1017 | X-linked spinal muscular atrophy type 2 | 301830 | UBA1 |
| 1018 | Yunis-Varon syndrome | 216340 | FIG4 |
| 1019 | Zellweger syndrome 1A | 214100 | PEX1 |
| 1020 | Zellweger syndrome 7A | 614872 | PEX26 |
| | | | |