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NEWBORN GENOMIC SEQUENCING
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Gene List

Disorder	Gene
GENERALIZED ARTERIAL CALCIFICATION OF INFANCY 2	*ABCC6**
PERMANENT NEONATAL DIABETES MELLITUS 3; FAMILIAL HYPERINSULINEMIC HYPOGLYCEMIA 1	*ABCC8*
METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblJ TYPE	*ABCD4*
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 20	*ACAD9*
DEFICIENCY OF MEDIUM-CHAIN ACYL-CoA DEHYDROGENASE	*ACADM*
DEFICIENCY OF VERY LONG-CHAIN ACYL-CoA DEHYDROGENASE	*ACADVL*
α-METHYLACETOACETIC ACIDURIA	*ACAT1*
HEREDITARY HEMORRHAGIC TELANGIECTASIA TYPE 2	*ACVRL1*
T CELL-NEG., B CELL-NEG., NK CELL-NEG. AR SCID DUE TO ADA DEFICIENCY	*ADA*
VASCULITIS, AUTOINFLAMMATION, IMMUNODEFICIENCY & HEMATOLOGIC DEFECTS SYNDROME	*ADA2*
HEREDITARY THROMBOTIC THROMBOCYTOPENIC PURPURA	*ADAMTS13*
GLYCOGEN STORAGE DISEASE III	*AGL*
CONGENITAL MYASTHENIC SYNDROME 8	*AGRN*
HYPERMETHIONINEMIA WITH S-ADENOSYLHOMOCYSTEINE HYDROLASE DEFICIENCY	*AHCY*
RETICULAR DYSGENESIS	*AK2*
PYRIDOXINE-DEPENDENT EPILEPSY	*ALDH7A1*
HEREDITARY FRUCTOSE INTOLERANCE	*ALDOB*
CONGENITAL MYASTHENIC SYNDROME 15	*ALG14*
HYPOPHOSPHATASIA, CHILDHOOD; HYPOPHOSPHATASIA, INFANTILE	*ALPL*
IMERSLUND-GRASBECK SYNDROME 2	*AMN*
AUTOSOMAL NEPHROGENIC DIABETES INSIPIDUS	*AQP2*
ARGININEMIA	*ARG1*
IMMUNODEFICIENCY 71 WITH INFLAMMATORY DISEASE & CONGENITAL THROMBOCYTOPENIA	*ARPC1B*
METACHROMATIC LEUKODYSTROPHY	*ARSA*
MUCOPOLYSACCHARIDOSIS TYPE VI	*ARSB*
ARGININOSUCCINIC ACIDURIA	*ASL*
CLASSIC CITRULLINEMIA	*ASS1*
DISTAL RENAL TUBULAR ACIDOSIS 3 W./WITHOUT SENSORINEURAL HEARING LOSS	*ATP6V0A4*
DISTAL RENAL TUBULAR ACIDOSIS 2 WITH SENSORINEURAL HEARING LOSS	*ATP6V1B1*
MENKES DISEASE; OCCIPTAL HORN SYNDROME	*ATP7A*
X-LINKED NEPHROGENIC DIABETES INSIPIDUS; NEPHROGENIC SYNDROME OF INAPPROPRIATE ANTIDIURESIS	*AVPR2*
BRANCHED-CHAIN KETO ACID DEHYDROGENASE KINASE DEFICIENCY	*BCKDK*
AR AGAMMAGLOBULINEMIA 4	*BLNK*
FANCONI ANEMIA, COMPLEMENTATION GROUP D1	*BRCA2*
FANCONI ANEMIA, COMPLEMENTATION GROUP J	*BRIP1*
BIOTINIDASE DEFICIENCY	*BTD*
XL AGAMMAGLOBULINEMIA	*BTK*
COMPLEMENT COMPONENT 3 DEFICIENCY	*C3*

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COMPLEMENT COMPONENT 5 DEFICIENCY	*C5*
COMPLEMENT COMPONENT 6 DEFICIENCY	*C6*
COMPLEMENT COMPONENT 7 DEFICIENCY	*C7*
COMPLEMENT COMPONENT 8 DEFICIENCY TYPE I	*C8a*
COMPLEMENT COMPONENT 8 DEFICIENCY TYPE II	*C8b*
COMPLEMENT COMPONENT 9 DEFICIENCY	*C9*
HYPERAMMONEMIA DUE TO CARBONIC ANHYDRASE VA DEFICIENCY	*CA5A*
LONG QT SYNDROME 8; TIMOTHY SYNDROME	*CACNA1C*
PRIMARY ALDOSTERONISM, SEIZURES, & NEUROLOGIC ABNORMALITIES	*CACNA1D*
CAD-CDG, EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY 50	*CAD*
NEONATAL SEVERE HYPERPARATHYROIDISM; AD HYPOCALCEMIA 1	*CASR*
INTRINSIC FACTOR DEFICIENCY	*CBLIF*
HOMOCYSTINURIA DUE TO CYSTATHIONINE BETA-SYNTHASE DEFICIENCY	*CBS*
IMMUNODEFICIENCY 19	*CD3D*
IMMUNODEFICIENCY 18	*CD3E*
AR AGAMMAGLOBULINEMIA 3	*CD79A*
AR AGAMMAGLOBULINEMIA 6	*CD79B*
COMPLEMENT FACTOR B DEFICIENCY	*CFB*
COMPLEMENT FACTOR D DEFICIENCY	*CFD*
XL PROPERDIN DEFICIENCY	*CFP*
CYSTIC FIBROSIS	*CFTR*
PRESYNAPTIC CONGENITAL MYASTHENIC SYNDROME 6,	*CHAT*
CONGENITAL. MYASTHENIC SYNDROME 1A, SLOW-CHANNEL; CONGENITAL MYASTHENIC SYNDROME 1B, FAST-CHANNEL	*CHRNA1*
CONGENITAL MYASTHENIC SYNDROME 2A, SLOW-CHANNEL; CONGENITAL MYASTHENIC SYNDROME 2C, ASSOC. WITH ACETYLCHOLINE RECEPTOR DEFICIENCY	*CHRNB1*
CONGENITAL MYASTHENIC SYNDROME 3A, SLOW-CHANNEL; CONGENITAL MYASTHENIC SYNDROME 3B, FAST-CHANNEL; CONGENITAL MYASTHENIC SYNDROME 3C, ASSOC. WITH ACETYLCHOLINE RECEPTOR DEFICIENCY	*CHRND*
CONGENITAL MYASTHENIC SYNDROME 4A, SLOW-CHANNEL; CONGENITAL MYASTHENIC SYNDROME 4B, FAST-CHANNEL; CONGENITAL MYASTHENIC SYNDROME 4C, ASSOC. WITH ACETYLCHOLINE RECEPTOR DEFICIENCY	*CHRNE*
AR OSTEOPETROSIS 4; AD OSTEOPETROSIS 2	*CLCN7*
CONGENITAL MYASTHENIC SYNDROME 19	*COL13A1*
CONGENITAL MYASTHENIC SYNDROME 5	*COLQ*
PRIMARY COENZYME Q10 DEFICIENCY 1	*COQ2*
PRIMARY COENZYME Q10 DEFICIENCY7	*COQ4*

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Disorder	Gene
PRIMARY COENZYME Q10 DEFICIENCY 6	*COQ6*
PRIMARY COENZYME Q10 DEFICIENCY 8	*COQ7*
PRIMARY COENZYME Q10 DEFICIENCY 4	*COQ8A*
NEPHROTIC SYNDROME TYPE 9	*COQ8B*
PRIMARY COENZYME Q10 DEFICIENCY 5	*COQ9*
HEREDITARY COPROPORPHYRIA	*CPOX*
HYPERAMMONEMIA DUE TO CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY	*CPS1*
CARNITINE PALMITOYLTRANSFERASE I DEFICIENCY	*CPT1A*
AR SEVERE NEUTROPENIA 7	*CSF3R*
CYSTINOSIS	*CTNS*
IMMUNODEFICIENCY 24	*CTPS1*
IMERSLUND-GRASBECK SYNDROME 1	*CUBN*
WHIM SYNDROME	*CXCR4*
AR CHRONIC GRANULOMATOUS DISEASE 4	*CYBA*
XL CHRONIC GRANULOMATOUS DISEASE	*CYBB*
AR CHRONIC GRANULOMATOUS DISEASE 5	*CYBC1*
CONGENITAL ADRENAL INSUFFICIENCY WITH PARTIAL OR COMPLETE 46,XY SEX REVERSAL	*CYP11A1*
CONGENITAL ADRENAL HYPERPLASIA DUE TO STEROID 11-β-HYDROXYLASE DEFICIENCY	*CYP11B1*
CORTICOSTERONE METHYLOXIDASE TYPE I DEFICIENCY; CORTICOSTERONE METHYLOXIDASE TYPE II DEFICIENCY	*CYP11B2*
CONGENITAL ADRENAL HYPERPLASIA DUE TO 17-ALPHA-HYDROXYLASE DEFICIENCY	*CYP17A1*
CONGENITAL ADRENAL HYPERPLASIA DUE TO 21-HYDROXYLASE DEFICIENCY	*CYP21A2**
CEREBROTENDINOUS XANTHOMATOSIS	*CYP27A1*
MAPLE SYRUP URINE DISEASE	*DBT*
OMENN SYNDROME	*DCLRE1C**
AROMATIC L-AMINO ACID DECARBOXYLASE DEFICIENCY	*DDC*
PROTEIN-LOSING ENTEROPATHY DIARRHEA 7	*DGAT1*
XL DYSKERATOSIS CONGENITA	*DKC1*
DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY	*DLD*
BECKER MUSCULAR DYSTROPHY; DUCHENNE MUSCULAR DYSTROPHY	*DMD*
AR HYPER-IgE RECURRENT INFECTION SYNDROME 2	*DOCK8*
CONGENITAL MYASTHENIC SYNDROME 10	*DOK7*
CONGENITAL MYASTHENIC SYNDROME 13	*DPAGT1*
CARDIOMYOPATHY, DILATED, WITH WOOLLY HAIR AND KERATODERMA; CARDIOMYOPATHY, DILATED, WITH WOOLLY HAIR, KERATODERMA, & TOOTH AGENESIS	*DSP*
SHWACHMAN-DIAMOND SYNDROME 2	*EFL1*
MULTIPLE EPIPHYSEAL DYSPLASIA WITH EARLY-ONSET DIABETES MELLITUS	*EIF2AK3*
CYCLIC NEUTROPENIA; SEVERE CONGENITAL AD NEUTROPENIA 1	*ELANE*
FANCONI ANEMIA, COMPLEMENTATION GROUP Q; XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP F	*ERCC4*
MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY	*ETFA*

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MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY	*ETFB*
MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY	*ETFDH*
ETHYLMALONIC ENCEPHALOPATHY	*ETHE1*
DEFICIENCY OF FACTOR XIII, A SUBUNIT	*F13A1*
DEFICIENCY OF FACTOR XIII, B SUBUNIT	*F13B*
CONGENITAL PROTHROMBIN DEFICIENCY	*F2*
HEMOPHILIA A	*F8**
HEMOPHILIA B	*F9*
TYROSINEMIA, TYPE I	*FAH*
FANCONI ANEMIA, COMPLEMENTATION GROUP A	*FANCA*
FANCONI ANEMIA, COMPLEMENTATION GROUP B; VACTERL WITH HYDROCEPHALUS	*FANCB*
FANCONI ANEMIA, COMPLEMENTATION GROUP C	*FANCC*
FANCONI ANEMIA, COMPLEMENTATION GROUP D2	*FANCD2*
FANCONI ANEMIA, COMPLEMENTATION GROUP E	*FANCE*
FANCONI ANEMIA, COMPLEMENTATION GROUP F	*FANCF*
FANCONI ANEMIA, COMPLEMENTATION GROUP G	*FANCG*
FANCONI ANEMIA, COMPLEMENTATION GROUP I	*FANCI*
FANCONI ANEMIA, COMPLEMENTATION GROUP L	*FANCL*
AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME	*FAS*
FRUCTOSE-1,6-BISPHOSPHATASE DEFICIENCY	*FBP1*
LEUKOCYTE ADHESION DEFICIENCY, TYPE III	*FERMT3*
CONGENITAL AFIBRINOGENEMIA; CONGENITAL DYSFIBRINOGENEMIA; CONGENITAL HYPODYSFIBRINOGENEMIA	*FGA*
CONGENITAL AFIBRINOGENEMIA; CONGENITAL DYSFIBRINOGENEMIA; CONGENITAL HYPODYSFIBRINOGENEMIA	*FGB*
CONGENITAL AFIBRINOGENEMIA; CONGENITAL DYSFIBRINOGENEMIA; CONGENITAL HYPODYSFIBRINOGENEMIA	*FGG*
LIPID STORAGE MYOPATHY D.T. FLAVIN ADENINE DINUCLEOTIDE SYNTHETASE DEFICIENCY	*FLAD1*
NEURODEGENERATION DUE TO CEREBRAL FOLATE TRANSPORT DEFICIENCY	*FOLR1*
COMBINED PITUITARY HORMONE DEFICIENCIES, GENETIC FORMS	*FOXA2*
T-CELL IMMUNODEFICIENCY, CONGENITAL ALOPECIA, AND NAIL DYSTROPHY; AD T-CELL LYMPHOPENIA, INFANTILE, WITH OR WITHOUT NAIL DYSTROPHY	*FOXN1*
XL IMMUNODYSREGULATION, POLYENDOCRINOPATHY, & ENTEROPATHY	*FOXP3*
GLYCOGEN STORAGE DISEASE Ia	*G6PC1*
AR SEVERE CONGENITAL NEUTROPENIA 4	*G6PC3*
NONSPHEROCYTIC HEMOLYTIC ANEMIA DUE TO G6PD DEFICIENCY	*G6PD*
GLYCOGEN STORAGE DISEASE II	*GAA*
GALACTOSEMIA III	*GALE*
GALACTOSEMIA II	*GALK1*

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Disorder	Gene
MUCOPOLYSACCHARIDOSIS IVA	*GALNS*
GALACTOSEMIA I	*GALT*
CEREBRAL CREATINE DEFICIENCY SYNDROME 2	*GAMT*
ANEMIA, XL, WITH/WITHOUT NEUTROPENIA AND/OR PLATELET ABNORMALITIES; BLACKFAN-DIAMOND ANEMIA; CONGENITAL ERYTHROPOIETIC PORPHYRIA; THROMBOCYTOPENIA, X-LINKED, WITH OR WITHOUT DYSERYTHROPOIETIC ANEMIA	*GATA1*
GAUCHER DISEASE type I; GAUCHER DISEASE type II; GAUCHER DISEASE type III; GAUCHER DISEASE type IIIc	*GBA1**
GLUTARIC ACIDEMIA I; GA1	*GCDH*
DOPA-RESPONSIVE DYSTONIA; BH4-DEFICIENT HYPERPHENYLALANINEMIA B	*GCH1*
PERMANENT NEONATAL DIABETES MELLITUS 1; FAM. HYPERINSULINEMIC HYPOGLYCEMIA 3	*GCK*
CONGENITAL MYASTHENIC SYNDROME 12	*GFPT1*
FABRY DISEASE	*GLA*
NEONATAL DIABETES MELLITUS WITH CONGENITAL HYPOTHYROIDISM	*GLIS3*
HYPEREKPLEXIA 1	*GLRA1*
HYPEREKPLEXIA 2	*GLRB*
FAMILIAL HYPERINSULINEMIC HYPOGLYCEMIA 6	*GLUD1*
EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY 82	*GOT2*
MUCOPOLYSACCHARIDOSIS TYPE VII	*GUSB*
FAMILIAL HYPERINSULINEMIC HYPOGLYCEMIA 4	*HADH*
LONG-CHAIN 3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY; MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY	*HADHA*
MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY	*HADHB*
AR SEVERE CONGENITAL NEUTROPENIA 3	*HAX1*
ALPHA-THALASSEMIA; HEMOGLOBIN H DISEASE	*HBA1**
ALPHA-THALASSEMIA; HEMOGLOBIN H DISEASE	*HBA2**
BETA-THALASSEMIA; SICKLE CELL ANEMIA	*HBB*
CBLX TYPE METHYLMALONIC ACIDEMIA AND HOMOCYSTEINEMIA	*HCFC1*
SEPTOOPTIC DYSPLASIA	*HESX1*
NONSPHEROCYTIC HEMOLYTIC ANEMIA DUE TO HEXOKINASE DEFICIENCY	*HK1*
HOLOCARBOXYLASE SYNTHETASE DEFICIENCY	*HLCS*
3-HYDROXY-3-METHYLGLUTARYL-CoA LYASE DEFICIENCY	*HMGCL*
HYPERINSULINISM DUE TO HNF1A DEFICIENCY; MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 3	*HNF1A*

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Disorder	Gene
FANCONI RENOTUBULAR SYNDROME 4 WITH MATURITY-ONSET DIABETES OF YOUNG; HYPERINSULINISM DUE TO HNF4A DEFICIENCY; MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 1	*HNF4A*
TYROSINEMIA, TYPE III	*HPD*
CONGENITAL ADRENAL HYPERPLASIA D.T. 3-β-HYDROXYSTEROID DEHYDROGENASE 2 DEFICIENCY	*HSD3B2*
MUCOPOLYSACCHARIDOSIS II	*IDS*
HURLER SYNDROME; HURLER-SCHEIE SYNDROME; SCHEIE SYNDROME	*IDUA**
AR AGAMMAGLOBULINEMIA 1	*IGHM*
AR AGAMMAGLOBULINEMIA 2	*IGLL1*
AR INFLAMMATORY BOWEL DISEASE 28	*IL10RA*
AR INFLAMMATORY BOWEL DISEASE 25	*IL10RB*
IMMUNODEFICIENCY 63 WITH LYMPHOPROLIFERATION AND AUTOIMMUNITY	*IL2RB*
XL COMBINED IMMUNODEFICIENCY	*IL2RG*
T CELL-NEG., B CELL-POS., NK CELL-POS.AR SEVERE COMBINED IMMUNODEFICIENCY	*IL7R*
PERMANENT NEONATAL DIABETES MELLITUS 4; MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 10	*INS*
IMMUNODEFICIENCY 67	*IRAK4*
LEUKOCYTE ADHESION DEFICIENCY, TYPE I	*ITGB2*
ISOVALERIC ACIDEMIA	*IVD*
T CELL-NEG., B CELL-POS., NK CELL-NEG. aR SEVERE COMBINED IMMUNODEFICIENCY	*JAK3*
PERMANENT NEONATAL DIABETES MELLITUS 2; TRANSIENT NEONATAL DIABETES MELLITUS 3; FAM. HYPERINSULINEMIC HYPOGLYCEMIA 2	*KCNJ11*
JERVELL AND LANGE-NIELSEN SYNDROME 1; LONG QT SYNDROME 1; SHORT QT SYNDROME 2	*KCNQ1*
EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY 7; BENIGN FAMILIAL NEONATAL SEIZURES 1	*KCNQ2*
EPILEPSY, NOCTURNAL FRONTAL LOBE, 5; EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY 14	*KCNT1*
COMBINED PITUITARY HORMONE DEFICIENCY 3	*LHX3*
COMBINED PITUITARY HORMONE DEFICIENCY 4	*LHX4*
LYSOSOMAL ACID LIPASE DEFICIENCY	*LIPA*
CBLF TYPE METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA	*LMBRD1*
CONGENITAL MYASTHENIC SYNDROME 17	*LRP4*
CHEDIAK-HIGASHI SYNDROME	*LYST*
α MANNOSIDOSIS, TYPES I and II	*MAN2B1*
GLUCOCORTICOID DEFICIENCY 1	*MC2R*
3-METHYLCROTONYL-CoA CARBOXYLASE 1 DEFICIENCY	*MCCC1*
3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY	*MCCC2*

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METHYLMALONIC ACIDURIA, cblA TYPE	*MMAA*
METHYLMALONIC ACIDURIA, cblB TYPE	*MMAB*
METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblC TYPE	*MMACHC*
METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblD TYPE	*MMADHC*
METHYLMALONIC ACIDURIA DUE TO METHYLMALONYL-CoA MUTASE DEFICIENCY	*MMUT*
MOLYBDENUM COFACTOR DEFICIENCY, COMPLEMENTATION GROUP A	*MOCS1*
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ib	*MPI*
GLUCOCORTICOID DEFICIENCY 2	*MRAP*
HOMOCYSTINURIA-MEGALOBLASTIC ANEMIA, cblG COMPLEMENTATION TYPE	*MTR*
HOMOCYSTINURIA-MEGALOBLASTIC ANEMIA, cblE COMPLEMENTATION TYPE	*MTRR*
CONGENITAL MYASTHENIC SYNDROME 9 ASSOC. WITH ACETYLCHOLINE RECEPTOR DEFICIENCY	*MUSK*
MEVALONIC ACIDURIA	*MVK*
BONE MARROW FAILURE SYNDROME 4	*MYSM1*
N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY	*NAGS*
EARLY ONSET PROGRESSIVE ENCEPHALOPATHY WITH BRAIN EDEMA +/- LEUKOENCEPHATLOPATHY	*NAXE*
NIJMEGEN BREAKAGE SYNDROME	*NBN*
AR CHRONIC GRANULOMATOUS DISEASE 1	*NCF1**
AR CHRONIC GRANULOMATOUS DISEASE 2	*NCF2*
AR CHRONIC GRANULOMATOUS DISEASE 3	*NCF4*
CONGENITAL MALABSORPTIVE DIARRHEA 4	*NEUROG3*
SCID WITH MICROCEPHALY, GROWTH RETARDATION & SENS. TO IONIZING RADIATION	*NHEJ1*
GLUCOCORTICOID DEFICIENCY 4 W./WITHOUT MINERALOCORTICOID DEFICIENCY	*NNT*
NIEMANN-PICK DISEASE C1	*NPC1*
NIEMANN-PICK DISEASE C2	*NPC2*
TYPE I AD PSEUDOHYPOALDOSTERONISM	*NR3C2*
46,XX SEX REVERSAL 4; 46,XY SEX REVERSAL 3	*NR5A1*
HYPERAMMONEMIA DUE TO ORNITHINE TRANSCARBAMYLASE DEFICIENCY	*OTC*
SUCCINYL-CoA:3-OXOACID-CoA TRANSFERASE DEFICIENCY	*OXCT1*
PHENYLKETONURIA	*PAH*
BH4-DEFICIENT HYPERPHENYLALANINEMIA D	*PCBD1*
PROPIONIC ACIDEMIA	*PCCA*
PROPIONIC ACIDEMIA	*PCCB*
PYRUVATE DEHYDROGENASE E1-α DEFICIENCY	*PDHA1*
PYRUVATE DEHYDROGENASE E1-β DEFICIENCY	*PDHB*
PYRUVATE DEHYDROGENASE PHOSPHATASE DEFICIENCY	*PDP1*
PRIMARY COENZYME Q10 DEFICIENCY 2	*PDSS1*
PRIMARY COENZYME Q10 DEFICIENCY 3	*PDSS2*
TYPE IT CONGENITAL DISORDER OF GLYCOSYLATION	*PGM1*
IMMUNODEFICIENCY 23	*PGM3*
XLD HYPOPHOSPHATEMIC RICKETS	*PHEX*
PHOSPHOGLYCERATE DEHYDROGENASE DEFICIENCY	*PHGDH*

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AR AGAMMAGLOBULINEMIA 7; IMMUNODEFICIENCY 36	*PIK3R1*
PYRUVATE KINASE DEFICIENCY OF RED CELLS	*PKLR*
PLASMINOGEN DEFICIENCY TYPE 1	*PLG*
EPILEPSY, EARLY-ONSET, VITAMIN B6-DEPENDENT	*PLPBP*
PYRIDOXAMINE 5-PRIME-PHOSPHATE OXIDASE DEFICIENCY	*PNPO*
ANTLEY-BIXLER SYNDROME WITH GENITAL ANOMALIES & DISORDERED STEROIDOGENESIS	*POR*
COMBINED PITUITARY HORMONE DEFICIENCY 1	*POU1F1*
CONGENITAL MYASTHENIC SYNDROME 22	*PREPL*
FAM. HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS 2	*PRF1*
IMMUNODEFICIENCY 26 WITH OR WITHOUT NEUROLOGIC ABNORMALITIES	*PRKDC*
COMBINED PITUITARY HORMONE DEFICIENCY 2	*PROP1*
PHOSPHOSERINE AMINOTRANSFERASE DEFICIENCY	*PSAT1*
PHOSPHOSERINE PHOSPHATASE DEFICIENCY	*PSPH*
PANCREATIC AGENESIS 2	*PTF1A*
BH4-DEFICIENT HYPERPHENYLALANINEMIA A	*PTS*
BH4-DEFICIENT HYPERPHENYLALANINEMIA C	*QDPR*
GRISCELLI SYNDROME, TYPE 2	*RAB27A*
α/β T-CELL LYMPHOPENIA WITH γ/δ T-CELL EXPANSION, SEVERE CMV INF. & AUTOIMMUNITY; COMBINED CELLULAR AND HUMORAL IMMUNE DEFECTS WITH GRANULOMAS; OMENN SYNDROME	*RAG1*
COMBINED CELLULAR AND HUMORAL IMMUNE DEFECTS WITH GRANULOMAS; OMENN SYNDROME	*RAG2*
CONGENITAL MYASTHENIC SYNDROME 11 ASSOC. WITH ACETYLCHOLINE RECEPTOR DEFICIENCY	*RAPSN*
TYPE II BARE LYMPHOCYTE SYNDROME	*RFXANK*
DIAMOND-BLACKFAN ANEMIA 7	*RPL11*
DIAMOND-BLACKFAN ANEMIA 12	*RPL15*
DIAMOND-BLACKFAN ANEMIA 18	*RPL18*
DIAMOND-BLACKFAN ANEMIA 11	*RPL26*
DIAMOND-BLACKFAN ANEMIA 16	*RPL27*
DIAMOND-BLACKFAN ANEMIA 5	*RPL35A*
DIAMOND-BLACKFAN ANEMIA 6	*RPL5*
DIAMOND-BLACKFAN ANEMIA 9	*RPS10*
DIAMOND-BLACKFAN ANEMIA 4	*RPS17**
DIAMOND-BLACKFAN ANEMIA 1	*RPS19*
DIAMOND-BLACKFAN ANEMIA 3	*RPS24*
DIAMOND-BLACKFAN ANEMIA 10	*RPS26*
DIAMOND-BLACKFAN ANEMIA 15 WITH MANDIBULOFACIAL DYSOSTOSIS	*RPS28*
DIAMOND-BLACKFAN ANEMIA 13	*RPS29*
DIAMOND-BLACKFAN ANEMIA 8	*RPS7*
MALIGNANT HYPERTHERMIA, SUSCEPTIBILITY TO, 1	*RYR1*
SHWACHMAN-DIAMOND SYNDROME 1	*SBDS**

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EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY 6; TYPE 2 GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS; MIGRAINE, FAM. HEMIPLEGIC, 3	*SCN1A*
EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY 11	*SCN2A*
EPILEPSY, FAM. FOCAL, WITH VARIABLE FOCI 4	*SCN3A*
HYPERKALEMIC PERIODIC PARALYSIS; HYPOKALEMIC PERIODIC PARALYSIS TYPE 2; CONGENITAL MYASTHENIC SYNDROME 16; PARAMYOTONIA CONGENITA OF VON EULENBURG	*SCN4A*
DILATED CARDIOMYOPATHY 1E; LONG QT SYNDROME 3; PROGRESSIVE FAMILIAL HEART BLOCK TYPE IA	*SCN5A*
EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY 13; BENIGN FAMILIAL INFANTILE SEIZURES 5	*SCN8A*
AR PSEUDOHYPOALDOSTERONISM TYPE I	*SCNN1A*
AR PSEUDOHYPOALDOSTERONISM TYPE I	*SCNN1B*
AR PSEUDOHYPOALDOSTERONISM TYPE I	*SCNN1G*
XL LYMPHOPROLIFERATIVE SYNDROME 1	*SH2D1A*
CONGENITAL SUCRASE-ISOMALTASE DEFICIENCY	*SI*
FAM. HYPERINSULINEMIC HYPOGLYCEMIA 7	*SLC16A1*
INFANTILE PARKINSONISM-DYSTONIA 2	*SLC18A2*
PRESYNAPTIC CONGENITAL MYASTHENIC SYNDROME 21	*SLC18A3*
THIAMINE METABOLISM DYSFUNCTION SYNDROME 2 (BIOTIN/THIAMINE-RESPONSIVE TYPE)	*SLC19A3*
PRIMARY SYSTEMIC CARNITINE DEFICIENCY	*SLC22A5*
COMBINED D-2- & L-2-HYDROXYGLUTARIC ACIDURIA; PRESYNAPTIC CONGENITAL MYASTHENIC SYNDROME 23	*SLC25A1*
NEONATAL-ONSET CITRULLINEMIA TYPE II	*SLC25A13*
HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA SYNDROME	*SLC25A15*
CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	*SLC25A20*
SUSC. TO IDIOPATHIC GENERALIZED EPILEPSY 12; GLUT1 DEFICIENCY SYNDROME 1; GLUT1 DEFICIENCY SYNDROME 2	*SLC2A1*
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IIc	*SLC35C1*
GLYCOGEN STORAGE DISEASE Ib	*SLC37A4*
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IIn	*SLC39A8*
CYSTINURIA	*SLC3A1*
HEREDITARY FOLATE MALABSORPTION	*SLC46A1*
BROWN-VIALETTO-VAN LAERE SYNDROME 2	*SLC52A2*
BROWN-VIALETTO-VAN LAERE SYNDROME 1; FAZIO-LONDE DISEASE	*SLC52A3*
GLUCOSE/GALACTOSE MALABSORPTION	*SLC5A1*
BIOTIN-RESPONSIVE CHILDHOOD-ONSET PERIPHERAL MOTOR NEUROPATHY; SODIUM-DEPENDENT MULTIVITAMIN TRANSPORTER DEFICIENCY	*SLC5A6*

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Gene List

Disorder	Gene
HYPEREKPLEXIA 3	*SLC6A5*
CREATINE TRANSPORT DEFICIENCY	*SLC6A8**
CYSTINURIA	*SLC7A9*
FANCONI ANEMIA, COMPLEMENTATION GROUP P	*SLX4*
SPINAL MUSCULAR ATROPHY TYPE I; SPINAL MUSCULAR ATROPHY TYPE II; SPINAL MUSCULAR ATROPHY TYPE III	*SMN1*
NIEMANN-PICK A; NIEMANN-PICK B	*SMPD1*
CONGENITAL MYASTHENIC SYNDROME 18	*SNAP25*
DYSTONIA, DOPA-RESPONSIVE DUE TO SEPIAPTERIN REDUCTASE DEFICIENCY	*SPR*
AD SEVERE CONGENITAL NEUTROPENIA 8	*SRP54*
LIPOID CONGENITAL ADRENAL HYPERPLASIA	*STAR*
FAM. HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS 4	*STX11*
FAM. HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS 5	*STXBP2*
BARTH SYNDROME	*TAFAZZIN*
TYROSINEMIA, TYPE II	*TAT*
ISOLATED ACTH DEFICIENCY	*TBX19*
AGAMMAGLOBULINEMIA 8	*TCF3*
TRANSCOBALAMIN II DEFICIENCY	*TCN2*
SEGAWA SYNDROME	*TH*
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IIk	*TMEM165*
OSTEOPETROSIS 7	*TNFRSF11A*
NEURONAL CEROID LIPOFUSCINOSIS 2	*TPP1*
TUBEROUS SCLEROSIS 1	*TSC1*
TUBEROUS SCLEROSIS 2	*TSC2*
GASTROINTESTINAL DEFECTS AND IMMUNODEFICIENCY SYNDROME	*TTC7A*
ATAXIA WITH ISOLATED VITAMIN E DEFICIENCY	*TTPA*
FANCONI ANEMIA, COMPLEMENTATION GROUP T	*UBE2T*
FAM. HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS 3	*UNC13D*
CONGENITAL MYASTHENIC SYNDROME 25, PRESYNAPTIC	*VAMP1*
SEVERE CONGENITAL NEUTROPENIA 5	*VPS45*
XL SEVERE CONGENITAL NEUTROPENIA; THROMBOCYTOPENIA 1; WISKOTT-ALDRICH SYNDROME	*WAS*
LYMPHOPROLIFERATIVE SYNDROME 2	*XIAP*

*genes with analytical challenges due to current sequencing and bioinformatic laboratory practices