Supplementary Table 1. The List of 198 Genes

Gene	Genetic disorder	Inheritance	Phenotype MIM number
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type	AR	614857
ACAD8	Isobutyryl-CoA dehydrogenase deficiency	AR	611283
ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency of	AR	201450
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of	AR	201470
ACADSB	2-methylbutyrylglycinuria	AR	610006
ACADVL	Very long-chain acyl-CoA dehydrogenase deficiency	AR	201475
ACSF3	Combined malonic and methylmalonic aciduria	AR	614265
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2	AD	600376
AGL	Glycogen storage disease IIIa, Glycogen storage disease IIIb	AR	232400
AIPL1	Leber congenital amaurosis-4 (LCA4)	AR	604393
ALDH4A1	Hyperprolinemia, type II	AR	239510
ALDOB	Fructose intolerance, hereditary	AR	229600
ALPL	Hypophosphatasia, infantile	AR	241500
ARFGEF2	Periventricular heterotopia with microcephaly	AR	608097
ARSA	Metachromatic leukodystrophy	AR	250100
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	AR	253200
ARX	Epileptic encephalopathy, early infantile, 1	XLR	308350
ASL	Argininosuccinic aciduria	AR	207900
ASPA	Canavan disease	AR	271900
ASS1	Citrullinemia	AR	215700
ATM	Ataxia-telangiectasia	AR	208900
ATP7A	Menkes disease	XLR	309400
ATP7B	Wilson disease	AR	253200
ATPBD1	Cholestasis, progressive familial intrahepatic 1	AR	211600
AUH	3-methylglutaconic aciduria, type I	AR	250950
BCAT1	Hyperleucinemia-isoleucinemia or hypervalinemia	AR	113520
BCAT2	Hypervalinemia or hyperleucine-isoleucinemia	AR	113530
BMPR2	Pulmonary hypertension, familial primary, 1, with or without hereditary hemorrhagic telangiectasia	AD	178600
BRAF	Cardiofaciocutaneous syndrome	AD	115150
BTD	Biotinidase deficiency	AR	253260
BTK	Agammaglobulinemia, X-linked 1	XLR	300755
C2CD3	Orofaciodigital syndrome XIV	AR	615948
C5orf42	Orofaciodigital syndrome VI,/Joubert syndrome 17	AR	277170/614615
CACNA1S	Hypokalemic periodic paralysis, type 1	AD	170400
CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect	AR	613646
CDKL5	Epileptic encephalopathy, early infantile, 2	XLD	300672
CEP290	Leber congenital amaurosis 10/Joubert syndrome 5	AR	611755/610188
CFTR	Cystic fibrosis	AR	219700
CHD7	CHARGE syndrome	AD	214800
CLCN7	Osteopetrosis, autosomal dominant 2/Osteopetrosis, autosomal recessive 4	AD/AR	166600/611490
CLN3	Ceroid lipofuscinosis, neuronal, 3	AR	204200
CLPB	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia	AR	616271
CNBP	Myotonic dystrophy 2	AD	602668
COL11A1	Marshall syndrome/Stickler syndrome, type II	AD	154780/604841
COL11A2	Otospondylomegaepiphyseal dysplasia	AD/AR	184840/215150
COL1A2	Osteogenesis imperfecta, type III/typeIV	AD	259420/166220
COL2A1	Achondrogenesis, type II or hypochondrogenesis	AD	200610
COL4A3	Alport syndrome, autosomal dominant/autosomal recessive	AD/AR	104200/203780
COL4A4	Alport syndrome, autosomal recessive	AR	203780
COL4A5	Alport syndrome	XLD	301050
COL9A1	Epiphyseal dysplasia, multiple, 6/Stickler syndrome, type IV	AD/AR	614135/614134
COMP	Epiphyseal dysplasia, multiple, 1	AD	132400
CPT1A	Carnitine palmitoyltransferase deficiency, hepatic, type IA	AR	255120
CPT2	Carnitine palmitoyltransferase II deficiency, infantile	AR	600649
CTNS	Cystinosis, nephropathic	AR	219800
CYBB	Chronic granulomatous disease, X-linked	XLR	306400
CYP21A2	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	AR	201910
DDX59	Orofaciodigital syndrome V	AR	174300
DES	Cardiomyopathy, dilated, 1I/Muscular dystrophy, limb-girdle, type 2R	AD/AR	604765/615325
DHCR7	Smith-Lemli-Opitz syndrome	AR	270400
DLAT	Pyruvate dehydrogenase E2 deficiency	AR	245348
DMD	Duchenne muscular dystrophy	XLR	310200
DNAJC19	3-methylglutaconic aciduria, type V	AR	610198
DUOX2	Thyroid dyshormonogenesis 6	AR	607200
ENG	Telangiectasia, hereditary hemorrhagic, type 1	AD	187300
ETHE1	Ethylmalonic encephalopathy	AR	602473
EYA1	Otofaciocervical syndrome/Branchiootic syndrome 1	AD	166780/602588
F8	Hemophilia A	XLR	306700
FAH	Tyrosinemia, type I	AR	276700
FBN1	Marfan syndrome	AD	154700
FGFR1	Pfeiffer syndrome	AD	101600
FGFR2	Apert syndrome /Crouzon syndrome	AD	101200/123500
FGFR3	Achondroplasia/Hypochondroplasia	AD	100800/146000
FLNA	Congenital short bowel syndrome/Heterotopia, periventricular	XLR/XLD	300048/300049
G6PC	Glycogen storage disease Ia	AR	232200
GAA	Glycogen storage disease II	AR	232300
GALC	Krabbe disease	AR	245200
GALK1	Galactokinase deficiency with cataracts	AR	230200
GALT	Galactosemia	AR	230400
GBA	Gaucher disease I, II, III	AR	230800
GCDH	Glutaricaciduria, type I	AR	231670
GCH1	Dystonia, DOPA-responsive/Hyperphenylalaninemia, BH4-deficient, B	AD/AR	128230/233910
GLA	Fabry disease	XL	301500
GLB1	GM1-gangliosidosis, type I/Mucopolysaccharidosis type IV B (Morquio)	AR	230500/253010
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism	AR	610199
GNAS	Pseudohypoparathyroidism Ia/Ib/Ic	AD	103580/603233/612462
GUCY2D	Leber congenital amaurosis 1	AR	204000
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency /Hyperinsulinemic hypoglycemia, familial, 4	AR	231530/609975
HADHA	Long-chain hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency/Trifunctional protein deficiency	AR	609016/609015
HADHB	Trifunctional protein deficiency	AR	609015
HAL	Histidinemia	AD, AR	235800
HBA1	Thalassemias, alpha-	AD	604131
HBA2	Thalassemias, alpha-	AD	604131
HBB	Delta-beta thalassemia/Sickle cell anemia	AD/AR	141749/603903
HLCS	Holocarboxylase synthetase deficiency	AR	253270
HMGCL	HMG-CoA lyase deficiency	AR	246450
HSD17B10	HSD10 mitochondrial disease	XLD	300438
IDS	Mucopolysaccharidosis II	XLR	309900
IDUA	Mucopolysaccharidosis Ih/Ihs/Is	AS	607014/607015/607016
IGSF1	Hypothyroidism, central, and testicular enlargement	XLR	300888
IKBKG	Incontinentia pigmenti	XLD	308300
IL2RG	Severe combined immunodeficiency, X-linked	XLR	300400
IVD	Isovaleric acidemia	AR	243500
IYD	Thyroid dyshormonogenesis 4	AR	274800
JAG1	Alagille syndrome 1	AD	118450
KIF1B	Charcot-Marie-Tooth disease, type 2A1	AD	118210
KRAS	Noonan syndrome 3	AD	609942
L1CAM	MASA (mental retardation, aphasia, shuffling gait, and adducted thumbs) syndrome, CRASH syndrome	XLR	303350
LMNA	Muscular dystrophy, congenital/Emery-Dreifuss muscular dystrophy 2, AD/AR/Charcot-Marie-Tooth disease, type 2B1	AD/AD/AD/AR	613205/181350/616516/605588
LRP5	Osteopetrosis, autosomal dominant 1/Osteoporosis-pseudoglioma syndrome	AD/AR	607634/259770
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency	AR	210200
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency	AR	210210
MECP2	Rett syndrome	XLD	312750
MFN2	Charcot-Marie-Tooth disease, axonal, type 2A2A/2A2B	AD/AR	609260/617087
MLYCD	Malonyl-CoA decarboxylase deficiency	AR	248360
MUT	Methylmalonic aciduria, mut(0) type	AR	251000
NF1	Neurofibromatosis, type 1	AD	162200
NF2	Neurofibromatosis, type 2	AD	101000
NKX2-1	Choreoathetosis, hypothyroidism, and neonatal respiratory distress	AD	610978
NKX2-5	Hypothyroidism, congenital nongoitrous, 5	AD	225250
NRAS	Noonan syndrome 6	AD	613224
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia	AR	258870
OFD1	Orofaciodigital syndrome I /Joubert syndrome 10	XLD/XLR	311200/300804
OPA3	3-methylglutaconic aciduria, type III/Optic atrophy 3 with cataract	AR/AD	258501/165300
OSTM1	Osteopetrosis, autosomal recessive 5	AR	259720
OTC	Ornithine transcarbamylase deficiency	XLR	311250
PAH	Phenylketonuria	AR	261600
PCBD1	Hyperphenylalaninemia, BH4-deficient, D	AR	264070
PCCA	Propionicacidemia	AR	606054
PCCB	Propionicacidemia	AR	606054
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	XLD	312170
PDHB	Pyruvate dehydrogenase E1-beta deficiency	AR?	614111
PEX1	Peroxisome biogenesis disorder 1A (Zellweger)	AR	214100
PEX10	Peroxisome biogenesis disorder 6A (Zellweger)/6B	AR	614870/614871
PEX12	Peroxisome biogenesis disorder 3A (Zellweger)/3B	AR	614859/266510
PEX13	Peroxisome biogenesis disorder 11A (Zellweger)/11B	AR	614883/614885
PEX14	Peroxisome biogenesis disorder 13A (Zellweger)	AR	614887
PEX16	Peroxisome biogenesis disorder 8A (Zellweger)/8B	AR	614876/614877
PEX19	Peroxisome biogenesis disorder 12A (Zellweger)	AR	614886
PEX2	Peroxisome biogenesis disorder 5A (Zellweger)/5B	AR	614866/614867
PEX26	Peroxisome biogenesis disorder 7A (Zellweger)/7B	AR	614872/614873
PEX3	Peroxisome biogenesis disorder 10A (Zellweger)/10B	AR	614882/617370
PEX5	Peroxisome biogenesis disorder 2A (Zellweger)/2B	AR	214110/202370
PEX6	Peroxisome biogenesis disorder 4A (Zellweger)/4B	AR	614862/614863
PHEX	Hypophosphatemic rickets, X-linked dominant	XLD	307800
PHOX2B	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease	AD	209880
PKD1	Polycystic kidney disease 1	AD	173900
PKD2	Polycystic kidney disease 2	AD	613095
PKHD1	Polycystic kidney disease 4, with or without hepatic disease	AR	263200
PLEKHM1	Osteopetrosis, autosomal recessive 6	AR	611497
PMP22	Charcot-Marie-Tooth disease, type 1A/1E	AD	118220/118300
POLR1C	Leukodystrophy, hypomyelinating, 11/Treacher Collins syndrome 3	AR	616494/248390
POLR1D	Treacher Collins syndrome 2	AD/AR	613717
PRODH	Hyperprolinemia, type I	AR	239500
PSAP	Krabbe disease, atypical/Gaucher disease, atypical/ Metachromatic leukodystrophy due to SAP-b deficiency	AR	611722/610539/249900
PTPN11	Noonan syndrome 1	AD	163950
PTS	Hyperphenylalaninemia, BH4-deficient, A	AR	261640
QDPR	Hyperphenylalaninemia, BH4-deficient, C	AR	261630
RAF1	Noonan syndrome 5	AD	611553
RBMX	Mental retardation, X-linked, syndromic 11, Shashi type	XLR	300238
RPE65	Leber congenital amaurosis 2	AR	204100
RPGRIP1	Leber congenital amaurosis 6	AR	613826
RPS6KA3	Coffin-Lowry syndrome/Mental retardation, X-linked 19	XLD	303600/300844
RS1	Retinoschisis	XLR	312700
SCN1A	Epileptic encephalopathy, early infantile, 6 (Dravet syndrome)	AD	607208
SCN4A	Myotonia congenita, atypical, acetazolamide-responsive/Hypokalemic periodic paralysis, type 2	AD	608390/613345
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	AR	614739
SERPINA1	Emphysema due to alpha-1-antitrypsin deficiency	AR	613490
SIX1	Branchiootic syndrome 3	AD	608389
SIX5	Branchiootorenal syndrome 2	AD	610896
SLC22A5	Carnitine deficiency, systemic primary	AR	212140
SLC25A13	Citrullinemia, type II, neonatal-onset	AR	605814
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	AR	238970
SLC37A4	Glycogen storage disease Ib/Ic	AR	232220/232240
SLC5A5	Thyroid dyshormonogenesis 1	AR	274400
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome	AD	175050
SNX10	Osteopetrosis, autosomal recessive 8	AR	615085
SOS1	Noonan syndrome 4	AD	610733
SPATA7	Leber congenital amaurosis 3	AR	604232
STXI1	Hemophagocytic lymphohistiocytosis, familial, 4	AR	603552
STX16	Pseudohypoparathyroidism, type IB	AD	603233
TAZ	Barth syndrome	XLR	302060
TCIRG1	Osteopetrosis, autosomal recessive 1	AR	259700
TCOF1	Treacher Collins syndrome 1	AD	154500
TCTN3	Orofaciodigital syndrome IV,/Joubert syndrome 18	AR	258860/614815
THRA	Hypothyroidism, congenital, nongoitrous, 6	AD	614450
TNFRSF11A	Osteopetrosis, autosomal recessive 7/Osteolysis, familial expansile	AR/AD	612301/174810
TNFSF11	Osteopetrosis, autosomal recessive 2	AR	259710
TSC1	Tuberous sclerosis-1	AD	191100
TSC2	Tuberous sclerosis-2	AD	613254
TSHB	Hypothyroidism, congenital, nongoitrous 4	AR	275100
TSHR	Hypothyroidism, congenital, nongoitrous, 1/Hyperthyroidism, nonautoimmune	AR/AD	275200/609152
UGT1A1	Crigler-Najjar syndrome, type I/II	AR	218800/606785
USH2A	Usher syndrome, type 2A	AR	276901
VHL	von Hippel-Lindau syndrome	AD	193300
VPS33B	Arthrogryposis, renal dysfunction, and cholestasis 1	AR	208085
VWF	von Willebrand disease, type 1/von Willebrand disease, types 2A, 2B, 2M, and 2N	AD/AR	193400/613554
WT1	Wilms tumor, type 1 /Denys-Drash syndrome	AD	194070/194080

MIM, Mendelian Inheritance in Man; AD, autosomal dominat; AR, autosomal recessive; XLD, X-linked dominant; XLR, X-linked recessive.