異常である。
（倫理面への配慮）
遺伝子変異解析はヒトゲノム・遺伝子解析研究に関する倫理指針を遵守して実施された。随時、慶應義塾大学臨床遺伝学センター・国立成育医療研究センター遺伝診療科において遺伝カウンセリングを提供した。眼科所見については、患者家族に十分な説明を行い、書面にて検査結果の二次利用について同意を得た。診療録の調査や選択された症例の解析にあたっては、匿名化し、個人が特定できないように配慮した。

C．研究結果および考察

１）眼先天異常の遺伝子変異解析パネルの設計

方法に示した順に解析を進め、857 遺伝子・1130 疾患の眼科疾患関連遺伝子名と疾患名の対照表を得ることができた。
（付表）

２）眼先天異常に対する遺伝子診断と病態解明

両眼性の小角膜・小眼球・先天白内障など前眼部の眼先天異常患児に対し、解析パネルの運用を進めている。また小眼球症や先天無虹彩、視神経低形成などの疾患に対し、光干渉断層計（optical coherence tomography: OCT）、全視野および黄斑局所網膜電図（electroretinogram: ERG）を新たに導入して精密な形態・機能解析を行った。

D．考察

ヒトの眼科疾患に関わる遺伝子のリストを編纂することが出来た。この 857 遺伝子を標的遺伝子とした解析系を作成すれば、効率よく遺伝性眼疾患のスクリーニングが可能になると期待される。また最近、次世代シーケンサーのコストが低下し、あらかじめ決めた数百個の遺伝子に限って調べる分析を進める代わりに、全 2 万 5000 遺伝子を先に全部しらべて（エクソーム解析）、見つかった変異と様々なリストを比較して見つかった変異の病的意義を決めて行くというようなアプローチが欧米の一部の施設で一般化しつつある。上述の眼科疾患関連遺伝子名と疾患名の対照表はエクソーム解析の解釈にも極めて有用と期待される。

本研究班には全国から多種多様な眼先天異常患者が集まっており、新たな眼科的形態・機能解析評価法の導入も進んでいる。遺伝子変異解析パネルを運用して、効率的な遺伝子診断法を確立・普及させること、これに基づく病態解析・治療法の開発を推進することが今後の課題である。

E．結論

本研究班の目的を推進するために、眼科領域において臨床応用可能な遺伝子変異解析パネルの設計を開始した。これを用いることにより、眼先天異常に対しても、効率的な遺伝子診断が可能になり、病態解明に寄与すると期待される。

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G. 知的財産権の出願・登録状況(予定を含む。)

1. 特許取得
なし
2. 実用新案登録
なし
3. その他
なし

付表：眼科疾患関連遺伝子と疾患の対照表

ABCA4 "ATP-binding cassette, sub-family A (ABC1), member 4" 604116 Cone-rod dystrophy 3
ABCA4 "ATP-binding cassette, sub-family A (ABC1), member 4" 248200 Fundus flavimaculatus
ABCA4 "ATP-binding cassette, sub-family A (ABC1), member 4" 153800 " Macular degeneration, age-related, 2"
ABCA4 "ATP-binding cassette, sub-family A (ABC1), member 4" 248200 " Retinal dystrophy, early-onset severe"
ABCA4 "ATP-binding cassette, sub-family A (ABC1), member 4" 601718 Retinitis pigmentosa-19
ABCA4 "ATP-binding cassette, sub-family A (ABC1), member 4" 248200 Stargardt disease-1
ABCC2 "ATP-binding cassette, sub-family C (CFTR/MRP), member 2" 237500 Dubin-Johnson syndrome
ABCC6 "ATP-binding cassette, sub-family C (CFTR/MRP), member 6" 264800 Pseudoxanthoma elasticum
ABCC6 "ATP-binding cassette, sub-family C (CFTR/MRP), member 6" 177850 " Pseudoxanthoma elasticum, forme fruste"
ABCG5 "ATP-binding cassette, sub-family G (WHITE), member 5" 210250 Sitosterolemia
ABCG8 "ATP-binding cassette, sub-family G (WHITE), member 8" 611465 Gallbladder disease 4
ABCG8 "ATP-binding cassette, sub-family G (WHITE), member 8" 210250 Sitosterolemia
ACADVL "acyl-CoA dehydrogenase, very long chain" 201475 VLCAD deficiency
ACE angiotensin I converting enzyme (peptidyl-dipeptidase A) 1 267430 Renal tubular dysgenesis
ACTA1 "actin, alpha 1, skeletal muscle" 161800 " Myopathy, actin, congenital, with excess of thin myofilaments"
ACTA1 "actin, alpha 1, skeletal muscle" 255310 " Myopathy, congenital, with fiber-type disproportion 1"
ACTA1 "actin, alpha 1, skeletal muscle" 161800 " Myopathy, nemaline, 3"
ACTA2 "actin, alpha 2, smooth muscle, aorta" 611788 " Aortic aneurysm, familial thoracic 6"
ACTB "actin, beta" 607371 " Dystonia, juvenile-onset"
ACTC1 "actin, alpha, cardiac muscle 1" 612794 Atrial septal defect 5
ACTC1 "actin, alpha, cardiac muscle 1" 613424 " Cardiomyopathy, dilated, 1R"
ACTC1 "actin, alpha, cardiac muscle 1" 612098 " Cardiomyopathy, familial hypertrophic, 11"
ACTC1 "actin, alpha, cardiac muscle 1" 613424 Left ventricular noncompaction 4
ACTG1 "actin, gamma 1" 604717 " Deafness, autosomal dominant 20/26"
ACVRL1 activin A receptor type II-like 1 600376 Hereditary hemorrhagic telangiectasia-2
ADAM9 ADAM metallopeptidase domain 9 612775 Cone-rod dystrophy 9
ADAMTS10 "ADAM metallopeptidase with thrombospondin type 1 motif, 10" 277600 " Weill-Marchesani syndrome, recessive"
ADAMTS17 "ADAM metallopeptidase with thrombospondin type 1 motif, 17" 613195 Weill-Marchesani-like syndrome
ADAMTSL4 ADAMTS-like 4 225100 " Ectopia lentis, isolated, autosomal recessive"
ADIPOQ "adiponectin, C1Q and collagen domain containing" 612556 Adiponectin deficiency
AFG3L2 AFG3 ATPase family gene 3-like 2 (S. cerevisiae) 610246 Spinocerebellar ataxia 28
AGPS alkylglycerone phosphate synthase 600121 Rhizomelic chondrodysplasia punctata type 3
AGTR1 "angiotensin II receptor, type 1" 145500 " Hypertension, essential"
AGTR1 "angiotensin II receptor, type 1" 267430 Renal tubular dysgenesis
AHI1 Abelson helper integration site 1 608629 Joubert syndrome-3
AIFM1 "apoptosis-inducing factor, mitochondrion-associated, 1" 300816 Combined oxidative phosphorylation deficiency 6
AIPL1 aryl hydrocarbon receptor interacting protein-like 1 604393 Leber congenital amaurosis 4
AIRE autoimmune regulator 240300 " Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia"
ALDH7A1 "aldehyde dehydrogenase 7 family, member A1" 266100 " Epilepsy, pyridoxine-dependent"
ALMS1 Alstrom syndrome 1 203800 Alstrom syndrome
ALX1 ALX homeobox 1 613456 Frontonasal dysplasia 3
ALX3 ALX homeobox 3 136760 Frontorhiny
ALX4 ALX homeobox 4 613451 Frontonasal dysplasia 2
ALX4 ALX homeobox 4 609597 Parietal foramina 2
ANK2 "ankyrin 2, neuronal" 600919 " Cardiac arrhythmia, ankyrin-B-related"
ANK2 "ankyrin 2, neuronal" 600919 Long QT syndrome-4
AP3B1 "adaptor-related protein complex 3, beta 1 subunit" 608233 Hermansky-Pudlak syndrome 2

APC adenomatous polyposis coli 175100 Adenomatous polyposis coli
APC adenomatous polyposis coli 114500 " Colorectal cancer, somatic"
APC adenomatous polyposis coli 135290 " Desmoid disease, hereditary"
APC adenomatous polyposis coli 137215 " Gastric cancer, somatic"
APOA1 apolipoprotein A-I 105200 " Amyloidosis, 3 or more types"
APOA1 apolipoprotein A-I 604091 Hypoalphalipoproteinemia
APOB apolipoprotein B (including Ag(x) antigen) 144010 " Hypercholesterolemia, due to ligand-defective apo B"
APOE apolipoprotein E 104310 Alzheimer disease-2
APOE apolipoprotein E 611771 Lipoprotein glomerulopathy
APOE apolipoprotein E 269600 Sea-blue histiocyte disease
ARL6 ADP-ribosylation factor-like 6 209900 Bardet-Biedl syndrome 3
ARL6 ADP-ribosylation factor-like 6 613575 Retinitis pigmentosa 55
ARSB arylsulfatase B 253200 " Maroteaux-Lamy syndrome, several forms"
ARX aristaless related homeobox 300432 " Epilepsy, myoclonic, with mental retardation and spasticity"
ARX aristaless related homeobox 308350 " Epileptic encephalopathy, early infantile, 1"
ARX aristaless related homeobox 300215 Hydranencephaly with abnormal genitalia
ARX aristaless related homeobox 300215 " Lissencephaly, X-linked 2"
ARX aristaless related homeobox 300419 " Mental retardation, X-linked 36/43/54"
ARX aristaless related homeobox 309510 Partington syndrome
ARX aristaless related homeobox 300004 Proud syndrome
ASIP agouti signaling protein 611742 " Skin/hair/eye pigmentation 9, dark/light hair"
ASPA aspartoacylase 271900 Canavan disease
ASS1 argininosuccinate synthase 1 215700 Citrullinemia
ATP13A2 ATPase type 13A2 606693 Parkinson disease 9
ATRX alpha thalassemia/mental retardation syndrome X-linked 300448 " Alpha-thalassemia myelodysplasia syndrome, somatic"
ATRX alpha thalassemia/mental retardation syndrome X-linked 301040 Alpha-thalassemia mental retardation syndrome
ATRX alpha thalassemia/mental retardation syndrome X-linked 309580 " Mental retardation-hypotonic facies syndrome, X-linked"
ATXN1 ataxin 1 164400 Spinocerebellar ataxia-1
ATXN10 ataxin 10 603516 Spinocerebellar ataxia-10
ATXN2 ataxin 2 183090 Spinocerebellar ataxia-2
ATXN3 ataxin 3 109150 Machado-Joseph disease
ATXN7 ataxin 7 164500 Spinocerebellar ataxia-7
ATXN8 ataxin 8 608768 Spinocerebellar ataxia 8
AXIN1 axin 1 607864 Caudal duplication anomaly
AXIN1 axin 1 114550 " Hepatocellular carcinoma, somatic"
AXIN2 axin 2 114500 Colorectal cancer
AXIN2 axin 2 608615 Oligodontia-colorectal cancer syndrome
B3GALTL "beta 1,3-galactosyltransferase-like" 261540 Peters-plus syndrome B3GTL
BBS1 Bardet-Biedl syndrome 1 209900 Bardet-Biedl syndrome 1
BBS2 Bardet-Biedl syndrome 2 209900 Bardet-Biedl syndrome 2
BBS4 Bardet-Biedl syndrome 4 209900 Bardet-Biedl syndrome 4
BBS7 Bardet-Biedl syndrome 7 209900 Bardet-Biedl syndrome 7
BCOR BCL6 corepressor 300166 " Microphthalmia, syndromic 2"
BDNF brain-derived neurotrophic factor 209880 " Central hypoventilation syndrome, congenital"
BEST1 bestrophin 1 153700 Best macular dystrophy
BEST1 bestrophin 1 611809 Bestrophinopathy
BEST1 bestrophin 1 193220 " Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma"
BEST1 bestrophin 1 613194 " Retinitis pigmentosa, concentric"
BEST1 bestrophin 1 613194 Retinitis pigmentosa-50
BEST1 bestrophin 1 608161 " Vitelliform macular dystrophy, adult-onset"
BEST1 bestrophin 1 193220 Vitreoretinochoroidopathy

BFSP1 "beaded filament structural protein 1, filensin" 611391 " Cataract, cortical, juvenile-onset"
BFSP2 "beaded filament structural protein 2, phakinin" 611597 " Cataract, autosomal dominant, multiple types 1"
BFSP2 "beaded filament structural protein 2, phakinin" 604219 " Cataract, congenital"
BFSP2 "beaded filament structural protein 2, phakinin" 604219 " Cataract, juvenile-onset"
BLOC1S3 "biogenesis of lysosomal organelles complex-1, subunit 3" 203300 Hermansky-Pudlak syndrome 8
BMP4 bone morphogenetic protein 4 607932 " Microphthalmia, syndromic 6"
BMP4 bone morphogenetic protein 4 600625 Orofacial cleft 11
BMPR1A "bone morphogenetic protein receptor, type IA" 174900 " Juvenile polyposis syndrome, infantile form"
BMPR1A "bone morphogenetic protein receptor, type IA" 610069 " Polyposis syndrome, hereditary mixed, 2"
BMPR1A "bone morphogenetic protein receptor, type IA" 174900 " Polyposis, juvenile intestinal"
BMPR1B "bone morphogenetic protein receptor, type IB" 112600 Brachydactyly type A2
BMPR1B "bone morphogenetic protein receptor, type IB" 609441 " Chrondrodysplasia, acromesomelic, with genital anomalies"
BMPR2 "bone morphogenetic protein receptor, type II (serine/threonine kinase)" 178600 " Pulmonary hypertension, familial primary"
BMPR2 "bone morphogenetic protein receptor, type II (serine/threonine kinase)" 178600 " Pulmonary hypertension, primary, fenfluramine-associated"
BMPR2 "bone morphogenetic protein receptor, type II (serine/threonine kinase)" 265450 Pulmonary venoocclusive disease
BRAF v-raf murine sarcoma viral oncogene homolog B1 211980 " Adenocarcinoma of lung, somatic"
BRAF v-raf murine sarcoma viral oncogene homolog B1 115150 Cardiofaciocutaneous syndrome
BRCA2 "breast cancer 2, early onset" 605724 " Fanconi anemia, complementation group D1"
BRCA2 "breast cancer 2, early onset" 613347 Pancreatic cancer
BRCA2 "breast cancer 2, early onset" 176807 Prostate cancer
BRCA2 "breast cancer 2, early onset" 194070 Wilms tumor
BTD biotinidase 253260 Biotinidase deficiency
BUB1B budding uninhibited by benzimidazoles 1 homolog beta (yeast) 114500 Colorectal cancer
BUB1B budding uninhibited by benzimidazoles 1 homolog beta (yeast) 257300 Mosaic variegated aneuploidy syndrome
C10orf2 chromosome 10 open reading frame 2 251880 " Mitochondrial DNA depletion syndrome, hepatocerebral form"
C10orf2 chromosome 10 open reading frame 2 609286 Progressive external ophthalmoplegia with mitochondrial DNA deletions 3
C10orf2 chromosome 10 open reading frame 2 271245 " Spinocerebellar ataxia, infantile-onset"
C1QTNF5 C1q and tumor necrosis factor related protein 5 605670 " Retinal degeneration, late-onset, autosomal dominant"
CA2 carbonic anhydrase II 259730 " Osteopetrosis, autosomal recessive 3, with renal tubular acidosis"
CA4 carbonic anhydrase IV 600852 Retinitis pigmentosa-17
CABP4 calcium binding protein 4 610427 " Night blindness, congenital stationary, type 2B"
CACNA1A "calcium channel, voltage-dependent, P/Q type, alpha 1A subunit" 108500 " Episodic ataxia, type 2"
CACNA1A "calcium channel, voltage-dependent, P/Q type, alpha 1A subunit" 141500 " Hemiplegic migraine, familial"
CACNA1A "calcium channel, voltage-dependent, P/Q type, alpha 1A subunit" 183086 Spinocerebellar ataxia-6
CACNA2D4 "calcium channel, voltage-dependent, alpha 2/delta subunit 4" 610478 Retinal cone dystrophy 4
CACNB2 "calcium channel, voltage-dependent, beta 2 subunit" 611876 Brugada syndrome 4
CANT1 calcium activated nucleotidase 1 251450 Desbuquois dysplasia
CASR calcium-sensing receptor 239200 " Hyperparathyroidism, neonatal"
CASR calcium-sensing receptor 146200 " Hypocalcemia, autosomal dominant"
CASR calcium-sensing receptor 145980 " Hypocalciuric hypercalcemia, type I"
CAV1 "caveolin 1, caveolae protein, 22kDa" 612526 " Lipodystrophy, congenital generalized, type 3"
CAV3 caveolin 3 192600 " Cardiomyopathy, familial hypertrophic"
CAV3 caveolin 3 123320 " Creatine phosphokinase, elevated serum"
CAV3 caveolin 3 611818 Long QT syndrome-9
CAV3 caveolin 3 607801 " Muscular dystrophy, limb-girdle, type IC"
CAV3 caveolin 3 606072 Rippling muscle disease
CBS cystathionine-beta-synthase 236200 " Homocystinuria, B6-responsive and nonresponsive types"
CBS cystathionine-beta-synthase 236200 " Thrombosis, hyperhomocysteinemic"
CC2D2A coiled-coil and C2 domain containing 2A 216360 COACH syndrome

CC2D2A coiled-coil and C2 domain containing 2A 612285 Joubert syndrome 9
CC2D2A coiled-coil and C2 domain containing 2A 612284 " Meckel syndrome, type 6"
CCND1 cyclin D1 193300 " von Hippel-Lindau disease, modification of"
CD151 CD151 molecule (Raph blood group) 609057 Nephropathy with pretibial epidermolysis bullosa and deafness
CD320 CD320 molecule 613646 Methylmalonic aciduria due to transcobalamin receptor defect
CD36 CD36 molecule (thrombospondin receptor) 608404 Platelet glycoprotein IV deficiency
CD59 "CD59 molecule, complement regulatory protein" 612300 CD59 deficiency
CD81 CD81 molecule 613496 " Immunodeficiency, common variable, 6"
CDH23 cadherin-related 23 601386 " Deafness, autosomal recessive 12"
CDH23 cadherin-related 23 601067 " Usher syndrome, type 1D"
CDH23 cadherin-related 23 601067 " Usher syndrome, type 1D/F digenic"
CDHR1 cadherin-related family member 1 613660 Cone-rod dystrophy 15
CDK4 cyclin-dependent kinase 4 609048 Melanoma
CDK5RAP2 CDK5 regulatory subunit associated protein 2 604804 " Microcephaly, primary autosomal recessive, 3"
CDKN1B "cyclin-dependent kinase inhibitor 1B (p27, Kip1)" 610755 " Multiple endocrine neoplasia, type IV"
CDKN1C "cyclin-dependent kinase inhibitor 1C (p57, Kip2)" 130650 Beckwith-Wiedemann syndrome
CDKN2A cyclin-dependent kinase inhibitor 2A 151623 Li-Fraumeni syndrome
CDKN2A cyclin-dependent kinase inhibitor 2A 155755 Melanoma and neural system tumor syndrome
CDKN2A cyclin-dependent kinase inhibitor 2A 155601 " Melanoma, cutaneous malignant, 2"
CDKN2A cyclin-dependent kinase inhibitor 2A 606719 Pancreatic cancer/melanoma syndrome
CEL carboxyl ester lipase (bile salt-stimulated lipase) 609812 " Maturity-onset diabetes of the young, type VIII"
CEP290 centrosomal protein 290kDa 209900 Bardet-Biedl syndrome 14
CEP290 centrosomal protein 290kDa 610188 Joubert syndrome 5
CEP290 centrosomal protein 290kDa 611755 Leber congenital amaurosis 10
CEP290 centrosomal protein 290kDa 611134 Meckel syndrome type 4
CEP290 centrosomal protein 290kDa 610189 Senior-Loken syndrome 6
CFC1 "cripto, FRL-1, cryptic family 1" 217095 Double outlet right ventricle
CFC1 "cripto, FRL-1, cryptic family 1" 605376 " Heterotaxy, visceral, 2, autosomal"
CFTR "cystic fibrosis transmembrane conductance regulator (ATP-binding cassette sub-family C, member 7)" 277180 Congenital bilateral absence of vas deferens
CFTR "cystic fibrosis transmembrane conductance regulator (ATP-binding cassette sub-family C, member 7)" 219700 Cystic fibrosis
CHAT choline O-acetyltransferase 254210 " Myasthenic syndrome, congenital, associated with episodic apnea"
CHD7 chromodomain helicase DNA binding protein 7 214800 CHARGE syndrome
CHD7 chromodomain helicase DNA binding protein 7 146110 Hypogonadotropic hypogonadism
CHD7 chromodomain helicase DNA binding protein 7 612370 Kallmann syndrome 5
CHM choroideremia (Rab escort protein 1) 303100 Choroideremia
CHN1 chimerin (chimaerin) 1 604356 Duane retraction syndrome 2
CHRNB2 "cholinergic receptor, nicotinic, beta 2 (neuronal)" 605375 " Epilepsy, nocturnal frontal lobe, 3"
CHUK conserved helix-loop-helix ubiquitous kinase 613630 Cocoon syndrome
CISD2 CDGSH iron sulfur domain 2 604928 Wolfram syndrome 2
CLCN1 "chloride channel, voltage-sensitive 1" 160800 " Myotonia congenita, dominant"
CLCN1 "chloride channel, voltage-sensitive 1" 255700 " Myotonia congenita, recessive"
CLCN7 "chloride channel, voltage-sensitive 7" 166600 " Osteopetrosis, autosomal dominant 2"
CLCN7 "chloride channel, voltage-sensitive 7" 611490 " Osteopetrosis, autosomal recessive 4"
CLN3 "ceroid-lipofuscinosis, neuronal 3" 204200 " Ceroid lipofuscinosis, neuronal 3, juvenile"
CLN5 "ceroid-lipofuscinosis, neuronal 5" 256731 " Ceroid-lipofuscinosis, neuronal-5, variant late infantile"
CLN6 "ceroid-lipofuscinosis, neuronal 6, late infantile, variant" 601780 " Ceroid-lipofuscinosis, neuronal-6, variant late infantile"
CLN8 "ceroid-lipofuscinosis, neuronal 8 (epilepsy, progressive with mental retardation)" 600143 " Ceroid lipofuscinosis, neuronal 8"
CLN8 "ceroid-lipofuscinosis, neuronal 8 (epilepsy, progressive with mental retardation)" 610003 " Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant"

CLRN1 clarin 1 276902 " Usher syndrome, type 3"
CNGA3 cyclic nucleotide gated channel alpha 3 216900 Achromatopsia-2
CNGB1 cyclic nucleotide gated channel beta 1 268000 Retinitis pigmentosa-45
CNGB3 cyclic nucleotide gated channel beta 3 262300 Achromatopsia-3
CNGB3 cyclic nucleotide gated channel beta 3 248200 Macular degeneration juvenile
CNTN1 contactin 1 612540 " Myopathy, congenital, Compton-North"
COCH "coagulation factor C homolog, cochlin (Limulus polyphemus)" 601369 " Deafness, autosomal dominant 9"
COG7 component of oligomeric golgi complex 7 608779 " Congenital disorder of glycosylation, type IIe"
COL11A1 "collagen, type XI, alpha 1" 154780 Marshall syndrome
COL11A1 "collagen, type XI, alpha 1" 604841 " Stickler syndrome, type II"
COL11A2 "collagen, type XI, alpha 2" 601868 " Deafness, autosomal dominant 13"
COL11A2 "collagen, type XI, alpha 2" 609706 " Deafness, autosomal recessive 53"
COL11A2 "collagen, type XI, alpha 2" 215150 Otospondylomegaepiphyseal dysplasia
COL11A2 "collagen, type XI, alpha 2" 184840 " Stickler syndrome, type III"
COL11A2 "collagen, type XI, alpha 2" 277610 Weissenbacher-Zweymuller syndrome
COL18A1 "collagen, type XVIII, alpha 1" 267750 " Knobloch syndrome, type 1"
COL1A1 "collagen, type I, alpha 1" 114000 Caffey disease
COL1A1 "collagen, type I, alpha 1" 130000 " Ehlers-Danlos syndrome, type I"
COL1A1 "collagen, type I, alpha 1" 130060 " Ehlers-Danlos syndrome, type VIIA"
COL1A1 "collagen, type I, alpha 1" 166210 OI type II
COL1A1 "collagen, type I, alpha 1" 259420 OI type III
COL1A1 "collagen, type I, alpha 1" 166220 OI type IV
COL1A1 "collagen, type I, alpha 1" 166200 " Osteogenesis imperfecta, type I"
COL2A1 "collagen, type II, alpha 1" 200610 Achondrogenesis-hypochondrogenesis type 2
COL2A1 "collagen, type II, alpha 1" 132450 " Epiphyseal dysplasia, multiple, with myopia and deafness"
COL2A1 "collagen, type II, alpha 1" 156550 Kniest dysplasia
COL2A1 "collagen, type II, alpha 1" 604864 Osteoarthritis with mild chondrodysplasia
COL2A1 "collagen, type II, alpha 1" 183900 SED congenita
COL2A1 "collagen, type II, alpha 1" 184250 " SMED, Strudwick type"
COL2A1 "collagen, type II, alpha 1" 271700 Spondyloperipheral dysplasia
COL2A1 "collagen, type II, alpha 1" 108300 " Stickler syndrome, type I"
COL4A1 "collagen, type IV, alpha 1" 611773 " Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps"
COL4A1 "collagen, type IV, alpha 1" 607595 Brain small vessel disease with Axenfeld-Rieger anomaly
COL4A1 "collagen, type IV, alpha 1" 607595 Brain small vessel disease with hemorrhage
COL4A1 "collagen, type IV, alpha 1" 175780 Porencephaly
COL4A3 "collagen, type IV, alpha 3 (Goodpasture antigen)" 203780 " Alport syndrome, autosomal recessive"
COL4A3 "collagen, type IV, alpha 3 (Goodpasture antigen)" 141200 " Hematuria, benign familial"
COL4A4 "collagen, type IV, alpha 4" 203780 " Alport syndrome, autosomal recessive"
COL4A5 "collagen, type IV, alpha 5" 301050 Alport syndrome
COL5A1 "collagen, type V, alpha 1" 130000 " Ehlers-Danlos syndrome, type I"
COL5A1 "collagen, type V, alpha 1" 130010 " Ehlers-Danlos syndrome, type II"
COL5A2 "collagen, type V, alpha 2" 130000 " Ehlers-Danlos syndrome, type I"
COL8A2 "collagen, type VIII, alpha 2" 609140 " Corneal dystrophy polymorphous posterior, 2"
COL8A2 "collagen, type VIII, alpha 2" 136800 " Corneal dystrophy, Fuchs endothelial, 1"
COL9A2 "collagen, type IX, alpha 2" 600204 " Epiphyseal dysplasia, multiple, 2"
COL9A3 "collagen, type IX, alpha 3" 600969 " Epiphyseal dysplasia, multiple, 3"
COX15 "COX15 homolog, cytochrome c oxidase assembly protein (yeast)" 256000 Leigh syndrome due to cytochrome c oxidase deficiency
CP ceruloplasmin (ferroxidase) 604290 Cerebellar ataxia
CP ceruloplasmin (ferroxidase) 604290 " Hemosiderosis, systemic, due to aceruloplasminemia"
CRB1 crumbs homolog 1 (Drosophila) 172870 Pigmented paravenous chorioretinal atrophy

CRB1 crumbs homolog 1 (Drosophila) 600105 " Retinitis pigmentosa-12, autosomal recessive"
CRX cone-rod homeobox 120970 Cone-rod retinal dystrophy-2
CRX cone-rod homeobox 268000 " Retinitis pigmentosa, late-onset dominant"
CRYAB "crystallin, alpha B" 608810 " Myopathy, cardioskeletal, desmin-related, with cataract"
CRYBA1 "crystallin, beta A1" 600881 " Cataract, congenital zonular, with sutural opacities"
CRYBA4 "crystallin, beta A4" 610425 " Cataract, lamellar 2"
CRYBA4 "crystallin, beta A4" 610426 " Microphthalmia, isolated, with cataract 4"
CRYBB1 "crystallin, beta B1" 611544 " Cataract, congenital nuclear, autosomal recessive 3"
CRYBB2 "crystallin, beta B2" 601547 " Cataract, cerulean, type 2"
CRYBB2 "crystallin, beta B2" 604307 " Cataract, Coppock-like"
CRYBB2 "crystallin, beta B2" 607133 " Cataract, sutural, with punctate and cerulean opacities"
CRYBB3 "crystallin, beta B3" 609741 " Cataract, congenital nuclear, 2"
CRYGC "crystallin, gamma C" 604307 " Cataract, Coppock-like"
CRYGD "crystallin, gamma D" 608983 " Cataract, congenital, cerulean type, 3"
CRYGD "crystallin, gamma D" 115700 " Cataract, crystalline aculeiform"
CRYGD "crystallin, gamma D" 601286 " Cataract, nonnuclear polymorphic congenital"
CST3 cystatin C 105150 Cerebral amyloid angiopathy
CST3 cystatin C 611953 " Macular degeneration, age-related, 11"
CSTB cystatin B (stefin B) 254800 " Epilepsy, progressive myoclonic 1"
CTNNB1 "catenin (cadherin-associated protein), beta 1, 88kDa" 114550 Hepatocellular carcinoma
CTNNB1 "catenin (cadherin-associated protein), beta 1, 88kDa" 167000 Ovarian cancer
CTNNB1 "catenin (cadherin-associated protein), beta 1, 88kDa" 132600 Pilomatricoma
CTNS "cystinosin, lysosomal cystine transporter" 219900 " Cystinosis, late-onset juvenile or adolescent nephropathic"
CTNS "cystinosin, lysosomal cystine transporter" 219800 " Cystinosis, nephropathic"
CTNS "cystinosin, lysosomal cystine transporter" 219750 " Cystinosis, ocular nonnephropathic"
CTSD cathepsin D 610127 " Ceroid lipofuscinosis, neuronal, 10"
CXCR4 chemokine (C-X-C motif) receptor 4 193670 WHIM syndrome
CYCS "cytochrome c, somatic" 612004 Thrombocytopenia 4
CYP1B1 "cytochrome P450, family 1, subfamily B, polypeptide 1" 231300 " Glaucoma 3A, primary congenital"
CYP1B1 "cytochrome P450, family 1, subfamily B, polypeptide 1" 137760 " Glaucoma, primary open angle, adult-onset"
CYP1B1 "cytochrome P450, family 1, subfamily B, polypeptide 1" 137750 " Glaucoma, primary open angle, juvenile-onset"
CYP1B1 "cytochrome P450, family 1, subfamily B, polypeptide 1" 604229 Peters anomaly
CYP4V2 "cytochrome P450, family 4, subfamily V, polypeptide 2" 210370 Bietti crystalline corneoretinal dystrophy
DBH dopamine beta-hydroxylase (dopamine beta-monooxygenase) 223360 Dopamine beta-hydroxylase deficiency
DCC deleted in colorectal carcinoma 157600 " Mirror movements, congenital"
DCN decorin 610048 " Corneal dystrophy, congenital stromal"
DDC dopa decarboxylase (aromatic L-amino acid decarboxylase) 608643 Aromatic L-amino acid decarboxylase deficiency
DDR2 discoidin domain receptor tyrosine kinase 2 271665 " Spondylometaepiphyseal dysplasia, short limb-hand type"
DLL3 delta-like 3 (Drosophila) 277300 " Spondylocostal dysostosis, autosomal recessive, 1"
DMD dystrophin 300376 Becker muscular dystrophy
DMD dystrophin 302045 " Cardiomyopathy, dilated, 3B"
DMD dystrophin 310200 Duchenne muscular dystrophy
DMPK dystrophia myotonica-protein kinase 160900 Myotonic dystrophy
DNMT3B DNA (cytosine-5-)-methyltransferase 3 beta 242860 Immunodeficiency-centromeric instability-facial anomalies syndrome
DTNBP1 dystrobrevin binding protein 1 203300 Hermansky-Pudlak syndrome 7
DYNC2H1 "dynein, cytoplasmic 2, heavy chain 1" 613091 Asphyxiating thoracic dystrophy 3
DYNC2H1 "dynein, cytoplasmic 2, heavy chain 1" 263510 " Short rib-polydactyly syndrome, type III"
EDARADD EDAR-associated death domain 129490 " Ectodermal dysplasia, anhidrotic, autosomal dominant"
EDARADD EDAR-associated death domain 224900 " Ectodermal dysplasia, anhidrotic, autosomal recessive"
EDNRB endothelin receptor type B 600501 ABCD syndrome
EDNRB endothelin receptor type B 277580 Waardenburg syndrome type 4A

EFEMP1 EGF containing fibulin-like extracellular matrix protein 1 126600 Doyne honeycomb degeneration of retina
EFNB1 ephrin-B1 304110 Craniofrontonasal dysplasia
EGF epidermal growth factor 611718 " Hypomagnesemia 4, renal"
EGFR epidermal growth factor receptor 211980 " Adenocarcinoma of lung, response to tyrosine kinase inhibitor in"
EGFR epidermal growth factor receptor 211980 " Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in"
EGR2 early growth response 2 607678 Charcot-Marie-Tooth disease type 1D
EGR2 early growth response 2 145900 Dejerine-Sottas neuropathy
EGR2 early growth response 2 605253 " Neuropathy, congenital hypomyelinating, 1"
ELANE "elastase, neutrophil expressed" 162800 " Hematopoiesis, cyclic"
ELANE "elastase, neutrophil expressed" 202700 " Neutropenia, severe congenital, autosomal dominant 1"
ELN elastin 123700 " Cutis laxa, AD"
ELN elastin 185500 Supravalvar aortic stenosis
ELOVL4 ELOVL fatty acid elongase 4 600110 " Macular dystrophy, autosomal dominant, chromosome 6-linked"
ELOVL4 ELOVL fatty acid elongase 4 600110 Stargardt disease 3
EMX2 empty spiracles homeobox 2 269160 Schizencephaly
EP300 E1A binding protein p300 114500 Colorectal cancer
EP300 E1A binding protein p300 180849 Rubinstein-Taybi syndrome
EPAS1 endothelial PAS domain protein 1 611783 " Erythrocytosis, familial, 4"
EPHA2 EPH receptor A2 613020 " Cataract, posterior polar, 1"
EPHB2 EPH receptor B2 603688 " Prostate cancer, progression and metastasis of"
ERCC1 "excision repair cross-complementing rodent repair deficiency, complementation group 1 (includes overlapping antisense sequence)" 610758 Cerebrooculofacioskeletal syndrome 4
ERCC2 "excision repair cross-complementing rodent repair deficiency, complementation group 2" 610756 Cerebrooculofacioskeletal syndrome 2
ERCC2 "excision repair cross-complementing rodent repair deficiency, complementation group 2" 601675 Trichothiodystrophy
ERCC2 "excision repair cross-complementing rodent repair deficiency, complementation group 2" 278730 Xeroderma pigmentosum group D
ERCC6 "excision repair cross-complementing rodent repair deficiency, complementation group 6" 214150 Cerebrooculofacioskeletal syndrome 1
ERCC6 "excision repair cross-complementing rodent repair deficiency, complementation group 6" 133540 Cockayne syndrome type B
ERCC6 "excision repair cross-complementing rodent repair deficiency, complementation group 6" 278800 De Sanctis-Cacchione syndrome
ERCC6 "excision repair cross-complementing rodent repair deficiency, complementation group 6" 600630 UV-sensitive syndrome
ERCC8 "excision repair cross-complementing rodent repair deficiency, complementation group 8" 216400 Cockayne syndrome type A
EXT1 exostosin 1 215300 Chondrosarcoma
EXT1 exostosin 1 133700 " Exostoses, multiple, type 1"
EYA1 eyes absent homolog 1 (Drosophila) 113650 Branchiootorenal syndrome with cataract
EYA1 eyes absent homolog 1 (Drosophila) 113650 Branchiootorenal syndrome
EYA1 eyes absent homolog 1 (Drosophila) 166780 Otofaciocervical syndrome
EYA4 eyes absent homolog 4 (Drosophila) 605362 " Cardiomyopathy, dilated, 1J"
EYA4 eyes absent homolog 4 (Drosophila) 601316 " Deafness, autosomal dominant 10"
EYS eyes shut homolog (Drosophila) 602772 Retinitis pigmentosa-25
F5 "coagulation factor V (proaccelerin, labile factor)" 227400 Factor V deficiency
FA2H fatty acid 2-hydroxylase 612443 " Leukodystrophy, dysmyelinating, and spastic paraparesis with or without dystonia"
FAM20C "family with sequence similarity 20, member C" 259775 Raine syndrome
FANCA "Fanconi anemia, complementation group A" 227650 " Fanconi anemia, complementation group A"
FBLN5 fibulin 5 123700 " Cutis laxa, autosomal dominant"
FBLN5 fibulin 5 219100 " Cutis laxa, autosomal recessive"
FBLN5 fibulin 5 608895 " Macular degeneration, age-related, 3"

FBN1 fibrillin 1 129600 " Ectopia lentis, familial"
FBN1 fibrillin 1 154700 Marfan syndrome
FBN1 fibrillin 1 604308 MASS syndrome
FBN1 fibrillin 1 182212 Shprintzen-Goldberg syndrome
FBN1 fibrillin 1 184900 Stiff skin syndrome
FBN1 fibrillin 1 608328 " Weill-Marchesani syndrome, dominant"
FBN2 fibrillin 2 121050 " Contractural arachnodactyly, congenital"
FGA fibrinogen alpha chain 202400 " Afibrinogenemia, congenital"
FGA fibrinogen alpha chain 105200 " Amyloidosis, hereditary renal"
FGF10 fibroblast growth factor 10 180920 Aplasia of lacrimal and salivary glands
FGF10 fibroblast growth factor 10 149730 LADD syndrome
FGF14 fibroblast growth factor 14 609307 Spinocerebellar ataxia-27
FGF8 fibroblast growth factor 8 (androgen-induced) 612702 Kallmann syndrome 6
FGF9 fibroblast growth factor 9 (glia-activating factor) 612961 Multiple synostoses syndrome 3
FGFR1 fibroblast growth factor receptor 1 146110 Hypogonadotropic hypogonadism
FGFR1 fibroblast growth factor receptor 1 123150 Jackson-Weiss syndrome
FGFR1 fibroblast growth factor receptor 1 147950 Kallmann syndrome 2
FGFR1 fibroblast growth factor receptor 1 166250 Osteoglophonic dysplasia
FGFR1 fibroblast growth factor receptor 1 101600 Pfeiffer syndrome
FGFR1 fibroblast growth factor receptor 1 190440 Trigonocephaly
FGFR2 fibroblast growth factor receptor 2 207410 Antley-Bixler syndrome
FGFR2 fibroblast growth factor receptor 2 101200 Apert syndrome
FGFR2 fibroblast growth factor receptor 2 123790 Beare-Stevenson cutis gyrata syndrome
FGFR2 fibroblast growth factor receptor 2 123500 Crouzon syndrome
FGFR2 fibroblast growth factor receptor 2 137215 " Gastric cancer, somatic"
FGFR2 fibroblast growth factor receptor 2 123150 Jackson-Weiss syndrome
FGFR2 fibroblast growth factor receptor 2 101600 Pfeiffer syndrome
FGFR2 fibroblast growth factor receptor 2 101400 Saethre-Chotzen syndrome
FGFR3 fibroblast growth factor receptor 3 100800 Achondroplasia
FGFR3 fibroblast growth factor receptor 3 109800 " Bladder cancer, somatic"
FGFR3 fibroblast growth factor receptor 3 610474 CATSHL syndrome
FGFR3 fibroblast growth factor receptor 3 603956 " Cervical cancer, somatic"
FGFR3 fibroblast growth factor receptor 3 109800 " Colorectal cancer, somatic"
FGFR3 fibroblast growth factor receptor 3 612247 Crouzon syndrome with acanthosis nigricans
FGFR3 fibroblast growth factor receptor 3 146000 Hypochondroplasia
FGFR3 fibroblast growth factor receptor 3 149730 LADD syndrome
FGFR3 fibroblast growth factor receptor 3 602849 Muenke syndrome
FGFR3 fibroblast growth factor receptor 3 162900 " Nevus, keratinocytic, nonepidermolytic"
FGFR3 fibroblast growth factor receptor 3 187600 " Thanatophoric dysplasia, type I"
FKTN fukutin 611615 " Cardiomyopathy, dilated, 1X"
FLNA "filamin A, alpha" 300321 FG syndrome 2
FLNA "filamin A, alpha" 305620 Frontometaphyseal dysplasia
FLNA "filamin A, alpha" 300049 " Heterotopia, periventricular"
FLNA "filamin A, alpha" 300537 " Heterotopia, periventricular, ED variant"
FLNA "filamin A, alpha" 300048 " Intestinal pseudoobstruction, neuronal"
FLNA "filamin A, alpha" 309350 Melnick-Needles syndrome
FLNA "filamin A, alpha" 311300 " Otopalatodigital syndrome, type I"
FLNA "filamin A, alpha" 304120 " Otopalatodigital syndrome, type II"
FLT4 fms-related tyrosine kinase 4 602089 " Hemangioma, capillary infantile, somatic"
FLT4 fms-related tyrosine kinase 4 153100 " Lymphedema, hereditary I"
FMR1 fragile X mental retardation 1 300624 Fragile X syndrome

FMR1 fragile X mental retardation 1 300623 Fragile X tremor/ataxia syndrome
FN1 fibronectin 1 601894 Glomerulopathy with fibronectin deposits 2
FOLR1 folate receptor 1 (adult) 613068 Neurodegeneration due to cerebral folate transport deficiency
FOXC1 forkhead box C1 602482 " Axenfeld-Rieger syndrome, type 3"
FOXC1 forkhead box C1 601631 " Iridogoniodysgenesis, type 1"
FOXC1 forkhead box C1 601631 Iris hypoplasia and glaucoma
FOXC1 forkhead box C1 602482 Rieger or Axenfeld anomalies
FOXC2 "forkhead box C2 (MFH-1, mesenchyme forkhead 1)" 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus
FOXC2 "forkhead box C2 (MFH-1, mesenchyme forkhead 1)" 153400 Lymphedema-distichiasis syndrome
FOXE1 forkhead box E1 (thyroid transcription factor 2) 241850 Bamforth-Lazarus syndrome
FOXE3 forkhead box E3 107250 Anterior segment mesenchymal dysgenesis
FOXE3 forkhead box E3 610256 " Aphakia, congenital primary"
FOXL2 forkhead box L2 110100 " Blepharophimosis, epicanthus inversus, and ptosis, type 1"
FOXL2 forkhead box L2 110100 " Blepharophimosis, epicanthus inversus, and ptosis, type 2"
FOXL2 forkhead box L2 608996 Premature ovarian failure 3
FOXP2 forkhead box P2 602081 Speech-language disorder-1
FOXP3 forkhead box P3 304790 " Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked"
FRAS1 Fraser syndrome 1 219000 Fraser syndrome
FREM1 FRAS1 related extracellular matrix 1 608980 Bifid nose with or without anorectal and renal anomalies
FREM2 FRAS1 related extracellular matrix protein 2 219000 Fraser syndrome
FRMD7 FERM domain containing 7 310700 " Nystagmus 1, congenital, X-linked"
FSCN2 "fascin homolog 2, actin-bundling protein, retinal (Strongylocentrotus purpuratus)" 607921 Retinitis pigmentosa-30
FZD4 frizzled family receptor 4 133780 Exudative vitreoretinopathy
FZD4 frizzled family receptor 4 133780 Retinopathy of prematurity
GAD1 "glutamate decarboxylase 1 (brain, 67kDa)" 603513 " Cerebral palsy, spastic, symmetric, autosomal recessive"
GALC galactosylceramidase 245200 Krabbe disease
GALK1 galactokinase 1 230200 Galactokinase deficiency with cataracts
GALNS galactosamine (N-acetyl)-6-sulfate sulfatase 253000 Mucopolysaccharidosis IVA
GATA1 GATA binding protein 1 (globin transcription factor 1) 300367 Dyserythropoietic anemia with thrombocytopenia
GATA1 GATA binding protein 1 (globin transcription factor 1) 190685 " Leukemia, megakaryoblastic, of Down syndrome"
GATA1 GATA binding protein 1 (globin transcription factor 1) 190685 " Leukemia, megakaryoblastic, with or without Down syndrome"
GATA1 GATA binding protein 1 (globin transcription factor 1) 300367 Macrothrombocytopenia
GATA1 GATA binding protein 1 (globin transcription factor 1) 314050 " Thrombocytopenia with beta-thalassemia, X-linked"
GATA3 GATA binding protein 3 146255 " Hypoparathyroidism, sensorineural deafness, and renal dysplasia"
GATA4 GATA binding protein 4 607941 Atrial septal defect-2
GCNT2 "glucosaminyl (N-acetyl) transferase 2, I-branching enzyme (I blood group)" 110800 Adult i phenotype with congenital cataract
GCNT2 "glucosaminyl (N-acetyl) transferase 2, I-branching enzyme (I blood group)" 110800 Adult i phenotype without cataract
GDF6 growth differentiation factor 6 118100 " Klippel-Feil syndrome, autosomal dominant"
GDF6 growth differentiation factor 6 613094 " Microphthalmia, isolated 4"
GDF6 growth differentiation factor 6 122600 " Spondylocostal dystostosis 4, autosomal dominant"
GDNF glial cell derived neurotrophic factor 209880 Central hypoventilation syndrome
GDNF glial cell derived neurotrophic factor 142623 Hirschsprung disease
GFAP glial fibrillary acidic protein 203450 Alexander disease
GH1 growth hormone 1 262400 " Growth hormone deficiency, isolated, type IA"
GH1 growth hormone 1 612781 " Growth hormone deficiency, isolated, type IB"
GH1 growth hormone 1 173100 " Growth hormone deficiency, isolated, type II"
GH1 growth hormone 1 262650 Kowarski syndrome
GJA1 "gap junction protein, alpha 1, 43kDa" 600309 Atrioventricular septal defect

GJA1 "gap junction protein, alpha 1, 43kDa" 234100 Hallermann-Streiff syndrome
GJA1 "gap junction protein, alpha 1, 43kDa" 241550 Hypoplastic left heart syndrome
GJA1 "gap junction protein, alpha 1, 43kDa" 164200 Oculodentodigital dysplasia
GJA1 "gap junction protein, alpha 1, 43kDa" 257850 " Oculodentodigital dysplasia, autosomal recessive"
GJA1 "gap junction protein, alpha 1, 43kDa" 186100 " Syndactyly, type III"
GJA3 "gap junction protein, alpha 3, 46kDa" 601885 " Cataract, zonular pulverulent-3"
GJA8 "gap junction protein, alpha 8, 50kDa" 116200 " Cataract, zonular pulverulent-1"
GJA8 "gap junction protein, alpha 8, 50kDa" 116150 Cataract-microcornea syndrome
GJB1 "gap junction protein, beta 1, 32kDa" 302800 " Charcot-Marie-Tooth neuropathy, X-linked dominant, 1"
GJB2 "gap junction protein, beta 2, 26kDa" 149200 Bart-Pumphrey syndrome
GJB2 "gap junction protein, beta 2, 26kDa" 601544 " Deafness, autosomal dominant 3A"
GJB2 "gap junction protein, beta 2, 26kDa" 220290 " Deafness, autosomal recessive 1A"
GJB2 "gap junction protein, beta 2, 26kDa" 602540 Hystrix-like ichthyosis with deafness
GJB2 "gap junction protein, beta 2, 26kDa" 148210 Keratitis-ichthyosis-deafness syndrome
GJB2 "gap junction protein, beta 2, 26kDa" 148350 " Keratoderma, palmoplantar, with deafness"
GJB2 "gap junction protein, beta 2, 26kDa" 124500 Vohwinkel syndrome
GJC2 "gap junction protein, gamma 2, 47kDa" 608804 " Leukodystrophy, hypomyelinating, 2"
GJC2 "gap junction protein, gamma 2, 47kDa" 613480 " Lymphedema, hereditary, IC"
GJC2 "gap junction protein, gamma 2, 47kDa" 613206 " Spastic paraplegia, 44"
GK glycerol kinase 307030 Glycerol kinase deficiency
GLB1 "galactosidase, beta 1" 230500 " GM1-gangliosidosis, type I"
GLB1 "galactosidase, beta 1" 230600 " GM1-gangliosidosis, type II"
GLB1 "galactosidase, beta 1" 230650 " GM1-gangliosidosis, type III"
GLB1 "galactosidase, beta 1" 253010 Morquio syndrome B
GLI2 GLI family zinc finger 2 610829 Holoprosencephaly-9
GLI3 GLI family zinc finger 3 200990 Acrocallosal syndrome
GLI3 GLI family zinc finger 3 175700 Greig cephalopolysyndactyly syndrome
GLI3 GLI family zinc finger 3 146510 Pallister-Hall syndrome
GLI3 GLI family zinc finger 3 174200 " Polydactyly, postaxial, types A1 and B"
GLI3 GLI family zinc finger 3 174700 " Polydactyly, preaxial, type IV"
GLIS3 GLIS family zinc finger 3 610199 " Diabetes mellitus, neonatal, with congenital hypothyroidism"
GLRA1 "glycine receptor, alpha 1" 149400 " Startle disease/hyperekplexia, autosomal dominant"
GLUL glutamate-ammonia ligase 610015 " Glutamine deficiency, congenital"
GNAT1 "guanine nucleotide binding protein (G protein), alpha transducing activity polypeptide 1" 610444 " Night blindness, congenital stationary, autosomal dominant 3"
GNPAT glyceronephosphate O-acyltransferase 222765 " Chondrodysplasia punctata, rhizomelic, type 2"
GNPTAB "N-acetylglucosamine-1-phosphate transferase, alpha and beta subunits" 252500 Mucolipidosis II alpha/beta
GNPTAB "N-acetylglucosamine-1-phosphate transferase, alpha and beta subunits" 252600 Mucolipidosis III alpha/beta
GPHN gephyrin 149400 Hyperekplexia
GPHN gephyrin 252150 " Molybdenum cofactor deficiency, type C"
GPR143 G protein-coupled receptor 143 300814 " Nystagmus 6, congenital, X-linked"
GPR143 G protein-coupled receptor 143 300500 " Ocular albinism, type I, Nettleship-Falls type"
GPR98 G protein-coupled receptor 98 604352 " Convulsions, familial febrile, 4"
GPR98 G protein-coupled receptor 98 605472 " Usher syndrome, type IIC"
GRHL2 grainyhead-like 2 (Drosophila) 608641 " Deafness, autosomal dominant 28"
GRIA3 "glutamate receptor, ionotropic, AMPA 3" 300699 " Mental retardation, X-linked 94"
GRK1 G protein-coupled receptor kinase 1 613411 Oguchi disease-2
GRM6 "glutamate receptor, metabotropic 6" 257270 " Night blindness, congenital stationary, type 1B"
GUCA1A guanylate cyclase activator 1A (retina) 602093 Cone dystrophy-3
GUCA1A guanylate cyclase activator 1A (retina) 602093 Cone-rod dystrophy 14
GUCY2D "guanylate cyclase 2D, membrane (retina-specific)" 601777 Cone-rod dystrophy

GUCY2D "guanylate cyclase 2D, membrane (retina-specific)" 204000 Leber congenital amaurosis 1
H6PD hexose-6-phosphate dehydrogenase (glucose 1-dehydrogenase) 604931 Cortisone reductase deficiency
HADHA "hydroxyacyl-CoA dehydrogenase/3-ketoacyl-CoA thiolase/enoyl-CoA hydratase (trifunctional protein), alpha subunit" 609016 " Fatty liver, acute, of pregnancy"
HADHA "hydroxyacyl-CoA dehydrogenase/3-ketoacyl-CoA thiolase/enoyl-CoA hydratase (trifunctional protein), alpha subunit" 609016 " HELLP syndrome, maternal, of pregnancy"
HADHA "hydroxyacyl-CoA dehydrogenase/3-ketoacyl-CoA thiolase/enoyl-CoA hydratase (trifunctional protein), alpha subunit" 609016 LCHAD deficiency
HADHA "hydroxyacyl-CoA dehydrogenase/3-ketoacyl-CoA thiolase/enoyl-CoA hydratase (trifunctional protein), alpha subunit" 609015 Trifunctional protein deficiency
HCCS holocytochrome c synthase 309801 " Microphthalmia, syndromic 7"
HCRT hypocretin (orexin) neuropeptide precursor 161400 Narcolepsy 1
HESX1 HESX homeobox 1 182230 Growth hormone deficiency with pituitary anomalies
HESX1 HESX homeobox 1 182230 " Pituitary hormone deficiency, combined, 5"
HESX1 HESX homeobox 1 182230 Septo-optic dysplasia
HEXA hexosaminidase A (alpha polypeptide) 272800 " GM2-gangliosidosis, several forms"
HEXA hexosaminidase A (alpha polypeptide) 272800 Tay-Sachs disease
HEXB hexosaminidase B (beta polypeptide) 268800 " Sandhoff disease, infantile, juvenile, and adult forms"
HGF hepatocyte growth factor (hepapoietin A; scatter factor) 608265 " Deafness, autosomal recessive 39"
HMX1 H6 family homeobox 1 612109 Oculoauricular syndrome
HNF1A HNF1 homeobox A 612520 " Diabetes mellitus, insulin-dependent, 20"
HNF1A HNF1 homeobox A 142330 Hepatic adenoma
HNF1A HNF1 homeobox A 600496 " MODY, type III"
HNF1A HNF1 homeobox A 144700 Renal cell carcinoma
HOXA13 homeobox A13 176305 Guttmacher syndrome
HOXA13 homeobox A13 140000 Hand-foot-uterus Syndrome
HOXD13 homeobox D13 113200 Brachydactyly type D
HOXD13 homeobox D13 113300 Brachydactyly type E
HOXD13 homeobox D13 610713 Brachydactyly-syndactyly syndrome
HOXD13 homeobox D13 186300 " Syndactyly, type V"
HOXD13 homeobox D13 186000 Synpolydactyly with foot anomalies
HOXD13 homeobox D13 186000 " Synpolydactyly, type II"
HOXD13 homeobox D13 192350 VACTERL association
HPRT1 hypoxanthine phosphoribosyltransferase 1 300323 HPRT-related gout
HPRT1 hypoxanthine phosphoribosyltransferase 1 300322 Lesch-Nyhan syndrome
HPS1 Hermansky-Pudlak syndrome 1 203300 Hermansky-Pudlak syndrome 1
HPS3 Hermansky-Pudlak syndrome 3 203300 Hermansky-Pudlak syndrome 3
HPS4 Hermansky-Pudlak syndrome 4 203300 Hermansky-Pudlak syndrome 4
HPS5 Hermansky-Pudlak syndrome 5 203300 Hermansky-Pudlak syndrome 5
HPS6 Hermansky-Pudlak syndrome 6 203300 Hermansky-Pudlak syndrome 6
HR hairless homolog (mouse) 203655 Alopecia universalis
HR hairless homolog (mouse) 209500 Atrichia with papular lesions
HR hairless homolog (mouse) 146550 " Hypotrichosis, hereditary, Marie Unna type, 1"
HSD11B1 hydroxysteroid (11-beta) dehydrogenase 1 604931 Cortisone reductase deficiency
HSD17B10 hydroxysteroid (17-beta) dehydrogenase 10 300438 17-beta-hydroxysteroid dehydrogenase X deficiency
HSD17B10 hydroxysteroid (17-beta) dehydrogenase 10 300705 " Mental retardation, X-linked 17/31, microduplication"
HSD17B10 hydroxysteroid (17-beta) dehydrogenase 10 300220 " Mental retardation, X-linked syndromic 10"
HSD17B4 hydroxysteroid (17-beta) dehydrogenase 4 261515 D-bifunctional protein deficiency
HSF4 heat shock transcription factor 4 116800 " Cataract, lamellar"
HSF4 heat shock transcription factor 4 116800 " Cataract, Marner type"
HSPD1 heat shock 60kDa protein 1 (chaperonin) 612233 " Leukodystrophy, hypomyelinating, 4"

HSPD1 heat shock 60kDa protein 1 (chaperonin) 605280 Spastic paraplegia-13
HSPG2 heparan sulfate proteoglycan 2 224410 " Dyssegmental dysplasia, Silverman-Handmaker type"
HSPG2 heparan sulfate proteoglycan 2 255800 " Schwartz-Jampel syndrome, type 1"
IDUA "iduronidase, alpha-L-" 607014 Mucopolysaccharidosis Ih
IDUA "iduronidase, alpha-L-" 607015 Mucopolysaccharidosis Ih/s
IDUA "iduronidase, alpha-L-" 607016 Mucopolysaccharidosis Is
IFT122 intraflagellar transport 122 homolog (Chlamydomonas) 218330 Cranioectodermal dysplasia
IFT80 intraflagellar transport 80 homolog (Chlamydomonas) 611263 Asphyxiating thoracic dystrophy 2
IGF1 insulin-like growth factor 1 (somatomedin C) 608747 Growth retardation with deafness and mental retardation due to IGF1 deficiency
IGF1R insulin-like growth factor 1 receptor 270450 " Insulin-like growth factor I, resistance to"
IHH Indian hedgehog 607778 Acrocapitofemoral dysplasia
IHH Indian hedgehog 112500 Brachydactyly type A1
IKBKAP "inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein" 223900 " Dysautonomia, familial"
IKBKG "inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase gamma" 300291 " Ectodermal dysplasia, hypohidrotic, with immune deficiency"
IKBKG "inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase gamma" 300301 " Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency"
IKBKG "inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase gamma" 300584 " Immunodeficiency, isolated"
IKBKG "inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase gamma" 308300 " Incontinentia pigmenti, type II"
IL1RN interleukin 1 receptor antagonist 612852 Interleukin 1 receptor antagonist deficiency
IL2RA "interleukin 2 receptor, alpha" 606367 " Interleukin-2 receptor, alpha chain, deficiency of"
INPP5E "inositol polyphosphate-5-phosphatase, 72 kDa" 213300 Joubert syndrome 1
INPP5E "inositol polyphosphate-5-phosphatase, 72 kDa" 610156 " Mental retardation, truncal obesity, retinal dystrophy, and micropenis"
INS insulin 125852 " Diabetes mellitus, insulin-dependent, 2"
INS insulin 606176 " Diabetes mellitus, permanent neonatal"
INS insulin 125852 " Diabetes mellitus, type 1"
INS insulin 613370 " Maturity-onset diabetes of the young, type 10"
INSR insulin receptor 610549 " Diabetes mellitus, insulin-resistant, with acanthosis nigricans"
INSR insulin receptor 609968 " Hyperinsulinemic hypoglycemia, familial, 5"
INSR insulin receptor 246200 Leprechaunism
INSR insulin receptor 262190 Rabson-Mendenhall syndrome
IQCB1 IQ motif containing B1 609254 Senior-Loken syndrome 5
ITGA6 "integrin, alpha 6" 226730 " Epidermolysis bullosa, junctional, with pyloric stenosis"
ITGB2 "integrin, beta 2 (complement component 3 receptor 3 and 4 subunit)" 116920 Leukocyte adhesion deficiency
ITGB4 "integrin, beta 4" 131800 Epidermolysis bullosa of hands and feet
ITGB4 "integrin, beta 4" 226650 " Epidermolysis bullosa, junctional, non-Herlitz type"
ITGB4 "integrin, beta 4" 226730 " Epidermolysis bullosa, junctional, with pyloric atresia"
ITPR1 "inositol 1,4,5-trisphosphate receptor, type 1" 606658 Spinocerebellar ataxia 15
JAG1 jagged 1 118450 Alagille syndrome
JAG1 jagged 1 187500 Tetralogy of Fallot
KCNA1 "potassium voltage-gated channel, shaker-related subfamily, member 1 (episodic ataxia with myokymia)" 160120 Episodic ataxia/myokymia syndrome
KCNC3 "potassium voltage-gated channel, Shaw-related subfamily, member 3" 605259 Spinocerebellar ataxia-13
KCNJ10 "potassium inwardly-rectifying channel, subfamily J, member 10" 612780 SESAME syndrome
KCNJ13 "potassium inwardly-rectifying channel, subfamily J, member 13" 193230 Snowflake vitreoretinal degeneration
KCNMA1 "potassium large conductance calcium-activated channel, subfamily M, alpha member 1" 609446 Generalized epilepsy and paroxysmal dyskinesia
KCTD7 potassium channel tetramerisation domain containing 7 611726 " Epilepsy, progressive myoclonic 3"

KDR kinase insert domain receptor (a type III receptor tyrosine kinase) 602089 " Hemangioma, capillary infantile, somatic"
KERA keratocan 217300 " Cornea plana congenita, recessive"
KIF21A kinesin family member 21A 135700 " Fibrosis of extraocular muscles, congenital, 1"
KIF21A kinesin family member 21A 135700 " Fibrosis of extraocular muscles, congenital, 3B"
KIT v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog 606764 " Gastrointestinal stromal tumor, somatic"
KIT v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog 273300 Germ cell tumors
KIT v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog 601626 " Leukemia, acute myeloid"
KITLG KIT ligand 145250 " Hyperpigmentation, familial progressive"
KRAS v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog 109800 Bladder cancer
KRAS v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog 114480 " Breast cancer, somatic"
KRAS v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog 115150 Cardiofaciocutaneous syndrome
KRAS v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog 211980 Lung cancer
KRAS v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog 609942 Noonan syndrome 3
KRAS v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog 260350 " Pancreatic carcinoma, somatic"
KRAS v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog 137215 Stomach cancer
KRT12 keratin 12 122100 Meesmann corneal dystrophy
KRT14 keratin 14 125595 Dermatopathia pigmentosa reticularis
KRT14 keratin 14 131760 " Epidermolysis bullosa simplex, Dowling-Meara type"
KRT14 keratin 14 131900 " Epidermolysis bullosa simplex, Koebner type"
KRT14 keratin 14 601001 " Epidermolysis bullosa simplex, recessive"
KRT14 keratin 14 131800 " Epidermolysis bullosa simplex, Weber-Cockayne type"
KRT14 keratin 14 161000 Naegeli-Franceschetti-Jadassohn syndrome
KRT3 keratin 3 122100 Meesmann corneal dystrophy
KRT4 keratin 4 193900 White sponge nevus
L1CAM L1 cell adhesion molecule 304100 " Corpus callosum, partial agenesis of"
L1CAM L1 cell adhesion molecule 303350 CRASH syndrome
L1CAM L1 cell adhesion molecule 307000 Hydrocephalus due to aqueductal stenosis
L1CAM L1 cell adhesion molecule 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction
L1CAM L1 cell adhesion molecule 142623 Hydrocephalus with Hirschsprung disease and cleft palate
L1CAM L1 cell adhesion molecule 303350 MASA syndrome
LAMA2 "laminin, alpha 2" 607855 " Muscular dystrophy, congenital merosin-deficient"
LAMA2 "laminin, alpha 2" 607855 " Muscular dystrophy, congenital, due to partial LAMA2 deficiency"
LAMB2 "laminin, beta 2 (laminin S)" 609049 " Nephrosis, congenital, with or without ocular abnormalities"
LAMB2 "laminin, beta 2 (laminin S)" 609049 Pierson syndrome
LBR lamin B receptor 215140 Greenberg dysplasia
LBR lamin B receptor 169400 Pelger-Huet anomaly
LBR lamin B receptor 613471 Reynolds syndrome
LCA5 Leber congenital amaurosis 5 604537 Leber congenital amaurosis 5
LCAT lecithin-cholesterol acyltransferase 136120 Fish-eye disease
LCAT lecithin-cholesterol acyltransferase 245900 Norum disease
LDLR low density lipoprotein receptor 143890 " Hypercholesterolemia, familial"
LFNG LFNG O-fucosylpeptide 3-beta-N-acetylglucosaminyltransferase 609813 " Spondylocostal dysostosis, autosomal recessive 3"
LGI1 "leucine-rich, glioma inactivated 1" 600512 " Epilepsy, partial, with auditory features"
LMNA lamin A/C 115200 " Cardiomyopathy, dilated, 1A"
LMNA lamin A/C 605588 Charcot-Marie-Tooth disease type 2B1
LMNA lamin A/C 181350 " Emery-Dreifuss muscular dystrophy, AD"
LMNA lamin A/C 181350 " Emery-Dreifuss muscular dystrophy, AR"
LMNA lamin A/C 176670 Huchinson-Gilford Progeria
LMNA lamin A/C 151660 " Lipodystrophy, familial partial"
LMNA lamin A/C 248370 Mandibuloacral dysplasia
LMNA lamin A/C 159001 " Muscular dystrophy, limb-girdle, type 1B"

LMNB2 lamin B2 608709 " Lipodystrophy, partial, acquired"
LMX1B "LIM homeobox transcription factor 1, beta" 161200 Nail-patella syndrome
LPL lipoprotein lipase 144250 " Combined hyperlipidemia, familial"
LPL lipoprotein lipase 238600 Lipoprotein lipase deficiency
LRAT lecithin retinol acyltransferase (phosphatidylcholine--retinol O-acyltransferase) 613341 Leber congenital amaurosis 14
LRAT lecithin retinol acyltransferase (phosphatidylcholine--retinol O-acyltransferase) 613341 " Retinal dystrophy, early-onset severe"
LRAT lecithin retinol acyltransferase (phosphatidylcholine--retinol O-acyltransferase) 613341 " Retinitis pigmentosa, juvenile"
LRP2 low density lipoprotein receptor-related protein 2 222448 Donnai-Barrow syndrome
LRP5 low density lipoprotein receptor-related protein 5 601813 Exudative vitreoretinopathy 4
LRP5 low density lipoprotein receptor-related protein 5 144750 " Hyperostosis, endosteal"
LRP5 low density lipoprotein receptor-related protein 5 607634 " Osteopetrosis, AD type I"
LRP5 low density lipoprotein receptor-related protein 5 259770 Osteoporosis-pseudoglioma syndrome
LRP5 low density lipoprotein receptor-related protein 5 144750 Osteosclerosis
LRP5 low density lipoprotein receptor-related protein 5 607636 " van Buchem disease, type 2"
LTBP2 latent transforming growth factor beta binding protein 2 613086 " Glaucoma 3, primary congenital, D"
LTBP3 latent transforming growth factor beta binding protein 3 613097 " Tooth agenesis, selective, 6"
LYZ lysozyme 105200 " Amyloidosis, renal"
MAN2B1 "mannosidase, alpha, class 2B, member 1" 248500 " Mannosidosis, alpha-, types I and II"
MAP2K1 mitogen-activated protein kinase kinase 1 115150 Cardiofaciocutaneous syndrome
MAP2K2 mitogen-activated protein kinase kinase 2 115150 Cardiofaciocutaneous syndrome
MAPT microtubule-associated protein tau 600274 " Dementia, frontotemporal, with or without parkinsonism"
MAPT microtubule-associated protein tau 172700 Pick disease
MAPT microtubule-associated protein tau 260540 " Supranuclear palsy, progressive atypical"
MAPT microtubule-associated protein tau 601104 " Supranuclear palsy, progressive"
MBTPS2 "membrane-bound transcription factor peptidase, site 2" 308205 " Ichthyosis follicularis, atrichia, and photophobia syndrome"
MCOLN1 mucolipin 1 252650 Mucolipidosis IV
MEF2C myocyte enhancer factor 2C 613443 Chromosome 5q14.3 deletion syndrome
MEF2C myocyte enhancer factor 2C 613443 " Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations"
MERTK c-mer proto-oncogene tyrosine kinase 268000 Retinitis pigmentosa-38
MET met proto-oncogene (hepatocyte growth factor receptor) 114550 " Hepatocellular carcinoma, childhood type"
MET met proto-oncogene (hepatocyte growth factor receptor) 605074 " Renal cell carcinoma, papillary, familial and sporadic"
MFN2 mitofusin 2 609260 Charcot-Marie-Tooth disease type 2A2
MFN2 mitofusin 2 601152 Hereditary motor and sensory neuropathy VI
MFRP membrane frizzled-related protein 611040 " Microphthalmia, isolated 5"
MIP major intrinsic protein of lens fiber 604219 " Cataract, polymorphic and lamellar"
MITF microphthalmia-associated transcription factor 103500 Tietz albinism-deafness syndrome
MITF microphthalmia-associated transcription factor 193510 Waardenburg syndrome type IIA
MITF microphthalmia-associated transcription factor 103470 " Waardenburg syndrome/ocular albinism, digenic"
MKKS McKusick-Kaufman syndrome 209900 Bardet-Biedl syndrome 6
MKKS McKusick-Kaufman syndrome 236700 McKusick-Kaufman syndrome
MKS1 "Meckel syndrome, type 1" 209900 Bardet-Biedl syndrome 13
MKS1 "Meckel syndrome, type 1" 249000 " Meckel syndrome, type 1"
MLPH melanophilin 609227 Griscelli syndrome type 3
MMP2 "matrix metallopeptidase 2 (gelatinase A, 72kDa gelatinase, 72kDa type IV collagenase)" 259600 Torg-Winchester syndrome
MMP9 "matrix metallopeptidase 9 (gelatinase B, 92kDa gelatinase, 92kDa type IV collagenase)" 613073 Metaphyseal anadysplasia 2
MPO myeloperoxidase 254600 Myeloperoxidase deficiency
MSX2 msh homeobox 2 604757 " Craniosynostosis, type 2"
MSX2 msh homeobox 2 168500 Parietal foramina 1

MSX2 msh homeobox 2 168550 Parietal foramina with cleidocranial dysplasia
MTHFR methylenetetrahydrofolate reductase (NAD(P)H) 236250 Homocystinuria due to MTHFR deficiency
MYCN "v-myc myelocytomatosis viral related oncogene, neuroblastoma derived (avian)" 164280 Feingold syndrome
MYCN "v-myc myelocytomatosis viral related oncogene, neuroblastoma derived (avian)" 602585 Microcephaly and digital abnormalities with normal intelligence
MYD88 myeloid differentiation primary response gene (88) 612260 " Pyogenic bacterial infections, recurrent, due to MYD88 deficiency"
MYF6 myogenic factor 6 (herculin) 160150 " Myopathy, centronuclear"
MYH3 "myosin, heavy chain 3, skeletal muscle, embryonic" 193700 " Arthrogryposis, distal, type 2A"
MYH3 "myosin, heavy chain 3, skeletal muscle, embryonic" 601680 " Arthrogryposis, distal, type 2B"
MYO3A myosin IIIA 607101 " Deafness, autosomal recessive 30"
MYO5A "myosin VA (heavy chain 12, myoxin)" 214450 Griscelli syndrome type 1
MYO6 myosin VI 606346 " Deafness, autosomal dominant 22"
MYO6 myosin VI 607821 " Deafness, autosomal recessive 37"
MYO6 myosin VI 606346 " Deafness, sensorineural, with hypertrophic cardiomyopathy"
MYO7A myosin VIIA 601317 " Deafness, autosomal dominant 11, neurosensory"
MYO7A myosin VIIA 600060 " Deafness, autosomal recessive 2, neurosensory"
MYO7A myosin VIIA 276900 " Usher syndrome, type 1B"
MYOC "myocilin, trabecular meshwork inducible glucocorticoid response" 137750 " Glaucoma 1A, primary open angle, juvenile-onset"
NAGLU "N-acetylglucosaminidase, alpha" 252920 " Sanfilippo syndrome, type B"
NDP Norrie disease (pseudoglioma) 305390 " Exudative vitreoretinopathy, X-linked"
NDP Norrie disease (pseudoglioma) 310600 Norrie disease
NDRG1 N-myc downstream regulated 1 601455 Charcot-Marie-Tooth disease type 4D
NDUFS4 "NADH dehydrogenase (ubiquinone) Fe-S protein 4, 18kDa (NADH-coenzyme Q reductase)" 256000 Leigh syndrome
NDUFS4 "NADH dehydrogenase (ubiquinone) Fe-S protein 4, 18kDa (NADH-coenzyme Q reductase)" 252010 Mitochondrial complex I deficiency
NEB nebulin 256030 " Nemaline myopathy 2, autosomal recessive"
NEU1 sialidase 1 (lysosomal sialidase) 256550 " Sialidosis, type I"
NEU1 sialidase 1 (lysosomal sialidase) 256550 " Sialidosis, type II"
NEUROD1 neuronal differentiation 1 606394 Maturity-onset diabetes of the young 6
NF1 neurofibromin 1 607785 " Leukemia, juvenile myelomonocytic"
NF1 neurofibromin 1 162210 " Neurofibromatosis, familial spinal"
NF1 neurofibromin 1 162200 Neurofibromatosis type 1
NF1 neurofibromin 1 601321 Neurofibromatosis-Noonan syndrome
NF1 neurofibromin 1 193520 Watson syndrome
NF2 neurofibromin 2 (merlin) 607174 " Meningioma, NF2-related, somatic"
NF2 neurofibromin 2 (merlin) 101000 Neurofibromatosis type 2
NF2 neurofibromin 2 (merlin) 162091 Schwannomatosis
NHS Nance-Horan syndrome (congenital cataracts and dental anomalies) 302200 " Cataract, congenital, X-linked"
NHS Nance-Horan syndrome (congenital cataracts and dental anomalies) 302350 Nance-Horan syndrome
NIPBL Nipped-B homolog (Drosophila) 122470 Cornelia de Lange syndrome 1
NOD2 nucleotide-binding oligomerization domain containing 2 186580 Blau syndrome
NOD2 nucleotide-binding oligomerization domain containing 2 609464 " Sarcoidosis, early-onset"
NODAL nodal homolog (mouse) 270100 " Heterotaxy, visceral, 5"
NOG noggin 611377 Brachydactyly type B2
NOG noggin 184460 Stapes ankylosis with broad thumb and toes
NOG noggin 185800 " Symphalangism, proximal"
NOG noggin 186500 " Synostoses syndrome, multiple, 1"
NOG noggin 186570 Tarsal-carpal coalition syndrome
NOTCH1 notch 1 109730 Aortic valve disease

NOTCH2 notch 2 610205 Alagille syndrome 2
NPC1 "Niemann-Pick disease, type C1" 257220 " Niemann-Pick disease, type C1"
NPC1 "Niemann-Pick disease, type C1" 257220 Niemann-Pick disease type D
NPHP1 nephronophthisis 1 (juvenile) 609583 Joubert syndrome 4
NPHP1 nephronophthisis 1 (juvenile) 256100 " Nephronophthisis 1, juvenile"
NPHP1 nephronophthisis 1 (juvenile) 266900 Senior-Loken syndrome-1
NPHP4 nephronophthisis 4 606966 Nephronophthisis 4
NPHP4 nephronophthisis 4 606996 Senior-Loken syndrome 4
NPHS1 "nephrosis 1, congenital, Finnish type (nephrin)" 256300 " Nephrotic syndrome, type 1"
NR2E3 "nuclear receptor subfamily 2, group E, member 3" 268100 Enhanced S-cone syndrome
NR2E3 "nuclear receptor subfamily 2, group E, member 3" 611131 Retinitis pigmentosa-37
NR3C2 "nuclear receptor subfamily 3, group C, member 2" 605115 " Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy"
NR3C2 "nuclear receptor subfamily 3, group C, member 2" 177735 " Pseudohypoaldosteronism type I, autosomal dominant"
NRAS neuroblastoma RAS viral (v-ras) oncogene homolog 114500 Colorectal cancer
NRAS neuroblastoma RAS viral (v-ras) oncogene homolog 613224 Noonan syndrome 6
NRAS neuroblastoma RAS viral (v-ras) oncogene homolog 188470 " Thyroid carcinoma, follicular"
NSDHL NAD(P) dependent steroid dehydrogenase-like 308050 CHILD syndrome
NTF4 neurotrophin 4 613100 " Glaucoma 1, open angle, 1O"
NTRK1 "neurotrophic tyrosine kinase, receptor, type 1" 256800 " Insensitivity to pain, congenital, with anhidrosis"
NTRK1 "neurotrophic tyrosine kinase, receptor, type 1" 155240 " Medullary thyroid carcinoma, familial"
OAT ornithine aminotransferase 258870 Gyrate atrophy of choroid and retina with or without ornithinemia
OCA2 oculocutaneous albinism II 203200 " Albinism, brown oculocutaneous"
OCA2 oculocutaneous albinism II 203200 " Albinism, oculocutaneous, type II"
OPA1 optic atrophy 1 (autosomal dominant) 165500 Optic atrophy 1
OPA1 optic atrophy 1 (autosomal dominant) 125250 Optic atrophy and deafness
OPA3 "optic atrophy 3 (autosomal recessive, with chorea and spastic paraplegia)" 258501 3-Methylglutaconic aciduria type III
OPA3 "optic atrophy 3 (autosomal recessive, with chorea and spastic paraplegia)" 165300 Optic atrophy and cataract
OPN1MW "opsin 1 (cone pigments), medium-wave-sensitive" 303700 Blue cone monochromacy
OPN1MW "opsin 1 (cone pigments), medium-wave-sensitive" 303800 " Colorblindness, deutan"
OPN1SW "opsin 1 (cone pigments), short-wave-sensitive" 190900 " Colorblindness, tritan"
OPTN optineurin 613435 Amytrophic lateral sclerosis 12
OPTN optineurin 137760 " Glaucoma 1, open angle, E"
ORAI1 ORAI calcium release-activated calcium modulator 1 612782 Immune dysfunction with T-cell inactivation due to calcium entry defect 1
OSTM1 osteopetrosis associated transmembrane protein 1 259720 " Osteopetrosis, autosomal recessive 5"
OTC ornithine carbamoyltransferase 311250 Ornithine transcarbamylase deficiency
OTX2 orthodenticle homeobox 2 610125 " Microphthalmia, syndromic 5"
PAH phenylalanine hydroxylase 261600 Phenylketonuria
PANK2 pantothenate kinase 2 607236 HARP syndrome
PANK2 pantothenate kinase 2 234200 Neurodegeneration with brain iron accumulation 1
PAX2 paired box 2 120330 Optic nerve coloboma with renal disease
PAX3 paired box 3 122880 Craniofacial-deafness-hand syndrome
PAX3 paired box 3 268220 " Rhabdomyosarcoma 2, alveolar"
PAX3 paired box 3 193500 Waardenburg syndrome type 1
PAX3 paired box 3 148820 Waardenburg syndrome type 3
PAX4 paired box 4 612227 " Diabetes mellitus, ketosis-prone"
PAX4 paired box 4 125853 " Diabetes mellitus, type 2"
PAX4 paired box 4 612225 " Maturity-onset diabetes of the young, type IX"
PAX6 paired box 6 106210 Aniridia
PAX6 paired box 6 604219 Cataract with late-onset corneal dystrophy

PAX6 paired box 6 120430 Coloboma of optic nerve
PAX6 paired box 6 120200 " Coloboma, ocular"
PAX6 paired box 6 136520 Foveal hyperplasia
PAX6 paired box 6 206700 Gillespie syndrome
PAX6 paired box 6 148190 Keratitis
PAX6 paired box 6 120430 Morning glory disc anomaly
PAX6 paired box 6 165550 Optic nerve hypoplasia
PAX6 paired box 6 604229 Peters anomaly
PAX7 paired box 7 268220 " Rhabdomyosarcoma 2, alveolar"
PCDH15 protocadherin-related 15 609533 " Deafness, autosomal recessive 23"
PCDH15 protocadherin-related 15 601067 " Usher syndrome, type 1D/F digenic"
PCDH15 protocadherin-related 15 602083 " Usher syndrome, type 1F"
PDCD10 programmed cell death 10 603285 Cerebral cavernous malformations 3
PDE6B "phosphodiesterase 6B, cGMP-specific, rod, beta" 163500 " Night blindness, congenital stationary, autosomal dominant 2"
PDE6C "phosphodiesterase 6C, cGMP-specific, cone, alpha prime" 613093 Cone dystrophy 4
PDGFRA "platelet-derived growth factor receptor, alpha polypeptide" 606764 " Gastrointestinal stromal tumor, somatic"
PDGFRA "platelet-derived growth factor receptor, alpha polypeptide" 607685 " Hypereosinophilic syndrome, idiopathic, resistant to imatinib"
PDGFRB "platelet-derived growth factor receptor, beta polypeptide" 131440 Myeloproliferative disorder with eosinophilia
PEX7 peroxisomal biogenesis factor 7 266500 Refsum disease
PEX7 peroxisomal biogenesis factor 7 215100 Rhizomelic chondrodysplasia punctata type 1
PHOX2A paired-like homeobox 2a 602078 " Fibrosis of extraocular muscles, congenital, 2"
PIK3CA "phosphatidylinositol-4,5-bisphosphate 3-kinase, catalytic subunit alpha" 114480 " Breast cancer, somatic"
PIK3CA "phosphatidylinositol-4,5-bisphosphate 3-kinase, catalytic subunit alpha" 114500 " Colorectal cancer, somatic"
PIK3CA "phosphatidylinositol-4,5-bisphosphate 3-kinase, catalytic subunit alpha" 137215 " Gastric cancer, somatic"
PIK3CA "phosphatidylinositol-4,5-bisphosphate 3-kinase, catalytic subunit alpha" 114550 " Hepatocellular carcinoma, somatic"
PIK3CA "phosphatidylinositol-4,5-bisphosphate 3-kinase, catalytic subunit alpha" 182000 " Keratosis, seborrheic, somatic"
PIK3CA "phosphatidylinositol-4,5-bisphosphate 3-kinase, catalytic subunit alpha" 162900 " Nevus, epidermal"
PIK3CA "phosphatidylinositol-4,5-bisphosphate 3-kinase, catalytic subunit alpha" 211980 " Nonsmall cell lung cancer, somatic"
PIK3CA "phosphatidylinositol-4,5-bisphosphate 3-kinase, catalytic subunit alpha" 604370 " Ovarian cancer, somatic"
PITX2 paired-like homeodomain 2 180500 " Axenfeld-Rieger syndrome, type 1"
PITX2 paired-like homeodomain 2 137600 " Iridogoniodysgenesis, type 2"
PITX2 paired-like homeodomain 2 604229 Peters anomaly
PITX2 paired-like homeodomain 2 180550 Ring dermoid of cornea
PITX3 paired-like homeodomain 3 107250 Anterior segment mesenchymal dysgenesis
PITX3 paired-like homeodomain 3 610623 " Cataract, posterior polar, 4"
PITX3 paired-like homeodomain 3 610623 " Cataract, posterior polar, 4, syndromic"
PLA2G6 "phospholipase A2, group VI (cytosolic, calcium-independent)" 612953 " Dystonia-parkinsonism, adult-onset"
PLA2G6 "phospholipase A2, group VI (cytosolic, calcium-independent)" 256600 Infantile neuroaxonal dystrophy 1
PLA2G6 "phospholipase A2, group VI (cytosolic, calcium-independent)" 610217 Karak syndrome
PLA2G6 "phospholipase A2, group VI (cytosolic, calcium-independent)" 610217 Neurodegeneration with brain iron accumulation 2B
PLG plasminogen 217090 " Conjunctivitis, ligneous"
PLP1 proteolipid protein 1 312080 Pelizaeus-Merzbacher disease
PLP1 proteolipid protein 1 312920 Spastic paraplegia-2
PMM2 phosphomannomutase 2 212065 " Congenital disorder of glycosylation, type Ia"
POLG "polymerase (DNA directed), gamma" 203700 Alpers syndrome
POLG "polymerase (DNA directed), gamma" 603041 MNGIE without leukoencephalopathy
POLG "polymerase (DNA directed), gamma" 157640 " Progressive external ophthalmoplegia, autosomal dominant, with or without hypogonadism"
POLG "polymerase (DNA directed), gamma" 258450 " Progressive external ophthalmoplegia, autosomal recessive"

POLG "polymerase (DNA directed), gamma" 607459 " Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis"
POLG "polymerase (DNA directed), gamma" 607459 Spinocerebellar ataxia with epilepsy
POLH "polymerase (DNA directed), eta" 278750 " Xeroderma pigmentosum, variant type"
POR P450 (cytochrome) oxidoreductase 201750 " Adrenal hyperplasia, congenital, due to combined P450C17 and P450C21 deficiency"
POR P450 (cytochrome) oxidoreductase 201750 Antley-Bixler syndrome-like with disordered steroidogenesis
POR P450 (cytochrome) oxidoreductase 201750 " Disordered steroidogenesis, isolated"
POR P450 (cytochrome) oxidoreductase 201750 POR deficiency
POU4F3 POU class 4 homeobox 3 602459 " Deafness, autosomal dominant 15"
PPARG peroxisome proliferator-activated receptor gamma 609338 Carotid intimal medial thickness 1
PPARG peroxisome proliferator-activated receptor gamma 604367 " Insulin resistance, severe, digenic"
PPARG peroxisome proliferator-activated receptor gamma 604367 " Lipodystrophy, familial partial, type 3"
PPARG peroxisome proliferator-activated receptor gamma 601665 " Obesity, severe"
PPIB peptidylprolyl isomerase B (cyclophilin B) 259440 " Osteogenesis imperfecta, type IX"
PPP2R2B "protein phosphatase 2, regulatory subunit B, beta" 604326 Spinocerebellar ataxia 12
PPT1 palmitoyl-protein thioesterase 1 256730 " Ceroid lipofuscinosis, neuronal 1, infantile"
PRKAR1A "protein kinase, cAMP-dependent, regulatory, type I, alpha" 160980 " Carney complex, type 1"
PRKAR1A "protein kinase, cAMP-dependent, regulatory, type I, alpha" 255960 " Myxoma, intracardiac"
PRKAR1A "protein kinase, cAMP-dependent, regulatory, type I, alpha" 610489 " Pigmented adrenocortical disease, primary, 1"
PRKAR1A "protein kinase, cAMP-dependent, regulatory, type I, alpha" 188550 " Thyroid carcinoma, papillary"
PRKCG "protein kinase C, gamma" 605361 Spinocerebellar ataxia 14
PROM1 prominin 1 612657 Cone-rod dystrophy 12
PROM1 prominin 1 608051 " Macular dystrophy, retinal, 2"
PROM1 prominin 1 612095 Retinitis pigmentosa-41
PROM1 prominin 1 603786 Stargardt disease 4
PRPF31 PRP31 pre-mRNA processing factor 31 homolog (S. cerevisiae) 600138 Retinitis pigmentosa-11
PRPF8 PRP8 pre-mRNA processing factor 8 homolog (S. cerevisiae) 600059 Retinitis pigmentosa-13
PRPH2 "peripherin 2 (retinal degeneration, slow)" 613105 " Choriodal dystrophy, central areolar 2,"
PRPH2 "peripherin 2 (retinal degeneration, slow)" 608161 " Foveomacular dystrophy, adult-onset, with choroidal neovascularization"
PRPH2 "peripherin 2 (retinal degeneration, slow)" 169150 " Macular dystrophy, patterned"
PRPH2 "peripherin 2 (retinal degeneration, slow)" 608161 " Macular dystrophy, vitelliform"
PRPH2 "peripherin 2 (retinal degeneration, slow)" 608133 " Retinitis pigmentosa, digenic"
PRPH2 "peripherin 2 (retinal degeneration, slow)" 608133 Retinitis pigmentosa-7
PRPH2 "peripherin 2 (retinal degeneration, slow)" 136880 Retinitis punctata albescens
PSAP prosaposin 611721 Combined SAP deficiency
PSAP prosaposin 610539 " Gaucher disease, atypical"
PSAP prosaposin 611722 " Krabbe disease, atypical"
PSAP prosaposin 249900 Metachromatic leukodystrophy due to SAP-b deficiency
PSEN1 presenilin 1 607822 " Alzheimer disease, type 3"
PSEN1 presenilin 1 607822 " Alzheimer disease, type 3, with spastic paraparesis and apraxia"
PSEN1 presenilin 1 607822 " Alzheimer disease, type 3, with spastic paraparesis and unusual plaques"
PSEN1 presenilin 1 600274 " Dementia, frontotemporal"
PSEN1 presenilin 1 172700 Pick disease
PSEN2 presenilin 2 (Alzheimer disease 4) 606889 Alzheimer disease-4
PTCH1 patched 1 605462 " Basal cell carcinoma, somatic"
PTCH1 patched 1 109400 Basal cell nevus syndrome
PTCH1 patched 1 610828 Holoprosencephaly-7
PTCH2 patched 2 605462 " Basal cell carcinoma, somatic"
PTCH2 patched 2 155255 Medulloblastoma
PTEN phosphatase and tensin homolog 153480 Bannayan-Riley-Ruvalcaba syndrome