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

PTEN phosphatase and tensin homolog 158350 Cowden disease
PTEN phosphatase and tensin homolog 158350 Lhermitte-Duclos syndrome
PTEN phosphatase and tensin homolog 605309 Macrocephaly/autism syndrome
PTEN phosphatase and tensin homolog 276950 VATER association with macrocephaly and ventriculomegaly
PTF1A "pancreas specific transcription factor, 1a" 609069 " Diabetes mellitus, permanent neonatal, with cerebellar agenesis"
PTHLH parathyroid hormone-like hormone 613382 Brachydactyly type E2
PTPN11 "protein tyrosine phosphatase, non-receptor type 11" 151100 Leopard syndrome
PTPN11 "protein tyrosine phosphatase, non-receptor type 11" 607785 " Leukemia, juvenile myelomonocytic"
PTPN11 "protein tyrosine phosphatase, non-receptor type 11" 156250 Metachondromatosis
PTPN11 "protein tyrosine phosphatase, non-receptor type 11" 163950 Noonan syndrome 1
RAB23 "RAB23, member RAS oncogene family" 201000 Carpenter syndrome
RAB27A "RAB27A, member RAS oncogene family" 607624 Griscelli syndrome type 2
RAB3GAP1 RAB3 GTPase activating protein subunit 1 (catalytic) 600118 Warburg micro syndrome 1
RAB3GAP2 RAB3 GTPase activating protein subunit 2 (non-catalytic) 212720 Martsolf syndrome
RAF1 v-raf-1 murine leukemia viral oncogene homolog 1 611554 LEOPARD syndrome 2
RAF1 v-raf-1 murine leukemia viral oncogene homolog 1 611553 Noonan syndrome 5
RAG1 recombination activating gene 1 609889 " Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity"
RAG1 recombination activating gene 1 233650 Combined cellular and humoral immune defects with granulomas
RAG1 recombination activating gene 1 603554 Omenn syndrome
RAG1 recombination activating gene 1 601457 " Severe combined immunodeficiency, B cell-negative"
RAX retina and anterior neural fold homeobox 611038 " Microphthalmia, isolated 3"
RB1 retinoblastoma 1 109800 Bladder cancer
RB1 retinoblastoma 1 259500 Osteosarcoma
RD3 retinal degeneration 3 610612 Leber congenital amaurosis 12
RDH12 retinol dehydrogenase 12 (all-trans/9-cis/11-cis) 612712 Leber congenital amaurosis 13
RDH5 retinol dehydrogenase 5 (11-cis/9-cis) 136880 Fundus albipunctatus
RECQL4 RecQ protein-like 4 218600 Baller-Gerold syndrome
RECQL4 RecQ protein-like 4 266280 RAPADILINO syndrome
RECQL4 RecQ protein-like 4 268400 Rothmund-Thomson syndrome
RELN reelin 257320 " Lissencephaly syndrome, Norman-Roberts type"
REN renin 613092 " Hyperuricemic nephropathy, familial juvenile 2"
REN renin 267430 Renal tubular dysgenesis
RGS9 regulator of G-protein signaling 9 608415 Bradyopsia
RGS9BP regulator of G protein signaling 9 binding protein 608415 Bradyopsia
RHO rhodopsin 610445 " Night blindness, congenital stationery, autosomal dominant 1"
RLBP1 retinaldehyde binding protein 1 607475 Bothnia retinal dystrophy
RLBP1 retinaldehyde binding protein 1 136880 Fundus albipunctatus
RLBP1 retinaldehyde binding protein 1 607476 Newfoundland rod-cone dystrophy
RLBP1 retinaldehyde binding protein 1 136880 Retinitis punctata albescens
ROR2 receptor tyrosine kinase-like orphan receptor 2 113000 Brachydactyly type B1
ROR2 receptor tyrosine kinase-like orphan receptor 2 268310 " Robinow syndrome, autosomal recessive"
RP1 retinitis pigmentosa 1 (autosomal dominant) 180100 Retinitis pigmentosa-1
RP2 retinitis pigmentosa 2 (X-linked recessive) 312600 Retinitis pigmentosa-2
RPE65 retinal pigment epithelium-specific protein 65kDa 204100 Leber congenital amaurosis 2
RPGR retinitis pigmentosa GTPase regulator 304020 Cone-rod dystrophy-1
RPGR retinitis pigmentosa GTPase regulator 300455 " Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness"
RPGR retinitis pigmentosa GTPase regulator 300029 Retinitis pigmentosa-3
RPGRIP1 retinitis pigmentosa GTPase regulator interacting protein 1 608194 Cone-rod dystrophy 13
RPGRIP1L RPGRIP1-like 216360 COACH syndrome

RPGRIP1L RPGRIP1-like 611560 Joubert syndrome 7
RPGRIP1L RPGRIP1-like 611561 " Meckel syndrome, type 5"
RPS6KA3 "ribosomal protein S6 kinase, 90kDa, polypeptide 3" 303600 Coffin-Lowry syndrome
RUNX1 runt-related transcription factor 1 601626 " Leukemia, acute myeloid"
RUNX1 runt-related transcription factor 1 601399 " Platelet disorder, familial, with associated myeloid malignancy"
RUNX2 runt-related transcription factor 2 119600 Cleidocranial dysplasia
SAG S-antigen; retina and pineal gland (arrestin) 258100 Oguchi disease-1
SALL4 sal-like 4 (Drosophila) 607323 Duane-radial ray syndrome
SALL4 sal-like 4 (Drosophila) 147750 IVIC syndrome
SAT1 spermidine/spermine N1-acetyltransferase 1 308800 Keratosis follicularis spinulosa decalvans
SCN1A "sodium channel, voltage-gated, type I, alpha subunit" 607208 Dravet syndrome
SCN1A "sodium channel, voltage-gated, type I, alpha subunit" 604233 " Epilepsy, generalized, with febrile seizures plus, type 2"
SCN1A "sodium channel, voltage-gated, type I, alpha subunit" 607208 " Epilepsy, severe myoclonic, of infancy"
SCN1A "sodium channel, voltage-gated, type I, alpha subunit" 604403 " Febrile convulsions, familial, 3A"
SCN1A "sodium channel, voltage-gated, type I, alpha subunit" 609634 " Migraine, familial hemiplegic, 3"
SCN1B "sodium channel, voltage-gated, type I, beta subunit" 612838 Brugada syndrome 5
SCN1B "sodium channel, voltage-gated, type I, beta subunit" 612838 " Cardiac conduction defect, nonspecific"
SCN1B "sodium channel, voltage-gated, type I, beta subunit" 604233 Generalized epilepsy with febrile seizures plus
SDHA "succinate dehydrogenase complex, subunit A, flavoprotein (Fp)" 613642 " Cardiomyopathy, dilated, 1GG"
SDHA "succinate dehydrogenase complex, subunit A, flavoprotein (Fp)" 256000 Leigh syndrome
SDHA "succinate dehydrogenase complex, subunit A, flavoprotein (Fp)" 252011 Mitochondrial respiratory chain complex II deficiency
SDHB "succinate dehydrogenase complex, subunit B, iron sulfur (Ip)" 612359 Cowden-like syndrome
SDHB "succinate dehydrogenase complex, subunit B, iron sulfur (Ip)" 606864 Paraganglioma and gastric stromal sarcoma
SDHB "succinate dehydrogenase complex, subunit B, iron sulfur (Ip)" 115310 " Paraganglioma, familial chromaffin, 4"
SDHB "succinate dehydrogenase complex, subunit B, iron sulfur (Ip)" 171300 Pheochromocytoma
SEMA4A "sema domain, immunoglobulin domain (Ig), transmembrane domain (TM) and short cytoplasmic domain, (semaphorin) 4A" 610283 Cone-rod dystrophy 10
SEMA4A "sema domain, immunoglobulin domain (Ig), transmembrane domain (TM) and short cytoplasmic domain, (semaphorin) 4A" 610282 Retinitis pigmentosa-35
SERPINA1 "serpin peptidase inhibitor, clade A (alpha-1 antiproteinase, antitrypsin), member 1" 613490 Emphysema due to AAT deficiency
SERPINA1 "serpin peptidase inhibitor, clade A (alpha-1 antiproteinase, antitrypsin), member 1" 613490 " Emphysema-cirrhosis, due to AAT deficiency"
SERPINA1 "serpin peptidase inhibitor, clade A (alpha-1 antiproteinase, antitrypsin), member 1" 613490 Hemorrhagic diathesis due to ¥'antithrombin¥' Pittsburgh
SETX senataxin 602433 " Amyotrophic lateral sclerosis 4, juvenile"
SETX senataxin 606002 Ataxia-ocular apraxia-2
SFTPC surfactant protein C 610913 " Surfactant metabolism dysfunction, pulmonary, 2"
SGCA "sarcoglycan, alpha (50kDa dystrophin-associated glycoprotein)" 608099 " Muscular dystrophy, limb-girdle, type 2D"
SGCB "sarcoglycan, beta (43kDa dystrophin-associated glycoprotein)" 604286 " Muscular dystrophy, limb-girdle, type 2E"
SGSH N-sulfoglucosamine sulfohydrolase 252900 " Sanfilippo syndrome, type A"
SH3BP2 SH3-domain binding protein 2 118400 Cherubism
SH3PXD2B SH3 and PX domains 2B 249420 Frank-ter Haar syndrome
SHH sonic hedgehog 120200 " Coloboma, ocular"
SHH sonic hedgehog 142945 Holoprosencephaly-3
SHH sonic hedgehog 611638 " Microphthalmia, isolated, with coloboma 5"
SHH sonic hedgehog 147250 Solitary median maxillary central incisor
SHOX short stature homeobox 249700 Langer mesomelic dysplasia
SHOX short stature homeobox 127300 Leri-Weill dyschondrosteosis
SHOX short stature homeobox 300582 " Short stature, idiopathic familial"

SIX1 SIX homeobox 1 608389 Brachiootic syndrome 3
SIX1 SIX homeobox 1 605192 " Deafness, autosomal dominant 23"
SIX3 SIX homeobox 3 157170 Holoprosencephaly-2
SIX5 SIX homeobox 5 610896 Branchiootorenal syndrome 2
SIX6 SIX homeobox 6 212550 " Microphthalmia, isolated, with cataract 2"
SLC12A6 "solute carrier family 12 (potassium/chloride transporters), member 6" 218000 Agenesis of the corpus callosum with peripheral neuropathy
SLC17A5 "solute carrier family 17 (anion/sugar transporter), member 5" 604369 Salla disease
SLC17A5 "solute carrier family 17 (anion/sugar transporter), member 5" 269920 " Sialic acid storage disorder, infantile"
SLC1A1 "solute carrier family 1 (neuronal/epithelial high affinity glutamate transporter, system Xag), member 1" 222730 Dicarboxylicaminoaciduria
SLC1A3 "solute carrier family 1 (glial high affinity glutamate transporter), member 3" 612656 " Episodic ataxia, type 6"
SLC25A4 "solute carrier family 25 (mitochondrial carrier; adenine nucleotide translocator), member 4" 192600 " Cardiomyopathy, familial hypertrophic"
SLC25A4 "solute carrier family 25 (mitochondrial carrier; adenine nucleotide translocator), member 4" 609283 Progressive external ophthalmoplegia with mitochondrial DNA deletions 3
SLC2A1 "solute carrier family 2 (facilitated glucose transporter), member 1" 606777 GLUT1 deficiency syndrome 1
SLC2A1 "solute carrier family 2 (facilitated glucose transporter), member 1" 612126 GLUT1 deficiency syndrome 2
SLC34A2 "solute carrier family 34 (sodium phosphate), member 2" 265100 Pulmonary alveolar microlithiasis
SLC34A2 "solute carrier family 34 (sodium phosphate), member 2" 610441 Testicular microlithiasis
SLC39A13 "solute carrier family 39 (zinc transporter), member 13" 612350 " Spondylocheirodysplasia, Ehlers-Danlos syndrome-like"
SLC39A4 "solute carrier family 39 (zinc transporter), member 4" 201100 Acrodermatitis enteropathica
SLC40A1 "solute carrier family 40 (iron-regulated transporter), member 1" 606069 " Hemochromatosis, type 4"
SLC45A2 "solute carrier family 45, member 2" 606574 " Oculocutaneous albinism, type IV"
SLC4A11 "solute carrier family 4, sodium borate transporter, member 11" 613268 " Corneal dystrophy, Fuchs endothelial, 4"
SLC4A11 "solute carrier family 4, sodium borate transporter, member 11" 217700 Corneal endothelial dystrophy 2
SLC4A11 "solute carrier family 4, sodium borate transporter, member 11" 217400 Corneal endothelial dystrophy and perceptive deafness
SLC4A4 "solute carrier family 4, sodium bicarbonate cotransporter, member 4" 604278 " Renal tubular acidosis, proximal, with ocular abnormalities"
SLC6A3 "solute carrier family 6 (neurotransmitter transporter, dopamine), member 3" 613135 " Parkinsonism-dystonia, infantile"
SMARCA4 "SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a, member 4" 613325 Rhabdoid tumor predisposition syndrome 2
SMARCB1 "SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily b, member 1" 609322 Rhabdoid predisposition syndrome 1
SNAI2 snail homolog 2 (Drosophila) 172800 Piebaldism
SNAI2 snail homolog 2 (Drosophila) 608890 Waardenburg syndrome type 2D
SNCA "synuclein, alpha (non A4 component of amyloid precursor)" 127750 " Dementia, Lewy body"
SNCA "synuclein, alpha (non A4 component of amyloid precursor)" 605543 Parkinson disease 4
SOD1 "superoxide dismutase 1, soluble" 105400 " Amyotrophic lateral sclerosis, due to SOD1 deficiency"
SOS1 son of sevenless homolog 1 (Drosophila) 135300 " Fibromatosis, gingival"
SOS1 son of sevenless homolog 1 (Drosophila) 610733 Noonan syndrome 4
SOX2 SRY (sex determining region Y)-box 2 206900 " Microphthalmia, syndromic 3"
SOX2 SRY (sex determining region Y)-box 2 206900 Optic nerve hypoplasia and abnormalities of the central nervous system
SOX3 SRY (sex determining region Y)-box 3 300123 " Mental retardation, X-linked, with isolated growth hormone deficiency"
SOX3 SRY (sex determining region Y)-box 3 312000 " Panhypopituitarism, X-linked"
SOX9 SRY (sex determining region Y)-box 9 114290 Acampomelic campomelic dysplasia
SOX9 SRY (sex determining region Y)-box 9 114290 Campomelic dysplasia with autosomal sex reversal
SOX9 SRY (sex determining region Y)-box 9 114290 Campomelic dysplasia
SPINT2 "serine peptidase inhibitor, Kunitz type, 2" 270420 " Diarrhea 3, secretory sodium, congenital, syndromic"

SPTAN1 "spectrin, alpha, non-erythrocytic 1" 613477 " Epileptic encephalopathy, early infantile, 5"
SPTBN2 "spectrin, beta, non-erythrocytic 2" 600224 Spinocerebellar ataxia-5
STAT1 "signal transducer and activator of transcription 1, 91kDa" 209950 " Mycobacterial infection, atypical, familial disseminated"
STAT3 signal transducer and activator of transcription 3 (acute-phase response factor) 147060 Hyper-IgE recurrent infection syndrome
STIM1 stromal interaction molecule 1 612783 " Immune dysfunction, with T-cell inactivation due to calcium entry defect 2"
STK11 serine/threonine kinase 11 175200 Peutz-Jeghers syndrome
STK11 serine/threonine kinase 11 273300 " Testicular tumor, sporadic"
STRA6 stimulated by retinoic acid gene 6 homolog (mouse) 601186 " Microphthalmia, syndromic 9"
SYNE1 "spectrin repeat containing, nuclear envelope 1" 612998 Emery-Dreifuss muscular dystrophy 4
SYNE1 "spectrin repeat containing, nuclear envelope 1" 610743 " Spinocerebellar ataxia, autosomal recessive 8"
SYP synaptophysin 300802 " Mental retardation, X-linked, with or without epilepsy"
TBP TATA box binding protein 607136 Spinocerebellar ataxia 17
TBX15 T-box 15 260660 Cousin syndrome
TBX3 T-box 3 181450 Ulnar-mammary syndrome
TBX5 T-box 5 142900 Holt-Oram syndrome
TCOF1 Treacher Collins-Franceschetti syndrome 1 154500 Treacher Collins mandibulofacial dysostosis
TF transferrin 209300 Atransferrinemia
TFAP2A transcription factor AP-2 alpha (activating enhancer binding protein 2 alpha) 113620 Branchiooculofacial syndrome
TFAP2B transcription factor AP-2 beta (activating enhancer binding protein 2 beta) 169100 Char syndrome
TG thyroglobulin 274700 Thyroid dyshormonogenesis 3
TGFB1 "transforming growth factor, beta 1" 131300 Camurati-Engelmann disease
TGFB3 "transforming growth factor, beta 3" 107970 Arrhythmogenic right ventricular dysplasia 1
TGFBI "transforming growth factor, beta-induced, 68kDa" 607541 " Corneal dystrophy, Avellino type"
TGFBI "transforming growth factor, beta-induced, 68kDa" 121820 " Corneal dystrophy, epithelial basement membrane"
TGFBI "transforming growth factor, beta-induced, 68kDa" 121900 " Corneal dystrophy, Groenouw type I"
TGFBI "transforming growth factor, beta-induced, 68kDa" 122200 " Corneal dystrophy, lattice type I"
TGFBI "transforming growth factor, beta-induced, 68kDa" 608471 " Corneal dystrophy, lattice type IIIA"
TGFBI "transforming growth factor, beta-induced, 68kDa" 608470 " Corneal dystrophy, Reis-Bucklers type"
TGFBI "transforming growth factor, beta-induced, 68kDa" 602082 " Corneal dystrophy, Thiel-Behnke type"
TGFBR2 "transforming growth factor, beta receptor II (70/80kDa)" 133239 " Esophageal cancer, somatic"
TGFBR2 "transforming growth factor, beta receptor II (70/80kDa)" 610168 " Loeys-Dietz syndrome, type 1B"
TGFBR2 "transforming growth factor, beta receptor II (70/80kDa)" 610380 " Loeys-Dietz syndrome, type 2B"
TGM6 transglutaminase 6 609796 " Peeling skin syndrome, acral type"
TH tyrosine hydroxylase 605407 " Segawa syndrome, recessive"
THRB "thyroid hormone receptor, beta" 188570 Thyroid hormone resistance
THRB "thyroid hormone receptor, beta" 274300 " Thyroid hormone resistance, autosomal recessive"
THRB "thyroid hormone receptor, beta" 145650 " Thyroid hormone resistance, selective pituitary"
TIMP3 TIMP metallopeptidase inhibitor 3 136900 Sorsby fundus dystrophy
TNFRSF1A "tumor necrosis factor receptor superfamily, member 1A" 142680 " Periodic fever, familial"
TNXB tenascin XB 606408 Ehlers-Danlos due to tenascin X deficiency
TNXB tenascin XB 130020 " Ehlers-Danlos syndrome, hypermobility type"
TP53 tumor protein p53 202300 Adrenal cortical carcinoma
TP53 tumor protein p53 114480 Breast cancer
TP53 tumor protein p53 260500 Choroid plexus papilloma
TP53 tumor protein p53 114500 Colorectal cancer
TP53 tumor protein p53 114550 Hepatocellular carcinoma
TP53 tumor protein p53 151623 Li-Fraumeni syndrome
TP53 tumor protein p53 151623 Li-Fraumeni-like syndrome
TP53 tumor protein p53 607107 Nasopharyngeal carcinoma
TP53 tumor protein p53 259500 Osteosarcoma

TP53 tumor protein p53 260350 Pancreatic cancer
TP63 tumor protein p63 103285 ADULT syndrome
TP63 tumor protein p63 604292 " Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3"
TP63 tumor protein p63 106260 Hay-Wells syndrome
TP63 tumor protein p63 603543 Limb-mammary syndrome
TP63 tumor protein p63 129400 Orofacial cleft 8
TP63 tumor protein p63 129400 Rapp-Hodgkin syndrome
TP63 tumor protein p63 605289 " Split-hand/foot malformation, type 4"
TPO thyroid peroxidase 274500 Thyroid dyshormonogenesis 2A
TRIM37 tripartite motif containing 37 253250 Mulibrey nanism
TRPC6 "transient receptor potential cation channel, subfamily C, member 6" 603965 " Glomerulosclerosis, focal segmental, 2"
TRPM1 "transient receptor potential cation channel, subfamily M, member 1" 613216 " Night blindness, congenital stationary, type IC"
TSC2 tuberous sclerosis 2 606690 " Lymphangioleiomyomatosis, somatic"
TSC2 tuberous sclerosis 2 191100 Tuberous sclerosis-2
TSHR thyroid stimulating hormone receptor 603373 " Hyperthyroidism, familial gestational"
TSHR thyroid stimulating hormone receptor 609152 " Hyperthyroidism, nonautoimmune"
TSHR thyroid stimulating hormone receptor 275200 " Hypothyroidism, congenital, nongoitrous"
TSPAN12 tetraspanin 12 613310 Exudative vitreoretinopathy 5
TTBK2 tau tubulin kinase 2 604432 Spinocerebellar ataxia-11
TTC8 tetratricopeptide repeat domain 8 209900 Bardet-Biedl syndrome 8
TTC8 tetratricopeptide repeat domain 8 613464 Retinitis pigmentosa 51
TTN titin 604145 " Cardiomyopathy, dilated, 1G"
TTN titin 608807 " Muscular dystrophy, limb-girdle, type 2J"
TTN titin 611705 " Myopathy, early-onset, with fatal cardiomyopathy"
TTN titin 603689 " Myopathy, proximal, with early respiratory muscle involvement"
TTN titin 600334 " Tibial muscular dystrophy, tardive"
TTPA tocopherol (alpha) transfer protein 277460 Ataxia with isolated vitamin E deficiency
TTR transthyretin 105210 " Amyloidosis, hereditary, transthyretin-related"
TTR transthyretin 115430 " Carpal tunnel syndrome, familial"
TUBB3 "tubulin, beta 3 class III" 600638 " Fibrosis of extraocular muscles, congenital, 3A"
TULP1 tubby like protein 1 600132 Retinitis pigmentosa-14
TWIST1 twist homolog 1 (Drosophila) 123100 " Craniosynostosis, type 1"
TWIST1 twist homolog 1 (Drosophila) 101400 Saethre-Chotzen syndrome with eyelid anomalies
TWIST1 twist homolog 1 (Drosophila) 101400 Saethre-Chotzen syndrome
TYR tyrosinase (oculocutaneous albinism IA) 203100 " Albinism, oculocutaneous, type IA"
TYR tyrosinase (oculocutaneous albinism IA) 606952 " Albinism, oculocutaneous, type IB"
TYR tyrosinase (oculocutaneous albinism IA) 103470 " Waardenburg syndrome/albinism, digenic"
TYRP1 tyrosinase-related protein 1 203290 " Albinism, brown"
TYRP1 tyrosinase-related protein 1 278400 " Albinism, rufous"
USH1C "Usher syndrome 1C (autosomal recessive, severe)" 602092 " Deafness, autosomal recessive 18"
USH1C "Usher syndrome 1C (autosomal recessive, severe)" 276904 " Usher syndrome, type 1C"
USH2A "Usher syndrome 2A (autosomal recessive, mild)" 268000 Retinitis pigmentosa-39
USH2A "Usher syndrome 2A (autosomal recessive, mild)" 276901 " Usher syndrome, type 2A"
VCAN versican 143200 Wagner syndrome 1
VDR "vitamin D (1,25- dihydroxyvitamin D3) receptor" 166710 " Osteoporosis, involutional"
VDR "vitamin D (1,25- dihydroxyvitamin D3) receptor" 277440 " Rickets, vitamin D-resistant, type IIA"
VLDLR very low density lipoprotein receptor 224050 Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1
VPS13A vacuolar protein sorting 13 homolog A (S. cerevisiae) 200150 Choreoacanthocytosis
VSX1 visual system homeobox 1 122000 " Corneal dystrophy, hereditary polymorphous posterior"

VSX1 visual system homeobox 1 148300 Keratoconus
WDR36 WD repeat domain 36 609887 " Glaucoma 1, open angle, G"
WNT4 "wingless-type MMTV integration site family, member 4" 158330 Mullerian aplasia and hyperandrogenism
WNT4 "wingless-type MMTV integration site family, member 4" 611812 SERKAL syndrome
WNT7A "wingless-type MMTV integration site family, member 7A" 228930 Fuhrmann syndrome
WNT7A "wingless-type MMTV integration site family, member 7A" 276820 " Ulna and fibula, absence of, with sever limb deficiency"
WT1 Wilms tumor 1 194080 Denys-Drash syndrome
WT1 Wilms tumor 1 136680 Frasier syndrome
WT1 Wilms tumor 1 608978 Meacham syndrome
WT1 Wilms tumor 1 256370 " Nephrotic syndrome, type 4"
WT1 Wilms tumor 1 194070 " Wilms tumor, type 1"
XDH xanthine dehydrogenase 278300 " Xanthinuria, type I"
XPA "xeroderma pigmentosum, complementation group A" 278700 Xeroderma pigmentosum group A
XPC "xeroderma pigmentosum, complementation group C" 278720 Xeroderma pigmentosum group C
ZEB1 zinc finger E-box binding homeobox 1 613270 " Corneal dystrophy, Fuchs endothelial, 6"
ZEB1 zinc finger E-box binding homeobox 1 609141 " Corneal dystrophy, posterior polymorphous, 3"
ZEB2 zinc finger E-box binding homeobox 2 235730 Mowat-Wilson syndrome
ZIC2 Zic family member 2 609637 Holoprosencephaly-5
ZIC3 Zic family member 3 306955 " Congenital heart defects, nonsyndromic, 1, X-linked"
ZIC3 Zic family member 3 306955 " Heterotaxy, visceral, 1, S-linke"
ZNF513 zinc finger protein 513 613617 Retinitis pigmentosa 58
ZNF592 zinc finger protein 592 606937 " Spinocerebellar ataxia, autosomal recessive 5"
ZNF81 zinc finger protein 81 300498 " Mental retardation, X-linked 45"

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難病・がん等の疾患分野の医療の実用化研究事業（難病関連分野）
総合研究報告書

iPS細胞を用いた先天異常症の遺伝要因の解明のためのシステム構築

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研究要旨

臨床データ・検体・次世代シーケンサー技術という研究リソースを全国規模で共有して遺伝要因の解明を目指す。研究チーム内での分担研究者の役割は疾患特異的iPS細胞の樹立と蓄積である。従来の方法で疾患iPS細胞の解析を行いながら末梢血だけでなく不死化リンパ芽球から誘導したiPS細胞が神経疾患研究に使用できることをさらに明確に示し、当研究班内ですでに蓄積されている不死化リンパ芽球ストックがiPS細胞研究リソースとして有用であることを示した。

A．研究目的

本計画では中枢神経系・感覚器が発生学的に頭部外胚葉という共通の由来を持ち、先天異常の発症に共通の遺伝子経路が関与する可能性が高いことに着目して、既存3コンソーシアムの複合体を新たに編成の上、臨床データ・検体・次世代シーケンサー技術という研究リソースを全国規模で共有して遺伝要因の解明を目指す。研究チーム内での分担研究者の役割は疾患特異的iPS細胞の樹立と蓄積である。

B．研究方法

1．ヒトiPS細胞の樹立システムの開発

H23年度は患者に皮膚生検を行い樹立した線維芽細胞から疾患解析に用いるiPS細胞クローンの選定方法に関する検討を行った。レトロウィルス感染6日後に導入遺伝子が高発現している線維芽細胞と樹立したiPS細胞で導入遺伝子の発現を定量PCRにより比較し、導入遺伝子の発現がサイレンシングにより十分抑えられている line を選定し、神経系細胞への分化効率を検討した。

2．H24年度、H25年度は協力患者の拡大を図るために、侵襲の少ない末梢血を用いた方法の最適化を検討した。

①末梢血からのiPS細胞の樹立と神経分化

健康成人から末梢血を採取し、CD3陽性のT細胞を純化し、センダイウイルスを用いて遺伝子導入を行いiPS細胞を樹立した。樹立したT細胞由来のiPS細胞を神経分化誘導を行い、疾患解析に用いることが可能かを検討した。

②不死化リンパ芽球株からの iPS 細胞の樹立と神経分化

健康成人からEBVを用いて作成した**不死化リンパ芽球株**に遺伝子導入を行いiPS細胞を樹立した。樹立したT細胞由来のiPS細胞を神経分化誘導を行い、疾患解析に用いることが可能かを検討した。

（倫理面への配慮）

動物の飼育・管理は慶應義塾大学医学部動物実験ガイドラインを遵守して行われている。また、当研究室におけるヒトES細胞の使用については、文部科学省の「ヒトES細胞の樹立及び使用に関する指針」に基づき、平成19年10月31日に「ヒト胚性幹細胞を用いた中枢神経系の再生医学の基礎的研究」として承認され、研究計画はそれに準拠したものとなっている。患者からのiPS細胞の樹立は「神経疾患患者からのiPS細胞の樹立とそれを用いた疾患解析に関する研究」として慶應義塾大学医学部倫理委員会の承認を受けており（2008年6月）、十分な説明の上で患者の同意の下で行われる。他施設との共同研究においては当該施設においても倫理委員会の承認を受けている。

C．研究結果

1．H23年度はALS（FUS変異）、若年発症型パーキンソン病（PARK2）の解析を行い、iPS細胞の樹立と解析システムを確立した。H24年度以降は先天異常を示す疾患iPS細胞（CHARGE症候群・先天性ミエリン形成不全・シェーグレンラルソン症候群）の解析を行い、iPS細胞から誘導した細胞が患者と同じ表現型を持つことを示した。

2.末梢血由来iPS細胞の神経分化

T細胞から誘導したiPS細胞は良好に神経分化誘導が可能であり、従来の線維芽細胞由来の細胞と同様に神経疾患の病態解析に使用可能と考えられる。本年度は誘導した神経細胞における電気生理学的活動が存在することを確認した。

3.不死化リンパ芽球由来iPS細胞の神経分化
不死化リンパ芽球から誘導した全てのiPS細胞が良好に神経分化誘導が可能であり、これまでの解析ではEBVによる不死化の影響は分化細胞では従来の（不死化していない）血液細胞由来のiPS細胞と比較して有意な差を認めていない。本年度はさらに神経変性疾患において従来の線維芽細胞・血液細胞由来の細胞と同様に病態を再現することに成功した。現在当研究班内でストックされている不死化リンパ芽球ストックは数多く保有されており、それら全てがiPS細胞研究リソースとして用いることができるという重要な結果である。

D．考察

先天異常を示す疾患iPS細胞を患者線維芽細胞作成し、関連する表現型を生体外で再現することに成功した。

末梢血から作製した iPS 細胞は、T 細胞由来だけでなく、不死化リンパ芽球由来線維芽細胞由来の iPS 細胞も、従来の線維芽細胞由来 iPS 細胞とほぼ同様の分化誘導能力を示し、神経変性疾患の病態を再現できることを確認した。従ってこれらの細胞は十分に疾患解析に用いることが出来るのではないかと考えられる。

今後は、血液細胞からのiPS細胞を標準として患者協力を募っていく。受診のタイミングが合わない場合、樹立施設との連携が困難な受診施設では、不死化リンパ芽球化(SRLに依頼可能)を行い。ストックしておくことべきだと考えられる。

E．結論

先天異常を示す疾患iPS細胞を作成し、いくつかの疾患ではiPS細胞から誘導した細胞が患者と同じ表現型を持つことを示した。また、末梢血由来の細胞からのiPS細胞は線維芽細胞と似た性質を持つことが確認され、検体採取に末梢血を用いることにより、研究協力を得やすいのではないかと思われた。また、患者血液の不死化リンパ芽球化を予め行っておくことにより、樹立施設へ即時検体が運搬することが難しい施設でも研究参加が可能であると考えられた。さらに、当研究班内ですでに蓄積されている不死化リンパ芽球ストックがiPS細胞研究リソースとして用いることができると考えられる。

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5) **赤松　和土**：シンポジウム　ALSに対する再生医療の開発：幹細胞生物学を応用した神経疾患病態研究：第53回日本神経学会学術大会　2012年5月23日　東京
6) **赤松　和土**：招待講演：多能性幹細胞・神経幹細胞を用いた再生医療と病態解析：愛知医科大学　細胞治療研究会　2013年3月5日　愛知
7) **赤松　和土**：シンポジウム　疾患特異的iPS細胞を用いた病態解明および治療法確立研究：神経疾患患者由来細胞からの神経系細胞の誘導と病態解析：日本再生医療学会総会　2013年3月22日　横浜
8) **赤松　和土,** 岡野　栄之：シンポジウム45　iPS細胞技術を用いた中枢神経系の再生と創薬研究：iPS technology-based cell therapy for damaged CNS and investigation of neural disorders.：第90回日本生理学会大会2013年3月29日東京
9) **赤松　和土**：　特別講演：iPS細胞技術の神経疾患研究・治療への応用　第116回日本小児科学会学術集会2013年4月19日　広島
10) **赤松　和土：招待講演：題名未定　第11回**横浜小児先端医療セミナー　2013年5月24日(予定)　横浜
11) **赤松　和土：招待講演：**iPS細胞技術を用いた神経系の再生医療と疾患解析　第2回小児泌尿器科研究会　2013年6月1日　東京
12) **赤松　和土：招待講演：**iPS細胞を使った神経疾患の治療法の開発：第20回東京血管疾患研究所セミナー　2013年6月8日(予定)　東京
13) **赤松　和土**：シンポジウム　疾患iPS細胞と創薬：神経疾患患者由来細胞からの神経系細胞の誘導と病態解析：**炎症再生学会**　2013年7月2日(予定)　京都
14) **赤松　和土**：シンポジウム：「iPS technology-based regenerative medicine for damaged central nervous system」第11 回遺伝子治療学会シンポジウム　**遺伝子治療学会** 2013 年7 月6 日 岡山
15) **赤松　和土**：「iPS 細胞技術を用いた神経疾患研究と治療」第37 回日本血液事業学会総会シンポジウム 2013 年10 月21 日札幌

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