

| Gen      | OMIM-Gen # | OMIM-Phänotypen                                               | OMIM-Phänotypen # | Erbmodi |
|----------|------------|---------------------------------------------------------------|-------------------|---------|
| ACE      | 106180     | {Stroke, hemorrhagic}                                         | 614519            |         |
|          |            | Renal tubular dysgenesis                                      | 267430            | AR      |
|          |            | {Myocardial infarction, susceptibility to}                    |                   |         |
|          |            | {Microvascular complications of diabetes 3}                   | 612624            |         |
| ACTA1    | 102610     | [Angiotensin I-converting enzyme, benign serum increase]      |                   |         |
|          |            | {SARS, progression of}                                        |                   |         |
|          |            | ?Myopathy, scapulohumeroperoneal                              | 616852            | AD      |
|          |            | Nemaline myopathy 3, autosomal dominant or recessive          | 161800            | AR, AD  |
|          |            | Myopathy, actin, congenital, with excess of thin myofilaments | 161800            | AR, AD  |
| ACTA2    | 102620     | Myopathy, actin, congenital, with cores                       | 161800            | AR, AD  |
|          |            | Myopathy, congenital, with fiber-type disproportion 1         | 255310            | AR, AD  |
|          |            | Multisystemic smooth muscle dysfunction syndrome              | 613834            | AD      |
|          |            | Aortic aneurysm, familial thoracic 6                          | 611788            | AD      |
|          |            | Moyamoya disease 5                                            | 614042            |         |
| ACTN4    | 604638     | Glomerulosclerosis, focal segmental, 1                        | 603278            | AD      |
| ADAMTS13 | 604134     | Thrombotic thrombocytopenic purpura, hereditary               | 274150            | AR      |
| AGT      | 106150     | Renal tubular dysgenesis                                      | 267430            | AR      |
|          |            | {Preeclampsia, susceptibility to}                             |                   |         |
| AGTR1    | 106165     | {Hypertension, essential, susceptibility to}                  | 145500            | Mu      |
|          |            | {Hypertension, essential}                                     | 145500            | Mu      |
|          |            | Renal tubular dysgenesis                                      | 267430            | AR      |
| AGXT     | 604285     | Hyperoxaluria, primary, type 1                                | 259900            | AR      |
| AIP      | 605555     | Pituitary adenoma 1, multiple types                           | 102200            | AD, Som |
|          |            | Pituitary adenoma predisposition                              | 102200            | AD, Som |
| ALAD     | 125270     | Porphyria, acute hepatic                                      | 612740            | AR      |
|          |            | {Lead poisoning, susceptibility to}                           | 612740            | AR      |
| ALPL     | 171760     | Odontohypophosphatasia                                        | 146300            | AR, AD  |
|          |            | Hypophosphatasia, infantile                                   | 241500            | AR      |
|          |            | Hypophosphatasia, childhood                                   | 241510            | AR      |
|          |            | Hypophosphatasia, adult                                       | 146300            | AR, AD  |
| ALS2     | 606352     | Primary lateral sclerosis, juvenile                           | 606353            | AR      |
|          |            | Spastic paralysis, infantile onset ascending                  | 607225            | AR      |
|          |            | Amyotrophic lateral sclerosis 2, juvenile                     | 205100            | AR      |
| AP2S1    | 602242     | Hypocalciuric hypercalcemia, type III                         | 600740            | AD      |
| AQP2     | 107777     | Diabetes insipidus, nephrogenic, 2                            | 125800            | AR, AD  |
| ARHGDI1A | 601925     | Nephrotic syndrome, type 8                                    | 615244            | AR      |
| ARID1A   | 603024     | Coffin-Siris syndrome 2                                       | 614607            | AD      |
| ARID1B   | 614556     | Coffin-Siris syndrome 1                                       | 135900            | AD      |
| ASPM     | 605481     | Microcephaly 5, primary, autosomal recessive                  | 608716            | AR      |

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| ATR      | 601215 | Seckel syndrome 1                                                                           | 210600 | AR     |
|          |        | ?Cutaneous telangiectasia and cancer syndrome, familial                                     | 614564 | AD     |
| AVP      | 192340 | Diabetes insipidus, neurohypophyseal                                                        | 125700 | AD     |
| AVPR2    | 300538 | Diabetes insipidus, nephrogenic, 1                                                          | 304800 | XLR    |
|          |        | Nephrogenic syndrome of inappropriate antidiuresis                                          | 300539 | XLR    |
| B3GALNT2 | 610194 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11  | 615181 | AR     |
| B4GAT1   | 605517 | Muscular dystrophy-dystroglycanopathy [congenital with brain and eye anomalies], type A, 13 | 615287 | AR     |
| BBS10    | 610148 | Bardet-Biedl syndrome 10                                                                    | 615987 | AR     |
| BICD2    | 609797 | Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant                | 618291 | AD     |
|          |        | Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant                | 615290 | AD     |
| BMP1     | 112264 | Osteogenesis imperfecta, type XIII                                                          | 614856 | AR     |
| BRAT1    | 614506 | Neurodevelopmental disorder with cerebellar atrophy and with or without seizures            | 618056 | AR     |
|          |        | Rigidity and multifocal seizure syndrome, lethal neonatal                                   | 614498 | AR     |
| BSND     | 606412 | Sensorineural deafness with mild renal dysfunction                                          | 602522 | AR     |
|          |        | Bartter syndrome, type 4a                                                                   | 602522 | AR     |
| BUB1B    | 602860 | Colorectal cancer, somatic                                                                  | 114500 |        |
|          |        | [Premature chromatid separation trait]                                                      | 176430 | AD     |
|          |        | Mosaic variegated aneuploidy syndrome 1                                                     | 257300 | AR     |
| C3       | 120700 | C3 deficiency                                                                               | 613779 | AR     |
|          |        | {Hemolytic uremic syndrome, atypical, susceptibility to, 5}                                 | 612925 | AD     |
|          |        | {Macular degeneration, age-related, 9}                                                      | 611378 |        |
| CAPN3    | 114240 | Muscular dystrophy, limb-girdle, autosomal recessive 1                                      | 253600 | AR     |
|          |        | Muscular dystrophy, limb-girdle, autosomal dominant 4                                       | 618129 | AD     |
| CASK     | 300172 | Mental retardation, with or without nystagmus                                               | 300422 |        |
|          |        | Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia | 300749 | XLD    |
|          |        | FG syndrome 4                                                                               | 300422 |        |
| CASR     | 601199 | Hypocalcemia, autosomal dominant, with Bartter syndrome                                     | 601198 | AD     |
|          |        | {Epilepsy idiopathic generalized, susceptibility to, 8}                                     | 612899 |        |
|          |        | Hyperparathyroidism, neonatal                                                               | 239200 | AR, AD |
|          |        | Hypocalcemia, autosomal dominant                                                            | 601198 | AD     |
|          |        | Hypocalciuric hypercalcemia, type I                                                         | 145980 | AD     |
| CAV3     | 601253 | Myopathy, distal, Tateyama type                                                             | 614321 | AD     |
|          |        | Creatine phosphokinase, elevated serum                                                      | 123320 | AD     |
|          |        | Cardiomyopathy, familial hypertrophic                                                       | 192600 | DD, AD |
|          |        | Rippling muscle disease 2                                                                   | 606072 | AD     |
|          |        | Long QT syndrome 9                                                                          | 611818 | AD     |
| CBS      | 613381 | Thrombosis, hyperhomocysteinemic                                                            | 236200 | AR     |
|          |        | Homocystinuria, B6-responsive and nonresponsive types                                       | 236200 | AR     |
| CD46     | 120920 | {Hemolytic uremic syndrome, atypical, susceptibility to, 2}                                 | 612922 | AR, AD |
| CDK5RAP2 | 608201 | Microcephaly 3, primary, autosomal recessive                                                | 604804 | AR     |

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| CDKN1B | 600778 | Multiple endocrine neoplasia, type IV                                                   | 610755 | AD     |
| CENPJ  | 609279 | Microcephaly 6, primary, autosomal recessive                                            | 608393 | AR     |
|        |        | ?Seckel syndrome 4                                                                      | 613676 | AR     |
| CEP135 | 611423 | Microcephaly 8, primary, autosomal recessive                                            | 614673 | AR     |
| CEP152 | 613529 | Microcephaly 9, primary, autosomal recessive                                            | 614852 | AR     |
|        |        | Seckel syndrome 5                                                                       | 613823 | AR     |
| CEP63  | 614724 | ?Seckel syndrome 6                                                                      | 614728 | AR     |
| CFB    | 138470 | ?Complement factor B deficiency                                                         | 615561 | AR     |
|        |        | {Hemolytic uremic syndrome, atypical, susceptibility to, 4}                             | 612924 | AD     |
|        |        | {Macular degeneration, age-related, 14, reduced risk of}                                | 615489 | DD     |
| CFH    | 134370 | {Macular degeneration, age-related, 4}                                                  | 610698 |        |
|        |        | Basal laminar drusen                                                                    | 126700 | AD     |
|        |        | Complement factor H deficiency                                                          | 609814 | AR, AD |
|        |        | {Hemolytic uremic syndrome, atypical, susceptibility to, 1}                             | 235400 | AR, AD |
| CFI    | 217030 | {Hemolytic uremic syndrome, atypical, susceptibility to, 3}                             | 612923 | AD     |
|        |        | {Macular degeneration, age-related, 13, susceptibility to}                              | 615439 | AD     |
|        |        | Complement factor I deficiency                                                          | 610984 | AR     |
| CFTR   | 602421 | Cystic fibrosis                                                                         | 219700 | AR     |
|        |        | Sweat chloride elevation without CF                                                     |        |        |
|        |        | Congenital bilateral absence of vas deferens                                            | 277180 | AR     |
|        |        | {Pancreatitis, hereditary}                                                              | 167800 | AD     |
|        |        | {Bronchiectasis with or without elevated sweat chloride 1, modifier of}                 | 211400 | AD     |
|        |        | {Hypertrypsinemia, neonatal}                                                            |        |        |
| CHRNA1 | 100690 | Myasthenic syndrome, congenital, 1B, fast-channel                                       | 608930 | AR, AD |
|        |        | Myasthenic syndrome, congenital, 1A, slow-channel                                       | 601462 | AD     |
|        |        | Multiple pterygium syndrome, lethal type                                                | 253290 | AR     |
| CHRNA1 | 100720 | ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency | 616323 | AR     |
|        |        | Multiple pterygium syndrome, lethal type                                                | 253290 | AR     |
|        |        | Myasthenic syndrome, congenital, 3B, fast-channel                                       | 616322 | AR     |
|        |        | ?Myasthenic syndrome, congenital, 3A, slow-channel                                      | 616321 | AD     |
| CHRNA1 | 100725 | Myasthenic syndrome, congenital, 4A, slow-channel                                       | 605809 | AR, AD |
|        |        | Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency  | 608931 | AR     |
|        |        | Myasthenic syndrome, congenital, 4B, fast-channel                                       | 616324 | AR     |
| CHRNA1 | 100730 | Multiple pterygium syndrome, lethal type                                                | 253290 | AR     |
|        |        | Escobar syndrome                                                                        | 265000 | AR     |
| CLCN2  | 600570 | Leukoencephalopathy with ataxia                                                         | 615651 | AR     |
|        |        | Hyperaldosteronism, familial, type II                                                   | 605635 | AD     |
|        |        | {Epilepsy, juvenile myoclonic, susceptibility to, 8}                                    | 607628 | AD     |
|        |        | {Epilepsy, juvenile absence, susceptibility to, 2}                                      | 607628 | AD     |
|        |        | {Epilepsy, idiopathic generalized, susceptibility to, 11}                               | 607628 | AD     |

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| CLCN5  | 300008 | Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis | 308990 | XLR    |
|        |        | Hypophosphatemic rickets                                                | 300554 | XLR    |
|        |        | Dent disease 1                                                          | 300009 | XLR    |
|        |        | Nephrolithiasis, type I                                                 | 310468 | XLR    |
| CLCNKA | 602024 | Bartter syndrome, type 4b, digenic                                      | 613090 | DR     |
| CLCNKB | 602023 | Bartter syndrome, type 3                                                | 607364 | AR     |
|        |        | Bartter syndrome, type 4b, digenic                                      | 613090 | DR     |
| CLDN16 | 603959 | Hypomagnesemia 3, renal                                                 | 248250 | AR     |
| CLDN19 | 610036 | Hypomagnesemia 5, renal, with ocular involvement                        | 248190 | AR     |
| COL1A1 | 120150 | Osteogenesis imperfecta, type II                                        | 166210 | AD     |
|        |        | Caffey disease                                                          | 114000 | AD     |
|        |        | Ehlers-Danlos syndrome, arthrochalasia type, 1                          | 130060 | AD     |
|        |        | Osteogenesis imperfecta, type I                                         | 166200 | AD     |
|        |        | {Bone mineral density variation QTL, osteoporosis}                      | 166710 | AD     |
|        |        | Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1           | 619115 | AD     |
|        |        | Osteogenesis imperfecta, type IV                                        | 166220 | AD     |
|        |        | Osteogenesis imperfecta, type III                                       | 259420 | AD     |
|        |        | Osteogenesis imperfecta, type III                                       | 259420 | AD     |
|        |        | {Osteoporosis, postmenopausal}                                          | 166710 | AD     |
|        |        | Ehlers-Danlos syndrome, arthrochalasia type, 2                          | 617821 | AD     |
|        |        | Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2           | 619120 | AD     |
| COL1A2 | 120160 | Ehlers-Danlos syndrome, cardiac valvular type                           | 225320 | AR     |
|        |        | Osteogenesis imperfecta, type IV                                        | 166220 | AD     |
|        |        | Osteogenesis imperfecta, type II                                        | 166210 | AD     |
|        |        | Osteogenesis imperfecta, type II                                        | 166210 | AD     |
| COL3A1 | 120180 | Ehlers-Danlos syndrome, vascular type                                   | 130050 | AD     |
|        |        | Polymicrogyria with or without vascular-type EDS                        | 618343 | AR     |
| COL4A3 | 120070 | Hematuria, benign familial                                              | 141200 | AD     |
|        |        | Alport syndrome 3, autosomal dominant                                   | 104200 | AD     |
|        |        | Alport syndrome 2, autosomal recessive                                  | 203780 | AR     |
| COL4A4 | 120131 | Hematuria, familial benign                                              | 141200 | AD     |
|        |        | Alport syndrome 2, autosomal recessive                                  | 203780 | AR     |
|        |        | Alport syndrome 2, autosomal recessive                                  | 203780 | AR     |
| COL4A5 | 303630 | Alport syndrome 1, X-linked                                             | 301050 | XLD    |
| COL6A1 | 120220 | Bethlem myopathy 1                                                      | 158810 | AR, AD |
|        |        | Ullrich congenital muscular dystrophy 1                                 | 254090 | AR, AD |
| COL6A2 | 120240 | Bethlem myopathy 1                                                      | 158810 | AR, AD |
|        |        | ?Myosclerosis, congenital                                               | 255600 | AR     |
|        |        | Ullrich congenital muscular dystrophy 1                                 | 254090 | AR, AD |
|        |        | Ullrich congenital muscular dystrophy 1                                 | 254090 | AR, AD |
| COL6A3 | 120250 | Dystonia 27                                                             | 616411 | AR     |
|        |        | Bethlem myopathy 1                                                      | 158810 | AR, AD |

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| COLQ    | 603033 | Myasthenic syndrome, congenital, 5                                                          | 603034 | AR     |
| COQ2    | 609825 | {Multiple system atrophy, susceptibility to}                                                | 146500 | AR, AD |
|         |        | Coenzyme Q10 deficiency, primary, 1                                                         | 607426 | AR     |
| COQ6    | 614647 | Coenzyme Q10 deficiency, primary, 6                                                         | 614650 | AR     |
| COQ8B   | 615567 | Nephrotic syndrome, type 9                                                                  | 615573 | AR     |
| CRB2    | 609720 | Focal segmental glomerulosclerosis 9                                                        | 616220 | AR     |
|         |        | Ventriculomegaly with cystic kidney disease                                                 | 219730 | AR     |
| CRPPA   | 614631 | Muscular dystrophy-dystroglycanopathy [limb-girdle], type C, 7                              | 616052 | AR     |
|         |        | Muscular dystrophy-dystroglycanopathy [congenital with brain and eye anomalies], type A, 7  | 614643 | AR     |
| CRTAP   | 605497 | Osteogenesis imperfecta, type VII                                                           | 610682 | AR     |
| CTNS    | 606272 | Cystinosis, nephropathic                                                                    | 219800 | AR     |
|         |        | Cystinosis, ocular nonnephropathic                                                          | 219750 | AR     |
|         |        | Cystinosis, late-onset juvenile or adolescent nephropathic                                  | 219900 | AR     |
|         |        | Cystinosis, atypical nephropathic                                                           | 219800 | AR     |
| CUL3    | 603136 | Neurodevelopmental disorder with or without autism or seizures                              | 619239 | AD     |
|         |        | Pseudohypoaldosteronism, type IIE                                                           | 614496 | AD     |
| CYP11B1 | 610613 | Aldosteronism, glucocorticoid-remediable                                                    | 103900 | AD     |
|         |        | Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency                      | 202010 | AR     |
| CYP11B2 | 124080 | Hypoaldosteronism, congenital, due to CMO I deficiency                                      | 203400 | AR     |
|         |        | Aldosterone to renin ratio raised                                                           |        |        |
|         |        | {Low renin hypertension, susceptibility to}                                                 |        |        |
|         |        | Hypoaldosteronism, congenital, due to CMO II deficiency                                     | 610600 | AR     |
| CYP17A1 | 609300 | 17,20-lyase deficiency, isolated                                                            | 202110 | AR     |
|         |        | 17-alpha-hydroxylase/17,20-lyase deficiency                                                 | 202110 | AR     |
| CYP24A1 | 126065 | Hypercalcemia, infantile, 1                                                                 | 143880 | AR     |
| DGKE    | 601440 | {Hemolytic uremic syndrome, atypical, susceptibility to, 7}                                 | 615008 | AR     |
|         |        | Nephrotic syndrome, type 7                                                                  | 615008 | AR     |
| DICER1  | 606241 | Pleuropulmonary blastoma                                                                    | 601200 | AD     |
|         |        | Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors                          | 138800 | AD     |
|         |        | GLOW syndrome, somatic mosaic                                                               | 618272 |        |
|         |        | Rhabdomyosarcoma, embryonal, 2                                                              | 180295 |        |
| DIS3L2  | 614184 | Perlman syndrome                                                                            | 267000 | AR     |
| DMD     | 300377 | Becker muscular dystrophy                                                                   | 300376 | XLR    |
|         |        | Cardiomyopathy, dilated, 3B                                                                 | 302045 | XL     |
|         |        | Duchenne muscular dystrophy                                                                 | 310200 | XLR    |
| DNA2    | 601810 | ?Seckel syndrome 8                                                                          | 615807 | AR     |
|         |        | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6 | 615156 | AD     |
| DNAJB11 | 611341 | Polycystic kidney disease 6 with or without polycystic liver disease                        | 618061 | AD     |
| DNAJB6  | 611332 | Muscular dystrophy, limb-girdle, autosomal dominant 1                                       | 603511 | AD     |

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| DOK7   | 610285 | Fetal akinesia deformation sequence 3                                          | 618389 | AR |
|        |        | Myasthenic syndrome, congenital, 10                                            | 254300 | AR |
| DYSF   | 603009 | Muscular dystrophy, limb-girdle, autosomal recessive 2                         | 253601 | AR |
|        |        | Miyoshi muscular dystrophy 1                                                   | 254130 | AR |
|        |        | Myopathy, distal, with anterior tibial onset                                   | 606768 | AR |
| DZIP1L | 617570 | Polycystic kidney disease 5                                                    | 617610 | AR |
| ERF    | 611888 | Craniosynostosis 4                                                             | 600775 | AD |
|        |        | Chitayat syndrome                                                              | 617180 | AD |
| FAN1   | 613534 | Interstitial nephritis, karyomegalic                                           | 614817 | AR |
| FBN1   | 134797 | Geleophysic dysplasia 2                                                        | 614185 | AD |
|        |        | Weill-Marchesani syndrome 2, dominant                                          | 608328 | AD |
|        |        | Ectopia lentis, familial                                                       | 129600 | AD |
|        |        | MASS syndrome                                                                  | 604308 | AD |
|        |        | Marfan lipodystrophy syndrome                                                  | 616914 | AD |
|        |        | Acromicric dysplasia                                                           | 102370 | AD |
|        |        | Marfan syndrome                                                                | 154700 | AD |
|        |        | Stiff skin syndrome                                                            | 184900 | AD |
| FGF10  | 602115 | Aplasia of lacrimal and salivary glands                                        | 180920 | AD |
|        |        | LADD syndrome                                                                  | 149730 | AD |
| FGFR1  | 136350 | Pfeiffer syndrome                                                              | 101600 | AD |
|        |        | Hypogonadotropic hypogonadism 2 with or without anosmia                        | 147950 | AD |
|        |        | Jackson-Weiss syndrome                                                         | 123150 | AD |
|        |        | Hartsfield syndrome                                                            | 615465 | AD |
|        |        | Trigonocephaly 1                                                               | 190440 | AD |
|        |        | Osteoglophonic dysplasia                                                       | 166250 | AD |
|        |        | Encephalocraniocutaneous lipomatosis, somatic mosaic                           | 613001 |    |
| FGFR2  | 176943 | Bent bone dysplasia syndrome                                                   | 614592 | AD |
|        |        | LADD syndrome                                                                  | 149730 | AD |
|        |        | Scaphocephaly, maxillary retrusion, and mental retardation                     | 609579 |    |
|        |        | Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis | 207410 | AD |
|        |        | Scaphocephaly and Axenfeld-Rieger anomaly                                      |        |    |
|        |        | Jackson-Weiss syndrome                                                         | 123150 | AD |
|        |        | Gastric cancer, somatic                                                        | 613659 |    |
|        |        | Craniofacial-skeletal-dermatologic dysplasia                                   | 101600 | AD |
|        |        | Apert syndrome                                                                 | 101200 | AD |
|        |        | Pfeiffer syndrome                                                              | 101600 | AD |
|        |        | Craniosynostosis, nonspecific                                                  |        |    |
|        |        | Beare-Stevenson cutis gyrata syndrome                                          | 123790 | AD |
|        |        | Crouzon syndrome                                                               | 123500 | AD |
|        |        | Saethre-Chotzen syndrome                                                       | 101400 | AD |

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| FGFR3  | 134934 | Muenke syndrome                                                                                  | 602849 | AD     |
|        |        | SADDAN                                                                                           | 616482 | AD     |
|        |        | Hypochondroplasia                                                                                | 146000 | AD     |
|        |        | LADD syndrome                                                                                    | 149730 | AD     |
|        |        | Thanatophoric dysplasia, type II                                                                 | 187601 | AD     |
|        |        | Nevus, epidermal, somatic                                                                        | 162900 |        |
|        |        | CATSHL syndrome                                                                                  | 610474 | AR, AD |
|        |        | Thanatophoric dysplasia, type I                                                                  | 187600 | AD     |
|        |        | Spermatocytic seminoma, somatic                                                                  | 273300 |        |
|        |        | Bladder cancer, somatic                                                                          | 109800 |        |
|        |        | Achondroplasia                                                                                   | 100800 | AD     |
|        |        | Cervical cancer, somatic                                                                         | 603956 |        |
|        |        | Colorectal cancer, somatic                                                                       | 114500 |        |
|        |        | Crouzon syndrome with acanthosis nigricans                                                       | 612247 | AD     |
| FH     | 136850 | Leiomyomatosis and renal cell cancer                                                             | 150800 | AD     |
|        |        | Fumarase deficiency                                                                              | 606812 | AR     |
| FKBP10 | 607063 | Osteogenesis imperfecta, type XI                                                                 | 610968 | AR     |
|        |        | Bruck syndrome 1                                                                                 | 259450 | AR     |
| FKRP   | 606596 | Muscular dystrophy-dystroglycanopathy [congenital with or without mental retardation], type B, 5 | 606612 | AR     |
|        |        | Muscular dystrophy-dystroglycanopathy [limb-girdle], type C, 5                                   | 607155 | AR     |
|        |        | Muscular dystrophy-dystroglycanopathy [congenital with brain and eye anomalies], type A, 5       | 613153 | AR     |
| FKTN   | 607440 | Muscular dystrophy-dystroglycanopathy [limb-girdle], type C, 4                                   | 611588 | AR     |
|        |        | Muscular dystrophy-dystroglycanopathy [congenital with brain and eye anomalies], type A, 4       | 253800 | AR     |
|        |        | Muscular dystrophy-dystroglycanopathy [congenital without mental retardation], type B, 4         | 613152 | AR     |
|        |        | Cardiomyopathy, dilated, 1X                                                                      | 611615 | AR     |
|        |        | Birt-Hogg-Dube syndrome                                                                          | 135150 | AD     |
| FLCN   | 607273 | Colorectal cancer, somatic                                                                       | 114500 |        |
|        |        | Pneumothorax, primary spontaneous                                                                | 173600 | AD     |
|        |        | Renal carcinoma, chromophobe, somatic                                                            | 144700 |        |
|        |        | Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia                         | 608030 |        |
| FUS    | 137070 | Essential tremor, hereditary, 4                                                                  | 614782 | AD     |
| GANAB  | 104160 | Polycystic kidney disease 3                                                                      | 600666 | AD     |
| GCK    | 138079 | MODY, type II                                                                                    | 125851 | AD     |
|        |        | Diabetes mellitus, permanent neonatal 1                                                          | 606176 | AR     |
|        |        | Hyperinsulinemic hypoglycemia, familial, 3                                                       | 602485 | AD     |
|        |        | Diabetes mellitus, noninsulin-dependent, late onset                                              | 125853 | AD     |

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| GJB2    | 121011 | Keratoderma, palmoplantar, with deafness                                                    | 148350 | AD     |
|         |        | Deafness, autosomal recessive 1A                                                            | 220290 | DD, AR |
|         |        | Deafness, autosomal dominant 3A                                                             | 601544 | AD     |
|         |        | Hystrix-like ichthyosis with deafness                                                       | 602540 | AD     |
|         |        | Bart-Pumphrey syndrome                                                                      | 149200 | AD     |
|         |        | Keratitis-ichthyosis-deafness syndrome                                                      | 148210 | AD     |
|         |        | Vohwinkel syndrome                                                                          | 124500 | AD     |
| GJB6    | 604418 | Ectodermal dysplasia 2, Clouston type                                                       | 129500 | AD     |
|         |        | Deafness, autosomal dominant 3B                                                             | 612643 | AD     |
|         |        | Deafness, autosomal recessive 1B                                                            | 612645 | AR     |
|         |        | Deafness, digenic GJB2/GJB6                                                                 | 220290 | DD, AR |
| GMPPB   | 615320 | Muscular dystrophy-dystroglycanopathy [limb-girdle], type C, 14                             | 615352 | AR     |
|         |        | Muscular dystrophy-dystroglycanopathy [congenital with mental retardation], type B, 14      | 615351 | AR     |
|         |        | Muscular dystrophy-dystroglycanopathy [congenital with brain and eye anomalies], type A, 14 | 615350 | AR     |
| GPHN    | 603930 | Molybdenum cofactor deficiency C                                                            | 615501 | AR     |
| GRHPR   | 604296 | Hyperoxaluria, primary, type II                                                             | 260000 | AR     |
| HAO1    | 605023 |                                                                                             |        |        |
| HNF1A   | 142410 | Hepatic adenoma, somatic                                                                    | 142330 |        |
|         |        | Diabetes mellitus, insulin-dependent, 20                                                    | 612520 |        |
|         |        | {Diabetes mellitus, noninsulin-dependent, 2}                                                | 125853 | AD     |
|         |        | MODY, type III                                                                              | 600496 | AD     |
|         |        | {Diabetes mellitus, insulin-dependent}                                                      | 222100 | AR     |
|         |        | Renal cell carcinoma                                                                        | 144700 |        |
| HNF1B   | 189907 | Type 2 diabetes mellitus                                                                    | 125853 | AD     |
|         |        | Renal cysts and diabetes syndrome                                                           | 137920 | AD     |
|         |        | {Renal cell carcinoma}                                                                      | 144700 |        |
| HNF4A   | 600281 | Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young                   | 616026 | AD     |
|         |        | {Diabetes mellitus, noninsulin-dependent}                                                   | 125853 | AD     |
|         |        | MODY, type I                                                                                | 125850 | AD     |
| HOGA1   | 613597 | Hyperoxaluria, primary, type III                                                            | 613616 | AR     |
| HPRT1   | 308000 | Hyperuricemia, HRPT-related                                                                 | 300323 | XLR    |
|         |        | Lesch-Nyhan syndrome                                                                        | 300322 | XLR    |
| HSD11B2 | 614232 | Apparent mineralocorticoid excess                                                           | 218030 | AR     |
| IFITM5  | 614757 | Osteogenesis imperfecta, type V                                                             | 610967 | AD     |
| IGHMBP2 | 600502 | Neuronopathy, distal hereditary motor, type VI                                              | 604320 | AR     |
|         |        | Charcot-Marie-Tooth disease, axonal, type 2S                                                | 616155 | AR     |
| IL11RA  | 600939 | Craniosynostosis and dental anomalies                                                       | 614188 | AR     |
| INF2    | 610982 | Glomerulosclerosis, focal segmental, 5                                                      | 613237 |        |
|         |        | Charcot-Marie-Tooth disease, dominant intermediate E                                        | 614455 | AD     |
| INVS    | 243305 | Nephronophthisis 2, infantile                                                               | 602088 | AR     |

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|--------|--------|--------------------------------------------------------------------------------------------|--------|--------|
| KCNJ1  | 600359 | Bartter syndrome, type 2                                                                   | 241200 | AR     |
| KCNJ10 | 602208 | Enlarged vestibular aqueduct, digenic                                                      | 600791 | AR     |
|        |        | SESAME syndrome                                                                            | 612780 | AR     |
| KCNJ5  | 600734 | Long QT syndrome 13                                                                        | 613485 | AD     |
|        |        | Hyperaldosteronism, familial, type III                                                     | 613677 | AD     |
| KDM6A  | 300128 | Kabuki syndrome 2                                                                          | 300867 | XLD    |
| KLHL3  | 605775 | Pseudohypoaldosteronism, type IID                                                          | 614495 | AR, AD |
| KMT2D  | 602113 | Kabuki syndrome 1                                                                          | 147920 | AD     |
| KNL1   | 609173 | Microcephaly 4, primary, autosomal recessive                                               | 604321 | AR     |
| LAMB2  | 150325 | Nephrotic syndrome, type 5, with or without ocular abnormalities                           | 614199 |        |
|        |        | Pierson syndrome                                                                           | 609049 | AR     |
| LARGE1 | 603590 | Muscular dystrophy-dystroglycanopathy [congenital with mental retardation], type B, 6      | 608840 | AR     |
|        |        | Muscular dystrophy-dystroglycanopathy [congenital with brain and eye anomalies], type A, 6 | 613154 | AR     |
| LIG4   | 601837 | LIG4 syndrome                                                                              | 606593 | AR     |
|        |        | {Multiple myeloma, resistance to}                                                          | 254500 | Som    |
| LMNA   | 150330 | Mandibuloacral dysplasia                                                                   | 248370 | AR     |
|        |        | Heart-hand syndrome, Slovenian type                                                        | 610140 | AD     |
|        |        | Cardiomyopathy, dilated, 1A                                                                | 115200 | AD     |
|        |        | Restrictive dermopathy, lethal                                                             | 275210 | AR     |
|        |        | Emery-Dreifuss muscular dystrophy 3, autosomal recessive                                   | 616516 | AR     |
|        |        | Charcot-Marie-Tooth disease, type 2B1                                                      | 605588 | AR     |
|        |        | Emery-Dreifuss muscular dystrophy 2, autosomal dominant                                    | 181350 | AD     |
|        |        | Hutchinson-Gilford progeria                                                                | 176670 | AD     |
|        |        | Lipodystrophy, familial partial, type 2                                                    | 151660 | AD     |
|        |        | Muscular dystrophy, congenital                                                             | 613205 | AD     |
|        |        | Malouf syndrome                                                                            | 212112 | AD     |
| LMX1B  | 602575 | Focal segmental glomerulosclerosis 10                                                      | 256020 | AD     |
|        |        | Nail-patella syndrome                                                                      | 161200 | AD     |
| LRP5   | 603506 | Osteopetrosis, autosomal dominant 1                                                        | 607634 | AD     |
|        |        | [Bone mineral density variability 1]                                                       | 601884 | AD     |
|        |        | {Osteoporosis}                                                                             | 166710 | AD     |
|        |        | Hyperostosis, endosteal                                                                    | 144750 | AD     |
|        |        | Osteosclerosis                                                                             | 144750 | AD     |
|        |        | Polycystic liver disease 4 with or without kidney cysts                                    | 617875 | AD     |
|        |        | Osteoporosis-pseudoglioma syndrome                                                         | 259770 | AR     |
|        |        | Exudative vitreoretinopathy 4                                                              | 601813 | AR, AD |
|        |        | van Buchem disease, type 2                                                                 | 607636 |        |
| MAGED2 | 300470 | Bartter syndrome, type 5, antenatal, transient                                             | 300971 | XLR    |
| MAGEL2 | 605283 | Schaaf-Yang syndrome                                                                       | 615547 | AD     |
| MAX    | 154950 | {Pheochromocytoma, susceptibility to}                                                      | 171300 | AD     |

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| MCPH1  | 607117 | Microcephaly 1, primary, autosomal recessive                                          | 251200 | AR     |
| MEGF10 | 612453 | Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant   | 614399 | AR     |
|        |        | Myopathy, areflexia, respiratory distress, and dysphagia, early-onset                 | 614399 | AR     |
| MEN1   | 613733 | Lipoma, somatic                                                                       |        |        |
|        |        | Angiofibroma, somatic                                                                 |        |        |
|        |        | Multiple endocrine neoplasia 1                                                        | 131100 | AD     |
|        |        | Carcinoid tumor of lung                                                               |        |        |
|        |        | Adrenal adenoma, somatic                                                              |        |        |
|        |        | Parathyroid adenoma, somatic                                                          |        |        |
| MET    | 164860 | Renal cell carcinoma, papillary, 1, familial and somatic                              | 605074 |        |
|        |        | Hepatocellular carcinoma, childhood type, somatic                                     | 114550 |        |
|        |        | {Osteofibrous dysplasia, susceptibility to}                                           | 607278 | AD     |
|        |        | ?Deafness, autosomal recessive 97                                                     | 616705 | AR     |
| MMACHC | 609831 | Methylmalonic aciduria and homocystinuria, cblC type                                  | 277400 | AR     |
| MMADHC | 611935 | Methylmalonic aciduria, cblD type, variant 2                                          | 277410 | AR     |
|        |        | Methylmalonic aciduria and homocystinuria, cblD type                                  | 277410 | AR     |
|        |        | Homocystinuria, cblD type, variant 1                                                  | 277410 | AR     |
| MRE11  | 600814 | Ataxia-telangiectasia-like disorder 1                                                 | 604391 | AR     |
| MUSK   | 601296 | Fetal akinesia deformation sequence 1                                                 | 208150 | AR     |
|        |        | Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency | 616325 | AR     |
| MYBPC1 | 160794 | Myopathy, congenital, with tremor                                                     | 618524 | AD     |
|        |        | Lethal congenital contracture syndrome 4                                              | 614915 | AR     |
|        |        | Arthrogryposis, distal, type 1B                                                       | 614335 | AD     |
| MYBPC3 | 600958 | Cardiomyopathy, hypertrophic, 4                                                       | 115197 | AR, AD |
|        |        | Cardiomyopathy, dilated, 1MM                                                          | 615396 | AD     |
|        |        | Left ventricular noncompaction 10                                                     | 615396 | AD     |
| MYH11  | 160745 | Megacystis-microcolon-intestinal hypoperistalsis syndrome 2                           | 619351 | AR     |
|        |        | Aortic aneurysm, familial thoracic 4                                                  | 132900 | AD     |
|        |        | Visceral myopathy 2                                                                   | 619350 | AD     |
| MYH3   | 160720 | Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A                   | 178110 | AD     |
|        |        | Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B                    | 618469 | AR     |
|        |        | Arthrogryposis, distal, type 2B3 [Sheldon-Hall]                                       | 618436 | AD     |
|        |        | Arthrogryposis, distal, type 2A [Freeman-Sheldon]                                     | 193700 | AD     |
| MYH7   | 160760 | Laing distal myopathy                                                                 | 160500 | AD     |
|        |        | Cardiomyopathy, hypertrophic, 1                                                       | 192600 | DD, AD |
|        |        | Left ventricular noncompaction 5                                                      | 613426 | AD     |
|        |        | Cardiomyopathy, dilated, 1S                                                           | 613426 | AD     |
|        |        | Scapuloperoneal syndrome, myopathic type                                              | 181430 | AD     |
|        |        | Myopathy, myosin storage, autosomal dominant                                          | 608358 | AD     |
|        |        | Myopathy, myosin storage, autosomal recessive                                         | 255160 | AR     |

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| MYH8  | 160741 | Carney complex variant                                                                                        | 608837 |     |
|       |        | Trismus-pseudocamptodactyly syndrome                                                                          | 158300 | AD  |
| MYH9  | 160775 | Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss      | 155100 | AD  |
|       |        | Deafness, autosomal dominant 17                                                                               | 603622 | AD  |
| MYLK  | 600922 | Megacystis-microcolon-intestinal hypoperistalsis syndrome 1                                                   | 249210 | AR  |
|       |        | Aortic aneurysm, familial thoracic 7                                                                          | 613780 | AD  |
| MYO1E | 601479 | Glomerulosclerosis, focal segmental, 6                                                                        | 614131 | AR  |
| NBN   | 602667 | Leukemia, acute lymphoblastic                                                                                 | 613065 |     |
|       |        | Aplastic anemia                                                                                               | 609135 |     |
|       |        | Nijmegen breakage syndrome                                                                                    | 251260 | AR  |
| NHEJ1 | 611290 | Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation | 611291 |     |
| NIN   | 608684 | ?Seckel syndrome 7                                                                                            | 614851 | AR  |
| NPHP1 | 607100 | Joubert syndrome 4                                                                                            | 609583 | AR  |
|       |        | Nephronophthisis 1, juvenile                                                                                  | 256100 | AR  |
|       |        | Senior-Loken syndrome-1                                                                                       | 266900 | AR  |
| NPHP3 | 608002 | Nephronophthisis 3                                                                                            | 604387 | AR  |
|       |        | Renal-hepatic-pancreatic dysplasia 1                                                                          | 208540 | AR  |
|       |        | Meckel syndrome 7                                                                                             | 267010 | AR  |
| NPHS1 | 602716 | Nephrotic syndrome, type 1                                                                                    | 256300 | AR  |
| NPHS2 | 604766 | Nephrotic syndrome, type 2                                                                                    | 600995 | AR  |
| NR3C2 | 600983 | Pseudohypoaldosteronism type I, autosomal dominant                                                            | 177735 | AD  |
|       |        | Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy                                 | 605115 |     |
| OSGEP | 610107 | Galloway-Mowat syndrome 3                                                                                     | 617729 | AR  |
| P3H1  | 610339 | Osteogenesis imperfecta, type VIII                                                                            | 610915 | AR  |
| P4HB  | 176790 | Cole-Carpenter syndrome 1                                                                                     | 112240 | AD  |
| PAX2  | 167409 | Glomerulosclerosis, focal segmental, 7                                                                        | 616002 | AD  |
|       |        | Papillorenal syndrome                                                                                         | 120330 | AD  |
| PAX6  | 607108 | Optic nerve hypoplasia                                                                                        | 165550 | AD  |
|       |        | Cataract with late-onset corneal dystrophy                                                                    | 106210 | AD  |
|       |        | ?Coloboma, ocular                                                                                             | 120200 | AD  |
|       |        | ?Coloboma of optic nerve                                                                                      | 120430 | AD  |
|       |        | Aniridia                                                                                                      | 106210 | AD  |
|       |        | Anterior segment dysgenesis 5, multiple subtypes                                                              | 604229 | AD  |
|       |        | ?Morning glory disc anomaly                                                                                   | 120430 | AD  |
|       |        | Foveal hypoplasia 1                                                                                           | 136520 | AD  |
|       |        | Keratitis                                                                                                     | 148190 | AD  |
| PCNT  | 605925 | Microcephalic osteodysplastic primordial dwarfism, type II                                                    | 210720 | AR  |
| PDE3A | 123805 | Hypertension and brachydactyly syndrome                                                                       | 112410 | AD  |
| PDSS2 | 610564 | Coenzyme Q10 deficiency, primary, 3                                                                           | 614652 | AR  |
| PHF6  | 300414 | Borjeson-Forssman-Lehmann syndrome                                                                            | 301900 | XLR |

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| PIEZO2  | 613629 | Arthrogryposis, distal, type 5                                                              | 108145 | AD         |
|         |        | Arthrogryposis, distal, with impaired proprioception and touch                              | 617146 | AR         |
|         |        | Arthrogryposis, distal, type 3                                                              | 114300 | AD         |
|         |        | ?Marden-Walker syndrome                                                                     | 248700 | AD         |
| PIGB    | 604122 | Developmental and epileptic encephalopathy 80                                               | 618580 | AR         |
| PKD1    | 601313 | Polycystic kidney disease 1                                                                 | 173900 | AD         |
| PKD2    | 173910 | Polycystic kidney disease 2                                                                 | 613095 | AD         |
| PKHD1   | 606702 | Polycystic kidney disease 4, with or without hepatic disease                                | 263200 | AR         |
| PLCE1   | 608414 | Nephrotic syndrome, type 3                                                                  | 610725 | AR         |
| PLOD2   | 601865 | Bruck syndrome 2                                                                            | 609220 | AR         |
| PLS3    | 300131 | Bone mineral density QTL18, osteoporosis                                                    | 300910 | XLD        |
| PNKP    | 605610 | ?Charcot-Marie-Tooth disease, type 2B2                                                      | 605589 | AR         |
|         |        | Ataxia-oculomotor apraxia 4                                                                 | 616267 | AR         |
|         |        | Microcephaly, seizures, and developmental delay                                             | 613402 | AR         |
|         |        | Mitochondrial recessive ataxia syndrome [includes SANDO and SCAE]                           | 607459 | AR         |
| POLG    | 174763 | Mitochondrial DNA depletion syndrome 4B [MNGIE type]                                        | 613662 | AR         |
|         |        | Mitochondrial DNA depletion syndrome 4A [Alpers type]                                       | 203700 | AR         |
|         |        | Progressive external ophthalmoplegia, autosomal dominant 1                                  | 157640 | AD         |
|         |        | Progressive external ophthalmoplegia, autosomal recessive 1                                 | 258450 | AR         |
| POMC    | 176830 | {Obesity, early-onset, susceptibility to}                                                   | 601665 | AR, AD, Mu |
|         |        | Obesity, adrenal insufficiency, and red hair due to POMC deficiency                         | 609734 | AR         |
| POMGNT1 | 606822 | Muscular dystrophy-dystroglycanopathy [congenital with mental retardation], type B, 3       | 613151 | AR         |
|         |        | Muscular dystrophy-dystroglycanopathy [limb-girdle], type C, 3                              | 613157 | AR         |
|         |        | Retinitis pigmentosa 76                                                                     | 617123 | AR         |
| POMGNT2 | 614828 | Muscular dystrophy-dystroglycanopathy [congenital with brain and eye anomalies], type A, 3  | 253280 | AR         |
|         |        | Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8                               | 618135 | AR         |
|         |        | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8  | 614830 | AR         |
| POMK    | 615247 | ?Muscular dystrophy-dystroglycanopathy [limb-girdle], type C, 12                            | 616094 | AR         |
| POMT1   | 607423 | Muscular dystrophy-dystroglycanopathy [congenital with brain and eye anomalies], type A, 12 | 615249 | AR         |
|         |        | Muscular dystrophy-dystroglycanopathy [congenital with brain and eye anomalies], type A, 1  | 236670 | AR         |
|         |        | Muscular dystrophy-dystroglycanopathy [limb-girdle], type C, 1                              | 609308 | AR         |
| POMT2   | 607439 | Muscular dystrophy-dystroglycanopathy [congenital with mental retardation], type B, 1       | 613155 | AR         |
|         |        | Muscular dystrophy-dystroglycanopathy [limb-girdle], type C, 2                              | 613158 | AR         |
|         |        | Muscular dystrophy-dystroglycanopathy [congenital with mental retardation], type B, 2       | 613156 | AR         |
|         |        | Muscular dystrophy-dystroglycanopathy [congenital with brain and eye anomalies], type A, 2  | 613150 | AR         |

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| PPARG    | 601487 | {Diabetes, type 2}                                                                          | 125853 | AD         |
|          |        | Insulin resistance, severe, digenic                                                         | 604367 | AD         |
|          |        | Lipodystrophy, familial partial, type 3                                                     | 604367 | AD         |
|          |        | [Obesity, resistance to]                                                                    |        |            |
|          |        | Obesity, severe                                                                             | 601665 | AR, AD, Mu |
|          |        | Carotid intimal medial thickness 1                                                          | 609338 |            |
| PIIB     | 123841 | Osteogenesis imperfecta, type IX                                                            | 259440 | AR         |
| PRSS1    | 276000 | Pancreatitis, hereditary                                                                    | 167800 | AD         |
| PTPRO    | 600579 | Nephrotic syndrome, type 6                                                                  | 614196 | AR         |
| RAD50    | 604040 | Nijmegen breakage syndrome-like disorder                                                    | 613078 | AR         |
| RAPSN    | 601592 | Fetal akinesia deformation sequence 2                                                       | 618388 | AR         |
|          |        | Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency      | 616326 | AR         |
| RARS2    | 611524 | Pontocerebellar hypoplasia, type 6                                                          | 611523 | AR         |
| RBBP8    | 604124 | Seckel syndrome 2                                                                           | 606744 | AR         |
|          |        | Jawad syndrome                                                                              | 251255 | AR         |
|          |        | Pancreatic carcinoma, somatic                                                               |        |            |
| REN      | 179820 | Renal tubular dysgenesis                                                                    | 267430 | AR         |
|          |        | [Hyperproreninemia]                                                                         |        |            |
|          |        | Tubulointerstitial kidney disease, autosomal dominant, 4                                    | 613092 | AD         |
| RET      | 164761 | {Hirschsprung disease, susceptibility to, 1}                                                | 142623 | AD         |
|          |        | Multiple endocrine neoplasia IIA                                                            | 171400 | AD         |
|          |        | {Hirschsprung disease, protection against}                                                  | 142623 | AD         |
|          |        | Medullary thyroid carcinoma                                                                 | 155240 | AD         |
|          |        | Pheochromocytoma                                                                            | 171300 | AD         |
|          |        | Multiple endocrine neoplasia IIB                                                            | 162300 | AD         |
| RIT1     | 609591 | Noonan syndrome 8                                                                           | 615355 | AD         |
| RNU4atac | 601428 | Roifman syndrome                                                                            | 616651 | AR         |
|          |        | Lowry-Wood syndrome                                                                         | 226960 | AR         |
|          |        | Microcephalic osteodysplastic primordial dwarfism, type I                                   | 210710 | AR         |
| RXYLT1   | 605862 | Muscular dystrophy-dystroglycanopathy [congenital with brain and eye anomalies], type A, 10 | 615041 | AR         |
| SCNN1A   | 600228 | Pseudohypoaldosteronism, type I                                                             | 264350 | AR         |
|          |        | ?Liddle syndrome 3                                                                          | 618126 | AD         |
|          |        | Bronchiectasis with or without elevated sweat chloride 2                                    | 613021 | AD         |
| SCNN1B   | 600760 | Bronchiectasis with or without elevated sweat chloride 1                                    | 211400 | AD         |
|          |        | Pseudohypoaldosteronism, type I                                                             | 264350 | AR         |
|          |        | Liddle syndrome 1                                                                           | 177200 | AD         |
| SCNN1G   | 600761 | Bronchiectasis with or without elevated sweat chloride 3                                    | 613071 | AD         |
|          |        | Pseudohypoaldosteronism, type I                                                             | 264350 | AR         |
|          |        | Liddle syndrome 2                                                                           | 618114 | AD         |

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| SDHA     | 600857 | Cardiomyopathy, dilated, 1GG                                            | 613642 | AR            |
|          |        | Mitochondrial complex II deficiency, nuclear type 1                     | 252011 | AR            |
|          |        | Neurodegeneration with ataxia and late-onset optic atrophy              | 619259 | AD            |
|          |        | Parangangliomas 5                                                       | 614165 | AD            |
| SDHAF2   | 613019 | Parangangliomas 2                                                       | 601650 | AD            |
| SDHB     | 185470 | Parangangliomas 4                                                       | 115310 | AD            |
|          |        | Mitochondrial complex II deficiency, nuclear type 4                     | 619224 | AR            |
|          |        | Gastrointestinal stromal tumor                                          | 606764 | IC, AD        |
|          |        | Pheochromocytoma                                                        | 171300 | AD            |
| SDHC     | 602413 | Paranganglioma and gastric stromal sarcoma                              | 606864 |               |
|          |        | Parangangliomas 3                                                       | 605373 | AD            |
|          |        | Paranganglioma and gastric stromal sarcoma                              | 606864 |               |
|          |        | Gastrointestinal stromal tumor                                          | 606764 | IC, AD        |
| SDHD     | 602690 | Parangangliomas 1, with or without deafness                             | 168000 | AD            |
|          |        | Paranganglioma and gastric stromal sarcoma                              | 606864 |               |
|          |        | Mitochondrial complex II deficiency, nuclear type 3                     | 619167 | AR            |
|          |        | Pheochromocytoma                                                        | 171300 | AD            |
| SEC23A   | 610511 | Craniofasciocranial dysplasia                                           | 607812 | AR            |
| SEC24D   | 607186 | Cole-Carpenter syndrome 2                                               | 616294 | AR            |
| SELENON  | 606210 | Myopathy, congenital, with fiber-type disproportion                     | 255310 | AR, AD        |
|          |        | Muscular dystrophy, rigid spine, 1                                      | 602771 | AR            |
| SEPSECS  | 613009 | Pontocerebellar hypoplasia type 2D                                      | 613811 | AR            |
| SERPINF1 | 172860 | Osteogenesis imperfecta, type VI                                        | 613982 | AR            |
| SERPINH1 | 600943 | {Preterm premature rupture of the membranes, susceptibility to}         | 610504 |               |
|          |        | Osteogenesis imperfecta, type X                                         | 613848 | AR            |
| SHOX     | 312865 |                                                                         |        | Pseudoautosom |
|          |        | Short stature, idiopathic familial                                      | 300582 | al dominant   |
|          |        | Leri-Weill dyschondrosteosis                                            | 127300 | Pseudoautosom |
|          |        | Langer mesomelic dysplasia                                              | 249700 | al recessive  |
| SLC12A3  | 600968 | Gitelman syndrome                                                       | 263800 | AR            |
| SLC25A11 | 604165 | Parangangliomas 6                                                       | 618464 | AD            |
| SLC26A4  | 605646 | Deafness, autosomal recessive 4, with enlarged vestibular aqueduct      | 600791 | AR            |
|          |        | Pendred syndrome                                                        | 274600 | AR            |
| SLC34A3  | 609826 | Hypophosphatemic rickets with hypercalciuria                            | 241530 | AR            |
| SLC3A1   | 104614 | Cystinuria                                                              | 220100 | AR, AD        |
| SLC5A2   | 182381 | Renal glucosuria                                                        | 233100 | AR, AD        |
| SLC7A9   | 604144 | Cystinuria                                                              | 220100 | AR, AD        |
| SMAD2    | 601366 | Loeys-Dietz syndrome 6                                                  | 619656 | AD            |
|          |        | Congenital heart defects, multiple types, 8, with or without heterotaxy | 619657 | AD            |

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| SMARCA2 | 600014 | Nicolaides-Baraitser syndrome                                   | 601358 | AD     |
|         |        | Blepharophimosis-impaired intellectual development syndrome     | 619293 | AD     |
| SMARCA4 | 603254 | Coffin-Siris syndrome 4                                         | 614609 | AD     |
|         |        | {Rhabdoid tumor predisposition syndrome 2}                      | 613325 | AD     |
| SMARCA1 | 606622 | Schimke immunoosseous dysplasia                                 | 242900 | AR     |
| SMARCB1 | 601607 | Rhabdoid tumors, somatic                                        | 609322 |        |
|         |        | {Schwannomatosis-1, susceptibility to}                          | 162091 | AD     |
|         |        | Coffin-Siris syndrome 3                                         | 614608 | AD     |
|         |        | {Rhabdoid tumor predisposition syndrome 1}                      | 609322 | AD     |
| SMARCE1 | 603111 | {Meningioma, familial, susceptibility to}                       | 607174 | AD     |
|         |        | Coffin-Siris syndrome 5                                         | 616938 | AD     |
| SMN1    | 600354 | Spinal muscular atrophy-2                                       | 253550 | AR     |
|         |        | Spinal muscular atrophy-4                                       | 271150 | AR     |
|         |        | Spinal muscular atrophy-3                                       | 253400 | AR     |
|         |        | Spinal muscular atrophy-1                                       | 253300 | AR     |
| SMN2    | 601627 | {Spinal muscular atrophy, type III, modifier of}                | 253400 | AR     |
| SOD1    | 147450 | Spastic tetraplegia and axial hypotonia, progressive            | 618598 | AR     |
|         |        | Amyotrophic lateral sclerosis 1                                 | 105400 | AR, AD |
| SP7     | 606633 | Osteogenesis imperfecta, type XII                               | 613849 | AR     |
| SPARC   | 182120 | Osteogenesis imperfecta, type XVII                              | 616507 | AR     |
| STIL    | 181590 | Microcephaly 7, primary, autosomal recessive                    | 612703 | AR     |
| SUOX    | 606887 | Sulfite oxidase deficiency                                      | 272300 | AR     |
| TARDBP  | 605078 | Frontotemporal lobar degeneration, TARDBP-related               | 612069 | AD     |
|         |        | Amyotrophic lateral sclerosis 10, with or without FTD           | 612069 | AD     |
| TGFB2   | 190220 | Loeys-Dietz syndrome 4                                          | 614816 | AD     |
| TGFBR1  | 190181 | {Multiple self-healing squamous epithelioma, susceptibility to} | 132800 | AD     |
|         |        | Loeys-Dietz syndrome 1                                          | 609192 | AD     |
| TGFBR2  | 190182 | Loeys-Dietz syndrome 2                                          | 610168 | AD     |
|         |        | Colorectal cancer, hereditary nonpolyposis, type 6              | 614331 |        |
|         |        | Esophageal cancer, somatic                                      | 133239 |        |
| THBD    | 188040 | Thrombophilia due to thrombomodulin defect                      | 614486 |        |
|         |        | {Hemolytic uremic syndrome, atypical, susceptibility to, 6}     | 612926 | AD     |
| TMEM127 | 613403 | {Pheochromocytoma, susceptibility to}                           | 171300 | AD     |
| TMEM38B | 611236 | Osteogenesis imperfecta, type XIV                               | 615066 |        |
| TNNI2   | 191043 | Arthrogryposis, distal, type 2B1                                | 601680 | AD     |
| TNNI3   | 191044 | ?Cardiomyopathy, dilated, 2A                                    | 611880 | AR     |
|         |        | Cardiomyopathy, hypertrophic, 7                                 | 613690 | AD     |
|         |        | Cardiomyopathy, familial restrictive, 1                         | 115210 | AD     |
|         |        | Cardiomyopathy, dilated, 1FF                                    | 613286 |        |

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|--------|--------|---------------------------------------------------|--------|---------|
| TNNT2  | 191045 | Cardiomyopathy, dilated, 1D                       | 601494 | AD      |
|        |        | Cardiomyopathy, hypertrophic, 2                   | 115195 | AD      |
|        |        | Cardiomyopathy, familial restrictive, 3           | 612422 | AD      |
|        |        | Left ventricular noncompaction 6                  | 601494 | AD      |
| TNNT3  | 600692 | Arthrogryposis, distal, type 2B2                  | 618435 | AD      |
| TP53   | 191170 | {Basal cell carcinoma 7}                          | 614740 | AD      |
|        |        | {Adrenocortical carcinoma, pediatric}             | 202300 | AD      |
|        |        | Hepatocellular carcinoma, somatic                 | 114550 |         |
|        |        | Breast cancer, somatic                            | 114480 |         |
|        |        | Li-Fraumeni syndrome                              | 151623 | AD      |
|        |        | Pancreatic cancer, somatic                        | 260350 |         |
|        |        | Nasopharyngeal carcinoma, somatic                 | 607107 |         |
|        |        | {Osteosarcoma}                                    | 259500 | Som     |
|        |        | {Choroid plexus papilloma}                        | 260500 | AD      |
|        |        | {Colorectal cancer}                               | 114500 | AD, Som |
|        |        | {Glioma susceptibility 1}                         | 137800 | AD, Som |
|        |        | Bone marrow failure syndrome 5                    | 618165 | AD      |
| TPM1   | 191010 | Left ventricular noncompaction 9                  | 611878 | AD      |
|        |        | Cardiomyopathy, hypertrophic, 3                   | 115196 | AD      |
|        |        | Cardiomyopathy, dilated, 1Y                       | 611878 | AD      |
| TPM2   | 190990 | Arthrogryposis, distal, type 2B4                  | 108120 | AD      |
|        |        | Arthrogryposis, distal, type 1A                   | 108120 | AD      |
|        |        | Nemaline myopathy 4, autosomal dominant           | 609285 | AD      |
|        |        | CAP myopathy 2                                    | 609285 | AD      |
| TRIP13 | 604507 | Oocyte maturation defect 9                        | 619011 | AR      |
|        |        | Mosaic variegated aneuploidy syndrome 3           | 617598 | AR      |
| TRPC6  | 603652 | Glomerulosclerosis, focal segmental, 2            | 603965 | AD      |
| TRPV4  | 605427 | Spondylometaphyseal dysplasia, Kozlowski type     | 184252 | AD      |
|        |        | Digital arthropathy-brachydactyly, familial       | 606835 | AD      |
|        |        | [Sodium serum level QTL 1]                        | 613508 |         |
|        |        | SED, Maroteaux type                               | 184095 | AD      |
|        |        | Metatropic dysplasia                              | 156530 | AD      |
|        |        | Scapuloperoneal spinal muscular atrophy           | 181405 | AD      |
|        |        | Hereditary motor and sensory neuropathy, type IIc | 606071 | AD      |
|        |        | ?Avascular necrosis of femoral head, primary, 2   | 617383 | AD      |
|        |        | Neuronopathy, distal hereditary motor, type VIII  | 600175 | AD      |
|        |        | Parastremmatic dwarfism                           | 168400 | AD      |
|        |        | Brachyolmia type 3                                | 113500 | AD      |

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| TSC1   | 605284 | Focal cortical dysplasia, type II, somatic                                           | 607341 | AD  |
|        |        | Tuberous sclerosis-1                                                                 | 191100 |     |
|        |        | Lymphangiomyomatosis                                                                 | 606690 |     |
| TSC2   | 191092 | Lymphangiomyomatosis, somatic                                                        | 606690 |     |
|        |        | ?Focal cortical dysplasia, type II, somatic                                          | 607341 |     |
|        |        | Tuberous sclerosis-2                                                                 | 613254 |     |
| TSEN2  | 608753 | Pontocerebellar hypoplasia type 2B                                                   | 612389 | AR  |
| TSEN34 | 608754 | ?Pontocerebellar hypoplasia type 2C                                                  | 612390 | AR  |
| TSEN54 | 608755 | Pontocerebellar hypoplasia type 2A                                                   | 277470 | AR  |
|        |        | Pontocerebellar hypoplasia type 4                                                    | 225753 | AR  |
|        |        | ?Pontocerebellar hypoplasia type 5                                                   | 610204 | AR  |
| TTR    | 176300 | Amyloidosis, hereditary, transthyretin-related                                       | 105210 | AD  |
|        |        | Carpal tunnel syndrome, familial                                                     | 115430 | AD  |
|        |        | [Dystransthyretinemic hyperthyroxinemia]                                             | 145680 | AD  |
| TWIST1 | 601622 | Craniosynostosis 1                                                                   | 123100 | AD  |
|        |        | Robinow-Sorauf syndrome                                                              | 180750 | AD  |
|        |        | Sweeney-Cox syndrome                                                                 | 617746 | AD  |
| UBA1   | 314370 | Saethre-Chotzen syndrome with or without eyelid anomalies                            | 101400 | AD  |
|        |        | Spinal muscular atrophy, X-linked 2, infantile                                       | 301830 | XLR |
|        |        | VEXAS syndrome, somatic                                                              | 301054 |     |
| UBE3A  | 601623 | Angelman syndrome                                                                    | 105830 | AD  |
| UMOD   | 191845 | Tubulointerstitial kidney disease, autosomal dominant, 1                             | 162000 | AD  |
| VAPB   | 605704 | Spinal muscular atrophy, late-onset, Finkel type                                     | 182980 | AD  |
|        |        | Amyotrophic lateral sclerosis 8                                                      | 608627 | AD  |
| VHL    | 608537 | Hemangioblastoma, cerebellar, somatic                                                |        |     |
|        |        | Erythrocytosis, familial, 2                                                          | 263400 | AR  |
|        |        | von Hippel-Lindau syndrome                                                           | 193300 | AD  |
|        |        | Renal cell carcinoma, somatic                                                        | 144700 |     |
|        |        | Pheochromocytoma                                                                     | 171300 | AD  |
| VWA1   | 611901 | Neuropathy, hereditary motor, with myopathic features                                | 619216 | AR  |
| WDR62  | 613583 | Microcephaly 2, primary, autosomal recessive, with or without cortical malformations | 604317 | AR  |
| WDR73  | 616144 | Galloway-Mowat syndrome 1                                                            | 251300 | AR  |
| WNK1   | 605232 | Neuropathy, hereditary sensory and autonomic, type II                                | 201300 | AR  |
|        |        | Pseudohypoaldosteronism, type IIC                                                    | 614492 | AD  |
| WNK4   | 601844 | Pseudohypoaldosteronism, type IIB                                                    | 614491 | AD  |
| WNT1   | 164820 | {Osteoporosis, early-onset, susceptibility to, autosomal dominant}                   | 615221 |     |
|        |        | Osteogenesis imperfecta, type XV                                                     | 615220 | AR  |

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| WT1    | 607102 | Mesothelioma, somatic                                  | 156240 |         |
|        |        | Meacham syndrome                                       | 608978 | AD      |
|        |        | Frasier syndrome                                       | 136680 | AD, Som |
|        |        | Nephrotic syndrome, type 4                             | 256370 | AD      |
|        |        | Denys-Drash syndrome                                   | 194080 | AD, Som |
|        |        | Wilms tumor, type 1                                    | 194070 | AD, Som |
| XRCC4  | 194363 | Short stature, microcephaly, and endocrine dysfunction | 616541 | AR      |
| ZNF335 | 610827 | Microcephaly 10, primary, autosomal recessive          | 615095 | AR      |