No.	diseases classification	broad category of diseases
1	Metabolic-amino acid metabolic diseases	hyperphenylalaninemia (HPA)
	Metabolic-amino acid metabolic diseases	
	Metabolic-amino acid metabolic diseases	
	Metabolic-amino acid metabolic diseases	
	Metabolic-amino acid metabolic diseases	
	Metabolic-amino acid metabolic diseases	
2	Metabolic-amino acid metabolic diseases	Carbamoyl phosphate synthase deficiency
3	Metabolic-amino acid metabolic diseases	Maple Diabetes
	Metabolic-amino acid metabolic diseases	
	Metabolic-amino acid metabolic diseases	
	Metabolic-amino acid metabolic diseases	
4	Metabolic-amino acid metabolic diseases	glycine encephalopathy (GEE)
5	Metabolic-amino acid metabolic diseases	Hypermethioninemia
	Metabolic-amino acid metabolic diseases	
	Metabolic-amino acid metabolic diseases	
	Metabolic-amino acid metabolic diseases	
6	Metabolic-amino acid metabolic diseases	ornithinemia-Hyperammonemia-Homocitrullin
7	Metabolic-amino acid metabolic diseases	Hyperprolinemia
	Metabolic-amino acid metabolic diseases	
8	Metabolic-amino acid metabolic diseases	Homocysteinemia
	Metabolic-amino acid metabolic diseases	
	Metabolic-amino acid metabolic diseases	
	Metabolic-amino acid metabolic diseases	
9	Metabolic-amino acid metabolic diseases	Citrullinemia
	Metabolic-amino acid metabolic diseases	
10	Metabolic-amino acid metabolic diseases	Argininosuccinic Aciduria
11	Metabolic-amino acid metabolic diseases	Arginase Deficiency
12	Metabolic-amino acid metabolic diseases	Tyrosinemia
	Metabolic-amino acid metabolic diseases	
	Metabolic-amino acid metabolic diseases	
13	Metabolic-amino acid metabolic diseases	Ornithine Carbamyltransferase Deficiency
14	Metabolic-amino acid metabolic diseases	N-Acetylglutamate Synthetase Deficiency
15	Metabolic-amino acid metabolic diseases	Hypervalinemia
16	Metabolic-amino acid metabolic diseases	Histidinemia
17	Metabolic-organic acid metabolism disease	2-Methylbutyrylglycineuria
18	Metabolic-organic acid metabolism disease	3-Methylcrotonyl coenzyme A carboxylase deficiency
	Metabolic-organic acid metabolism disease	
19	Metabolic-organic acid metabolism disease	3-Methylglutaconic aciduria
	Metabolic-organic acid metabolism disease	
	Metabolic-organic acid metabolism disease	
	Metabolic-organic acid metabolism disease	
	Metabolic-organic acid metabolism disease	
	Metabolic-organic acid metabolism disease	
20	Metabolic-organic acid metabolism disease	3-Hydroxy-3-methylglutaryl coenzyme A cleavage enzyme deficiency
21	Metabolic-organic acid metabolism disease	3-Hydroxy-3-methylglutaryl coenzyme A synthetase 2 deficiency
22	Metabolic-organic acid metabolism disease	Beta-ketothionein deficiency
23	Metabolic-organic acid metabolism disease	Malonyl-CoA decarboxylase deficiency
24	Metabolic-organic acid metabolism disease	Propionic acidemia

25	Metabolic-organic acid metabolism disease	Succinate semialdehyde dehydrogenase deficiency
26	Metabolic-organic acid metabolism disease	Methylmalonic acidemia
	Metabolic-organic acid metabolism disease	
	Metabolic-organic acid metabolism disease	
	Metabolic-organic acid metabolism disease	
	Metabolic-organic acid metabolism disease	
	Metabolic-organic acid metabolism disease	
	Metabolic-organic acid metabolism disease	
	Metabolic-organic acid metabolism disease	
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	Metabolic-organic acid metabolism disease	
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	Metabolic-organic acid metabolism disease	
	Metabolic-organic acid metabolism disease	
	Metabolic-organic acid metabolism disease	
27	Metabolic-organic acid metabolism disease	Fully carboxylase synthetase deficiency
28	Metabolic-organic acid metabolism disease	Biotinidase deficiency
29	Metabolic-organic acid metabolism disease	Glutaric acidemia
30	Metabolic-organic acid metabolism disease	-hydroxybutyryl coenzyme A dehydrogenase
31	Metabolic-organic acid metabolism disease	obutyryl Coenzyme A Dehydrogenase Deficier
32	Metabolic-organic acid metabolism disease	Isovaleric acidemia
33	Metabolic-fatty acid metabolism disease	,4-Dienoyl Coenzyme A Reductase Deficienc
34	Metabolic-fatty acid metabolism disease	-chain acyl-coenzyme A dehydrogenase defic
35	Metabolic-fatty acid metabolism disease	Multiple acyl coenzyme A dehydrogenase deficiency
	Metabolic-fatty acid metabolism disease	
	Metabolic-fatty acid metabolism disease	
36	Metabolic-fatty acid metabolism disease	ng chain acyl coenzyme A dehydrogenase def
37	Metabolic-fatty acid metabolism disease	rnitine-acylcarnitine transferase deficier
38	Metabolic-fatty acid metabolism disease	Carnitine palmitoyltransferase deficiency
	Metabolic-fatty acid metabolism disease	
39	Metabolic-fatty acid metabolism disease	Trifunctional Group Protein Deficiency
40	Metabolic-fatty acid metabolism disease	Primary carnitine deficiency
41	Metabolic-fatty acid metabolism disease	n 3-hydroxyacyl-coenzyme A dehydrogenase d
42	Metabolic-fatty acid metabolism disease	chain acyl coenzyme A dehydrogenase defi
43	Metabolic-fatty acid metabolism disease	hain 3-ketoacyl coenzyme A sulfurylase de
44	Metabolic-fatty acid metabolism disease	droxyacyl coenzyme A dehydrogenase defici
45	Metabolic-fatty acid metabolism disease	Ethylmalonic acid encephalopathy
46	Metabolic-Lysosomal Storage Disease	Krabbe diseases
47	Metabolic-Lysosomal Storage Disease	Fabry diseases
48	Metabolic-Lysosomal Storage Disease	Niemann-Pick disease
	Metabolic-Lysosomal Storage Disease	
	Metabolic-Lysosomal Storage Disease	
49	Metabolic-Lysosomal Storage Disease	Mucopolysaccharide storage disease
	Metabolic-Lysosomal Storage Disease	
	Metabolic-Lysosomal Storage Disease	
	Metabolic-Lysosomal Storage Disease	
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	Metabolic-Lysosomal Storage Disease	
	Metabolic-Lysosomal Storage Disease	
	Metabolic-Lysosomal Storage Disease	

	Metabolic-Lysosomal Storage Disease	
50	Metabolic-Lysosomal Storage Disease	Heterotrophic cerebral leukodystrophy
51	Metabolic-Lysosomal Storage Disease	Ganglioside Storage Disease
	Metabolic-Lysosomal Storage Disease	
	Metabolic-Lysosomal Storage Disease	
	Metabolic-Lysosomal Storage Disease	
	Metabolic-Lysosomal Storage Disease	
	Metabolic-Lysosomal Storage Disease	
52	Metabolic-Glycometabolic Diseases	Galactosemia
	Metabolic-Glycometabolic Diseases	
	Metabolic-Glycometabolic Diseases	
53	Metabolic-Glycometabolic Diseases	Glycogen Accumulation Disease
	Metabolic-Glycometabolic Diseases	
	Metabolic-Glycometabolic Diseases	
	Metabolic-Glycometabolic Diseases	
	Metabolic-Glycometabolic Diseases	
	Metabolic-Glycometabolic Diseases	
	Metabolic-Glycometabolic Diseases	
	Metabolic-Glycometabolic Diseases	
	Metabolic-Glycometabolic Diseases	
	Metabolic-Glycometabolic Diseases	
	Metabolic-Glycometabolic Diseases	
	Metabolic-Glycometabolic Diseases	
54	Metabolic-Glycometabolic Diseases	Hereditary fructose intolerance
55	Metabolic-peroxisomal disease	Disorders of peroxisome biosynthesis
	Metabolic-peroxisomal disease	
	Metabolic-peroxisomal disease	
	Metabolic-peroxisomal disease	
	Metabolic-peroxisomal disease	
	Metabolic-peroxisomal disease	
	Metabolic-peroxisomal disease	
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	Metabolic-peroxisomal disease	
	Metabolic-peroxisomal disease	
	Metabolic-peroxisomal disease	
	Metabolic-peroxisomal disease	
56	Metabolic-peroxisomal disease	Primary hyperoxaluria
	Metabolic-peroxisomal disease	
	Metabolic-peroxisomal disease	
57	Metabolic-peroxisomal disease	Glutaric acidemia type III
58	Metabolic-Lipid Metabolism Disease/Other	Glutathionemia
	Metabolic-Lipid Metabolism Disease/Others	
59	Metabolic-lipid metabolism disease/other	Familial hypercholesterolemia
60	Metabolic-lipid metabolism disease/other	Tendon xanthomatosis
61	Metabolism-Other	Menkes disease
62	Metabolism-Other	Hypophosphatemic rickets
63	Metabolism-Other	Hepatomegaly
64	Metabolism-other	Progressive familial intrahepatic cholestasis
	Metabolism-other	

	Metabolize-other	cholestasis
65	Metabolize-other	lucose-6-phosphate dehydrogenase deficienc
66	Metabolize-other	Congenital bile acid synthesis disorder
67	Metabolize-other	Hypoalkaline phosphatasemia
68	Metabolize-other	Dihydrolipoic acid dehydrogenase deficiency
69	Metabolize-other	**Brain Creatine Deficiency Syndrome**
	Metabolize-other	
70	Metabolize-other	Alagille syndrome
	Metabolize-other	
71	Metabolize-other	Crigler-Najjar syndrome
	Metabolize-other	
72	deafness	Pendred syndrome
73	deafness	Usher syndrome
	deafness	
	deafness	
	deafness	
	deafness	
74	deafness	drug-induced deafness
75	deafness	Autosomal Recessive Deafness
	deafness	
	deafness	
	deafness	
	deafness	
	deafness	
	deafness	
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	deafness	
76	deafness	Autosomal Dominant Deafness
	deafness	
77	deafness	Waardenburg Syndrome
78	Immune system diseases	Thrombocytopenia in Eczema with Immunodeficiency Syndrome
79	Immune system diseases	X-linked lymphoproliferative disorder
	Immune system diseases	
80	Immune system diseases	X-linked agammaglobulinemia
81	Immune system diseases	Chronic Granulomatous Disease
82	Immune system diseases	Primary combined immunodeficiency
	Immune system diseases	
	Immune system diseases	
	Immune system diseases	
	Immune system diseases	
83	Immune system diseases	Familial Mediterranean Fever
84	Immune system diseases	Immunodeficiency with elevated IgM
85	Immune system diseases	Severe Congenital Neutrophil Deficiency
86	Endocrine system diseases	Congenital adrenal hyperplasia
	Endocrine system diseases	

86	Endocrine system diseases	Congenital adrenal hyperplasia
	Endocrine system diseases	
87	Endocrine system diseases	Kallmann syndrome
	Endocrine system diseases	
	Endocrine system diseases	
	Endocrine system diseases	
88	Endocrine system diseases	Congenital Adrenal Hypoplasia
89	Endocrine system diseases	Congenital hypothyroidism
	Endocrine system diseases	
	Endocrine system diseases	
	Endocrine system diseases	
	Endocrine system diseases	
	Endocrine system diseases	
	Endocrine system diseases	
90	Endocrine system diseases	Neonatal diabetes mellitus
	Endocrine system diseases	
91	Endocrine system diseases	Familial Hyperinsulinemia
	Endocrine system diseases	
	Endocrine system diseases	
	Endocrine system diseases	
92	Neuromuscular diseases	Pyridoxine-dependent epilepsy
93	Neuromuscular diseases	Hereditary spastic paraplegia
	Neuromuscular diseases	
	Neuromuscular diseases	
	Neuromuscular diseases	
94	Neuromuscular diseases	Congenital myasthenia gravis
95	Neuromuscular diseases	Progressive muscular dystrophy
96	Neuromuscular diseases	Spinal Muscular Atrophy
97	Neuromuscular diseases	Dopa-responsive dystonia
	Neuromuscular diseases	
98	Neuromuscular diseases	Glucose transporter protein 1 deficiency syndrome
99	Neuromuscular diseases	Severe myoclonic epilepsy in infants
	Neuromuscular diseases	
100	Neuromuscular diseases	Brown-Vialetto-Van Laere syndrome
	Neuromuscular diseases	
101	Diseases of the blood system	Congenital Pure Red Blood Cell Aplastic Anemia
	Diseases of the blood system	
	Diseases of the blood system	
102	Diseases of the blood system	Thalassemia
	Diseases of the blood system	
103	Diseases of the blood system	Fanconi anemia
104	Diseases of the blood system	Familial hemophagocytic lymphohistiocytosis
	Diseases of the blood system	
105	Diseases of the blood system	Transcobalamin II deficiency
106	others	Gitelman syndrome
107	others	Leber’s Hereditary Optic Neuropathy
108	others	Alport syndrome
	others	
	others	

109	others	Nodular sclerosis
	others	
110	others	Cystic fibrosis
111	others	Retinoblastoma
112	others	Primary Coenzyme Q10 Deficiency

Disease subtype	Gene	inheritance pattern
Phenylketonuria	*PAH*	AR
BH4-Deficient Hyperphenylalaninemia A	*PTS*	AR
BH4-Deficient Hyperphenylalaninemia B	*GCH1*	AR
BH4-Deficient Hyperphenylalaninemia C	*QDPR*	AR
BH4-Deficient Hyperphenylalaninemia D	*PCBD1*	AR
Mild non-BH4-deficient Hyperphenylalani	*DNAJC12*	AR
Carbamoylphosphate Synthetase I Deficie	*CPS1*	AR
Maple Syrup Urine Disease,type Ia	*BCKDHA*	AR
Maple Syrup Urine Disease,type Ib	*BCKDHB*	AR
Maple Syrup Urine Disease,type II	*DBT*	AR
Mild Variant Maple Syrup Urine Disease	*PPM1K*	AR
Glycine Encephalopathy	*AMT*、*GLDC*、*GCSH*	AR
Hypermethioninemia	*MAT1A*	AR
Hypermethioninemia with S-Adenosylhomoc	*AHCY*	AR
Hypermethioninemia due to Adenosine Kir	*ADK*	AR
Glycine N-Methyltransferase Deficiency	*GNMT*	AR
Hyperornithinemia-Hyperammonemia- Homoc	*SLC25A15*	AR
Hyperprolinemia Type I	*PRODH*	AR
Hyperprolinemia Type II	*ALDH4A1*	AR
Homocystinuria Due to Cystathionine Bet	*CBS*	AR
Homocystinuria Due to MTHFR Deficiency	*MTHFR*	AR
Homocystinuria-Megaloblastic Anemia Cbl	*MTR*	AR
Homocystinuria-Megaloblastic Anemia Cbl	*MTRR*	AR
Citrullinemia, Type II, Neonatal-Onset	*SLC25A13*	AR
Citrullinemia, Type I	*ASS1*	AR
Argininosuccinic Aciduria	*ASL*	AR
Argininemia	*ARG1*	AR
Tyrosinemia Type I	*FAH*	AR
Tyrosinemia Type II	*TAT*	AR
Tyrosinemia Type III	*HPD*	AR
Ornithine Transcarbamylase Deficiency	*OTC*	XL
N-Acetylglutamate Synthase Deficiency	*NAGS*	AR
Hypervalinemia	*BCAT1*、*BCAT2*	AR
Histidinemia	*HAL*	AD,AR
2-Methylbutyryl Glycinuria	*ACADSB*	AR
3-Methylcrotonyl-CoA Carboxylase 1 Defi	*MCCC1*	AR
3-Methylcrotonyl-CoA Carboxylase 2 Defi	*MCCC2*	AR
3-Methylglutaconic Aciduria Type 1	*AUH*	AR
3-Methylglutaconic Aciduria Type 2	*TAFAZZIN*	XLR
3-Methylglutaconic Aciduria Type 3	*OPA3*	AR
3-Methylglutaconic Aciduria Type 5	*DNAJC19*	AR
3-Methylglutaconic Aciduria Type 7B	*CLPB*	AR
3-Methylglutaconic Aciduria with Deafne	*SERAC1*	AR
3-Hydroxy-3-Methylglutaryl-CoA Lyase De	*HMGCL*	AR
3-Hydroxy-3-Methylglutaryl-CoA Synthase	*HMGCS2*	AR
Beta-Ketothiolase Deficiency	*ACAT1*	AR
Malonyl-CoA Decarboxylase Deficiency	*MLYCD*	AR
Propionic Acidemia	*PCCA*、*PCCB*	AR

Succinic Semialdehyde Dehydrogenase Def	*ALDH5A1*	AR
Methylmalonic Aciduria, mut (0) Type	*MMUT*	AR
Methylmalonyl-Coa Epimerase Deficiency	*MCEE*	AR
Methylmalonic Acidemia, CblA Type	*MMAA*	AR
Methylmalonic Acidemia, CblB Type	*MMAB*	AR
Methylmalonic Acidemia, CblC Type	*MMACHC*	AR
Methylmalonic Aciduria and Homocystinur	*MMADHC*	AR
Methylmalonic Aciduria and Homocystinur	*PRDX1*	AR
Methylmalonic Aciduria and Homocystinur	*ABCD4*	AR
Methylmalonic Acidemia with Homocystinu	*HCFC1*	XLR
Methylmalonic Aciduria and Homocystinur	*LMBRD1*	AR
Combined Malonic and Methylmalonic Acid	*ACSF3*	AR
Methylmalonic Aciduria due to Transcoba	*CD320*	AR
Mitochondrial DNA Depletion Syndrome 5	*SUCLA2*	AR
Mitochondrial DNA Depletion Syndrome 9	*SUCLG1*	AR
Multiple Carboxylase Deficiency	*HLCS*	AR
Biotinidase Deficiency	*BTD*	AR
Glutaric Acidemia I	*GCDH*	AR
2-Methyl-3-Hydroxybutyryl-CoA Dehydroge	*HSD17B10*	XLD
Isobutyryl-CoA Dehydrogenase Deficiency	*ACAD8*	AR
Isovaleric Acidemia	*IVD*	AR
2,4-Dienoyl-CoA Reductase Deficiency	*NADK2*	AR
Short Chain Acyl-CoA Dehydrogenase Defi	*ACADS*	AR
Glutaric Acidemia IIA	*ETFA*	AR
Glutaric Acidemia IIB	*ETFB*	AR
Glutaric Acidemia IIC	*ETFDH*	AR
Acyl-CoA Dehydrogenase Deficiency, Very	*ACADVL*	AR
Carnitine-Acylcarnitine Translocase Def	*SLC25A20*	AR
Carnitine Palmitoyl Transferase I Defic	*CPT1A*	AR
Carnitine Palmitoyl Transferase II Defi	*CPT2*	AR
Trifunctional Protein Deficiency	*HADHA*、*HADHB*	AR
Primary Carnitine Deficiency	*SLC22A5*	AR
Long-Chain 3-Hydroxyacyl-Coa Dehydrogen	*HADHA*	AR
Medium-Chain Acyl-Coenzyme A Dehydrogen	*ACADM*	AR
Medium Chain 3-Ketoacyl-CoA Thiolase De	*ACAA1*、*ACAA2*	AR
3-Hydroxyacyl-CoA Dehydrogenase Deficie	*HADH*	AR
Ethylmalonic Encephalopathy	*ETHE1*	AR
Krabbe Disease	*GALC*	AR
Fabry Disease	*GLA*	XL
Niemann-Pick Disease A/B	*SMPD1*	AR
Niemann-Pick Disease Type C1	*NPC1*	AR
Niemann-Pick Disease Type C2	*NPC2*	AR
Mucopolysaccharidosis Type I	*IDUA*	AR
Mucopolysaccharidosis II	*IDS*	XLR
Mucopolysaccharidosis Type IIIA	*SGSH*	AR
Mucopolysaccharidosis Type IIIB	*NAGLU*	AR
Mucopolysaccharidosis Type IIIC	*HGSNAT*	AR
Mucopolysaccharidosis Type IVA	*GALNS*	AR
Mucopolysaccharidosis Type IVB	*GLB1*	AR
Mucopolysaccharidosis Type VI	*ARSB*	AR
Mucopolysaccharidosis Type VII	*GUSB*	AR

Mucopolysaccharidosis Type IX	*HYAL1*	AR
Metachromatic Leukodystrophy	*ARSA*	AR
GM1-gangliosidosis, Type I	*GLB1*	AR
GM1-gangliosidosis, Type II	*GLB1*	AR
GM1-gangliosidosis, Type III	*GLB1*	AR
Tay-Sachs Disease	*HEXA*	AR
Sandhoff Disease	*HEXB*	AR
GM2-gangliosidosis, AB variant	*GM2A*	AR
Galactokinase Deficiency	*GALK1*	AR
Galactosemia	*GALT*	AR
Epimerase Deficiency Galactosemia	*GALE*	AR
Glycogen Storage Disease Type Ia	*G6PC*	AR
Glycogen Storage Disease Type Ib/Ic	*SLC37A4*	AR
Glycogen Storage Disease II	*GAA*	AR
Glycogen Storage Disease Type III	*AGL*	AR
Glycogen Storage Disease Type IV	*GBE1*	AR
Glycogen Storage Disease Type V	*PYGM*	AR
Glycogen Storage Disease Type VI	*PYGL*	AR
Glycogen Storage Disease Type IXa1	*PHKA2*	XLR
Glycogen Storage Disease Type IXb	*PHKB*	AR
Glycogen Storage Disease Type IXc	*PHKG2*	AR
Glycogen Storage Disease Type IXd	*PHKA1*	XLR
Glycogen Storage Disease Type XIV	*PGM1*	AR
Hereditary Fructose Intolerance	*ALDOB*	AR
Peroxisome Biogenesis Disorder 1A (Zell	*PEX1*	AR
Peroxisome Biogenesis Disorder 1B	*PEX1*	AR
Peroxisome Biogenesis Disorder 3A (Zell	*PEX12*	AR
Peroxisome Biogenesis Disorder 3B	*PEX12*	AR
Peroxisome Biogenesis Disorder 4A	*PEX6*	AR
Peroxisome Biogenesis Disorder 4B	*PEX6*	AD, AR
Peroxisome Biogenesis Disorder 5A	*PEX2*	AR
Peroxisome Biogenesis Disorder 5B	*PEX2*	AR
Peroxisome Biogenesis Disorder 6A (Zell	*PEX10*	AR
Peroxisome Biogenesis Disorder 6B	*PEX10*	AR
Peroxisome Biogenesis Disorder 7A	*PEX26*	AR
Peroxisome Biogenesis Disorder 7B	*PEX26*	AR
Heimler Syndrome 1	*PEX1*	AR
Heimler Syndrome 2	*PEX6*	AR
Primary Hyperoxaluria Type I	*AGXT*	AR
Primary Hyperoxaluria Type II	*GRHPR*	AR
Primary Hyperoxaluria Type III	*HOGA1*	AR
Glutaric Aciduria III	*SUGCT*	AR
Sitosterolemia 1	*ABCG8*	AR
Sitosterolemia 2	*ABCG5*	AR
Hypercholesterolemia, Familial,1	*LDLR*	AD, AR
Cerebrotendinous Xanthomatosis	*CYP27A1*	AR
Menkes Disease	*ATP7A*	XLR
X-Linked Hypophosphatemia	*PHEX*	XLD
Wilson Disease	*ATP7B*	AR
Progressive Familial Intrahepatic Chole	*ATP8B1*	AR
Progressive Familial Intrahepatic Chole	*ABCB11*	AR

Progressive Familial Intrahepatic Chole	*ABCB4*	AR
Glucose-6-Phosphate Dehydrogenase Defic	*G6PD*	XLD
Congenital Bile Acid Synthesis Defect 1	*HSD3B7*	AR
Hypophosphatasia	*ALPL*	AR
Dihydrolipoamide Dehydrogenase Deficien	*DLD*	AR
Cerebral Creatine Deficiency Syndrome 1	*SLC6A8*	XLR
Cerebral Creatine Deficiency Syndrome 2	*GAMT*	AR
Alagille Syndrome 1	*JAG1*	AD
Alagille Syndrome 2	*NOTCH2*	AD
Crigler-Najjar Syndrome type 1	*UGT1A1*	AR
Crigler-Najjar Syndrome type 2	*UGT1A1*	AR
Pendred Syndrome	*SLC26A4*	AR
Usher Syndrome Type IB	*MYO7A*	AR
Usher Syndrome Type IC	*USH1C*	AR
Usher Syndrome Type IF	*PCDH15*	AR
Usher Syndrome Type IIA	*USH2A*	AR
Usher Syndrome Type IIC	*ADGRV1*	AR
Maternally Inherited Hearing Impairment	*MT-RNR1*	Mi
Autosomal Recessive Deafness 1A	*GJB2*	AR
Autosomal Recessive Deafness 3	*MYO15A*	AR
Autosomal Recessive Deafness 4 with En	*SLC26A4*	AR
Autosomal Recessive Deafness 7	*TMC1*	AR
Autosomal Recessive Deafness 8	*TMPRSS3*	AR
Autosomal Recessive Deafness 9	*OTOF*	AR
Autosomal Recessive Deafness 12	*CDH23*	AR
Autosomal Recessive Deafness 21	*TECTA*	AR
Autosomal Recessive Deafness 22	*OTOA*	AR
Autosomal Recessive Deafness 23	*PCDH15*	AR
Autosomal Recessive Deafness 36	*ESPN*	AR
Autosomal Recessive Deafness 42	*ILDR1*	AR
Autosomal Recessive Deafness 49	*MARVELD2*	AR
Autosomal Recessive Deafness 63	*LRTOMT*	AR
Autosomal Dominant Deafness 2B	*GJB3*	AD
Autosomal Dominant Deafness 5	*GSDME*	AD
Waardenburg Syndrome	*MITF*、*PAX3*、*SOX10*	AD
Wiskott-Aldrich Syndrome 1	*WAS*	XLR
X-Linked Lymphoproliferative Syndrome 1	*SH2D1A*	XLR
X-Linked Lymphoproliferative Syndrome 2	*XIAP*	XLR
X-Linked Agammaglobulinemia 1	*BTK*	XLR
X-linked Chronic Granulomatous Disease	*CYBB*	XLR
X-Linked Severe Combined Immunodeficien	*IL2RG*	XLR
Severe Combined Immunodeficiency, Autos	*RAG1*、*RAG2*	AR
Severe Combined Immunodeficiency Due to	*ADA*	AR
Severe Combined Immunodeficiency, T cel	*IL7R*	AR
Autosomal Recessive T Cell-Negative, B	*JAK3*	AR
Familial Mediterranean Fever	*MEFV*	AR
Immunodeficiency with Hyper-IgM, type 1	*CD40LG*	XLR
Neutropenia, Severe Congenital 1, Autos	*ELANE*	AD
Congenital Adrenal Hyperplasia due to 1	*CYP11B1*	AR
Congenital Adrenal Hyperplasia due to 1	*CYP17A1*	AR

Adrenal Hyperplasia, Congenital, due to	*HSD3B2*	AR
Lipoid Congenital Adrenal Hyperplasia	*STAR*	AR
Hypogonadotropic Hypogonadism 1 with or	*ANOS1*	XLR
Hypogonadotropic Hypogonadism 2 with or	*FGFR1*	AD
Kallmann Syndrome 3	*PROKR2*	AD
Hypogonadotropic Hypogonadism 5 with or	*CHD7*	AD
X-Linked Adrenal Hypoplasia Congenita	*NR0B1*	XLR
Thyroid Dyshormonogenesis 2A	*TPO*	AR
Thyroid Dyshormonogenesis 3	*TG*	AR
Thyroid Dyshormonogenesis 5	*DUOXA2*	AR
Thyroid Dyshormonogenesis 6	*DUOX2*	AR
Combined Pituitary Hormone Deficiency 2	*PROP1*	AR
Hypothyroidism Congenital Nongoitrous 1	*TSHR*	AR
Hypothyroidism Congenital Nongoitrous 2	*PAX8*	AD
Diabetes, Permanent Neonatal 2, with or	*KCNJ11*	AD
Diabetes Mellitus, Permanent Neonatal 3	*ABCC8*	AD, AR
Familial Hyperinsulinemic Hypoglycemia	*ABCC8*	AD, AR
Familial Hyperinsulinemic Hypoglycemia	*KCNJ11*	AR
Familial Hyperinsulinemic Hypoglycemia	*HADH*	AR
Familial Hyperinsulinemic Hypoglycemia	*INSR*	AD
Pyridoxine-Dependent Epilepsy	*ALDH7A1*	AR
Autosomal Dominant Spastic Paraplegia 3	*REEP1*	AD
Autosomal Dominant Spastic Paraplegia 3	*ATL1*	AD
Autosomal Dominant Spastic Paraplegia 4	*SPAST*	AD
Autosomal Recessive Spastic Paraplegia	*SPG11*	AR
Autosomal Dominate Myotonia Congenita	*CLCN1*	AD, AR
Duchenne Muscular Dystrophy	*DMD*	XLR
Spinal Muscular Atrophy, Type I	*SMN1*	AR
Tyrosine Hydroxylase Deficiency	*TH*	AR
Dopa-Responsive Dystonia due to Sepiapt	*SPR*	AR
Glucose Transporter Type 1 Deficiency S	*SLC2A1*	AD, AR
Early Infantile Epileptic Encephalopath	*SCN1A*	AD
Early Infantile Epileptic Encephalopath	*PCDH19*	XL
Brown-Vialetto-Van Laere Syndrome 1	*SLC52A3*	AR
Brown-Vialetto-Van Laere Syndrome 2	*SLC52A2*	AR
Diamond-Blackfan Anemia 10	*RPS26*	AD
Diamond-Blackfan Anemia 1	*RPS19*	AD
Diamond-Blackfan Anemia 7	*RPL11*	AD
Alpha-Thalassemia	*HBA1*、*HBA2*	AR
Beta-Thalassemia	*HBB*	AR
Fanconi Anemia, Complementation Group A	*FANCA*	AR
Familial Hemophagocytic Lymphohistiocyt	*PRF1*	AR
Familial Hemophagocytic Lymphohistiocyt	*UNC13D*	AR
Transcobalamin II Deficiency	*TCN2*	AR
Gitelman Syndrome	*SLC12A3*	AR
Leber Hereditary Optic Neuropathy	*MT-ND4*	Mi
Alport Syndrome 1, X-linked	*COL4A5*	XLD
Alport Syndrome, COL4A3-related	*COL4A3*	AD, AR
Alport Syndrome, COL4A4-related	*COL4A4*	AR

Tuberous Sclerosis 1	*TSC1*	AD
Tuberous Sclerosis-2	*TSC2*	AD
Cystic Fibrosis	*CFTR*	AR
Retinoblastoma	*RB1*	AD
Primary Coenzyme Q10 Deficiency 7	*COQ4*	AR