Condition group	Gene	Condition name (OMIM)
17-alpha-hydroxylase/17,20-lyase deficiency	CYP17A1	17-alpha-hydroxylase/17,20-lyase deficiency
Abetalipoproteinemia	MTTP	Abetalipoproteinemia
Achalasia-addisonianism-alacrimia syndrome	AAAS	Achalasia-addisonianism-alacrimia syndrome
Acrodermatitis enteropathica	SLC39A4	Acrodermatitis enteropathica
Adenine phosphoribosyltransferase deficiency	APRT	Adenine phosphoribosyltransferase deficiency
Adrenocorticotropic hormone deficiency	TBX19	Adrenocorticotropic hormone deficiency
Adrenoleukodystrophy	ABCD1	Adrenoleukodystrophy
Afibrinogenaemia	FGA	Afibrinogenemia, congenital
Afibrinogenaemia	FGB	Afibrinogenemia, congenital
Afibrinogenaemia	FGG	Afibrinogenemia, congenital
Agammaglobulinemia	BTK	X-linked agammaglobulinemia
Agammaglobulinemia	IGHM	Agammaglobulinemia 1
Agammaglobulinemia	IGLL1	Agammaglobulinemia 2
Agammaglobulinemia	CD79A	Agammaglobulinemia 3
Agammaglobulinemia	CD79B	Agammaglobulinemia 6
Agammaglobulinemia	BLNK	Agammaglobulinemia 4
Agammaglobulinemia	PIK3R1	Agammaglobulinemia 7
Agammaglobulinemia	TCF3	Agammaglobulinemia 8
Agammaglobulinemia	TCF3	Agammaglobulinemia 8
Agammaglobulinemia	SLC39A7	SLC39A7 associated agammaglobulinemia
Allan-Herndon-Dudley syndrome	SLC16A2	Allan-Herndon-Dudley syndrome
Apparent mineralocorticoid excess	HSD11B2	Apparent mineralocorticoid excess
Argininemia	ARG1	Argininemia
Argininosuccinic aciduria	ASL	Argininosuccinic aciduria
Aromatic amino acid decarboxylase deficiency	DDC	Aromatic amino acid decarboxylase deficiency
Ataxia pancytopenia syndrome	SAMD9L	Ataxia pancytopenia syndrome
Ataxia with vitamin E deficiency	TTPA	Ataxia with vitamin E deficiency
Atransferrinaemia	TF	Atransferrinaemia
Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia	AIRE	Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia
Autoinflammation, panniculitis, and dermatosis syndrome	OTULIN	Autoinflammation, panniculitis, and dermatosis syndrome
Bamforth-Lazarus syndrome	FOXE1	Bamforth-Lazarus syndrome
Bare lymphocyte syndrome	CIITA	Bare lymphocyte syndrome, type II, complementation group A
Bare lymphocyte syndrome	RFXANK	Bare lymphocyte syndrome, type II, complementation group B
Bare lymphocyte syndrome	RFX5	Bare lymphocyte syndrome, type II, complementation group C
Bare lymphocyte syndrome	RFXAP	Bare lymphocyte syndrome, type II, complementation group D
Barth Syndrome	TAFAZZIN	Barth Syndrome
Bartter syndrome	SLC12A1	Bartter syndrome, type 1
Bartter syndrome	KCNJ1	Bartter syndrome, type 2
Bartter syndrome	MAGED2	Bartter syndrome, type 5, antenatal, transient
Bernard-Soulier syndrome	GP1BA	Bernard-Soulier syndrome, type A1 (recessive)
Bernard-Soulier syndrome	GP1BB	Bernard-Soulier syndrome, type B
Bernard-Soulier syndrome	GP9	Bernard-Soulier syndrome, type C
Beta Thalassaemia	HBB	Beta Thalassaemia
Bile acid conjugation defect	BAAT	Bile acid conjugation defect 1
Biotinidase deficiency	BTD	Biotinidase deficiency
Bone marrow failure syndrome	DNAJC21	Bone marrow failure syndrome 3
Branched-chain ketoacid dehydrogenase kinase deficiency	BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency
Brown-Vialetto-Van Laere syndrome	SLC52A3	Brown-Vialetto-Van Laere syndrome 1
Brown-Vialetto-Van Laere syndrome	SLC52A2	Brown-Vialetto-Van Laere syndrome 2

Carbamoyl phosphate synthetase I deficiency	CPS1	Carbamoyl phosphate synthetase I deficiency
Cardiac arrhythmia syndrome, with or without skeletal muscle weakness	TRDN	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness
Carnitine palmitoyltransferase I deficiency	CPT1A	Carnitine palmitoyltransferase I deficiency
Carnitine palmitoyltransferase II deficiency infantile	CPT2	Carnitine palmitoyltransferase II deficiency infantile
Carnitine-acylcarnitine translocase deficiency	SLC25A20	Carnitine-acylcarnitine translocase deficiency
Catecholaminergic polymorphic ventricular tachycardia	TECRL	Ventricular tachycardia, catecholaminergic polymorphic, 3
Catecholaminergic polymorphic ventricular tachycardia	CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 2
Cerebral creatine deficiency syndrome	GAMT	Cerebral creatine deficiency syndrome 2
Cerebral creatine deficiency syndrome	GATM	Cerebral creatine deficiency syndrome 3
Cerebrotendinous xanthomatosis	CYP27A1	Cerebrotendinous xanthomatosis
Chediak-Higashi Syndrome	LYST	Chediak-Higashi Syndrome
Chronic granulomatous disorder	CYBB	Chronic granulomatous disease x-linked
Chronic granulomatous disorder	CYBA	Chronic granulomatous disease 4
Chronic granulomatous disorder	NCF2	Chronic granulomatous disease 3
Chronic granulomatous disorder	NCF4	Chronic granulomatous disease 2
Chronic granulomatous disorder	CYBC1	Chronic granulomatous disease 5
Chylomicron retention disease	SAR1B	Chylomicron retention disease
Citrullinemia	ASS1	Citrullinemia
Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	MTHFD1	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia
Combined pituitary hormone deficiency	PROP1	Combined Pituitary hormone deficiency 2
Combined pituitary hormone deficiency	POU1F1	Pituitary hormone deficiency, combined or isolated, 1
Combined pituitary hormone deficiency	POU1F1	Pituitary hormone deficiency, combined or isolated, 1
Combined pituitary hormone deficiency	HESX1	Pituitary hormone deficiency, combined, 5
Combined pituitary hormone deficiency	LHX3	Pituitary hormone deficiency, combined, 3
Congenital adrenal hyperplasia	CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency
Congenital adrenal hyperplasia	CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
Congenital adrenal hyperplasia	HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency
Congenital adrenal hypoplasia	NR0B1	Adrenal hypoplasia, congenital
Congenital adrenal insufficiency with 46XY sex reversal	CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete
Congenital bile acid synthesis defect	HSD3B7	Bile acid synthesis defect, congenital, 1
Congenital bile acid synthesis defect	AKR1D1	Bile acid synthesis defect, congenital, 2
Congenital bile acid synthesis defect	CYP7B1	Bile acid synthesis defect, congenital, 3
Congenital bile acid synthesis defect	AMACR	Bile acid synthesis defect, congenital, 4
Congenital diarrhoea	DGAT1	Diarrhea 7, protein-losing enteropathy type
Congenital Diarrhoea	SLC26A3	Diarrhea 1, secretory chloride, congenital
Congenital Diarrhoea	SLC9A3	Diarrhea 8, secretory sodium, congenital
Congenital Diarrhoea	EPCAM	Diarrhea 5, with tufting enteropathy, congenital
Congenital Diarrhoea	SPINT2	Diarrhea 3, secretory sodium, congenital, syndromic
Congenital diarrhoea	NEUROG3	Diarrhea 4, malabsorptive, congenital
Congenital disorder of glycosylation	MPI	Congenital disorder of glycosylation, type Ib
Congenital disorder of glycosylation	PGM1	Congenital disorder of glycosylation, type It
Congenital erythropoietic porphyria	UROS	Congenital erythropoietic porphyria
Congenital hyperinsulinism	HADH	Familial hyperinsulinemic hypoglycemia-4
Congenital hyperinsulinism	HK1	HK1 associated hyperinsulinism
Congenital hyperinsulinism	PMM2	Polycystic kidney disease with hyperinsulinemic hypoglycemia
Congenital hyperinsulinism	GCK	Familial hyperinsulinemic hypoglycemia-3
Congenital hyperinsulinism	ABCC8	Hyperinsulinemic hypoglycemia, familial, 1
Congenital hyperinsulinism	KCNJ11	Familial hyperinsulinemic hypoglycemia-2
Congenital hyperinsulinism	KCNJ11	Familial hyperinsulinemic hypoglycemia-2
Congenital hypoaldosteronism	CYP11B2	Hypoaldosteronism, congenital, due to CMO I deficiency

Congenital hypothyroidism	DUOX2	Thyroid dyshormonogenesis 6
Congenital hypothyroidism	DUOXA2	Thyroid dyshormonogenesis 5
Congenital hypothyroidism	TPO	Thyroid dyshormonogenesis 2A
Congenital hypothyroidism	TG	Thyroid dyshormonogenesis 3
Congenital hypothyroidism	SLC5A5	Thyroid dyshormonigenesis 1
Congenital hypothyroidism	SLC26A7	Thyroid dyshormonigenesis (no phenotype on OMIM)
Congenital hypothyroidism	TSHR	Hypothyroidism, congenital, nongoitrous, 1
Congenital hypothyroidism	TRHR	Hypothyroidism, congenital, nongoitrous, 7
Congenital hypothyroidism	PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia
Congenital hypothyroidism	IYD	Thyroid dyshormonogenesis 4
Congenital hypothyroidism	IGSF1	Hypothyroidism, central, and testicular enlargement
Congenital hypothyroidism	TBL1X	Hypothyroidism, congenital, nongoitrous, 8
Congenital hypothyroidism	IRS4	Hypothyroidism, congenital, nongoitrous, 9
Congenital hypothyroidism	TSHB	Hypothyroidism, congenital, nongoitrous 4
Congenital lipodystrophy	AGPAT2	Congenital generalized lipodystrophy type 1
Congenital lipodystrophy	BSCL2	Lipodystrophy, congenital generalized, type 2
Congenital lipodystrophy	CAV1	Lipodystrophy, congenital generalized, type 3
Congenital lipodystrophy	CAV1	Lipodystrophy, familial partial, type 7
Congenital lipodystrophy	CAVIN1	Lipodystrophy, congenital generalized, type 4
Congenital Myasthenic Syndrome	CHRNE	Myasthenic syndrome, congenital, 4
Congenital Myasthenic Syndrome	CHRNE	Myasthenic syndrome, congenital, 4
Congenital Myasthenic Syndrome	AGRN	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects
Congenital Myasthenic Syndrome	ALG14	Myasthenic syndrome, congenital, 15, without tubular aggregates
Congenital Myasthenic Syndrome	SYT2	Congenital myasthenic syndrome 7
Congenital Myasthenic Syndrome	SYT2	Congenital myasthenic syndrome 7
Congenital Myasthenic Syndrome	CHAT	Congenital myasthenic syndrome-6
Congenital Myasthenic Syndrome	CHRNA1	Congenital myasthenic syndrome-1
Congenital Myasthenic Syndrome	CHRNA1	Congenital myasthenic syndrome-1
Congenital Myasthenic Syndrome	CHRNB1	Congenital myasthenic syndrome-2
Congenital Myasthenic Syndrome	CHRNB1	Congenital myasthenic syndrome-2
Congenital Myasthenic Syndrome	CHRND	Congenital myasthenic syndrome-3
Congenital Myasthenic Syndrome	CHRND	Congenital myasthenic syndrome-3
Congenital Myasthenic Syndrome	COL13A1	Congenital myasthenic syndrome-19
Congenital Myasthenic Syndrome	COLQ	Congenital myasthenic syndrome-5
Congenital Myasthenic Syndrome	DOK7	Congenital myasthenic syndrome-10
Congenital Myasthenic Syndrome	DPAGT1	Congenital myasthenic syndrome-13
Congenital Myasthenic Syndrome	ALG2	Congenital myasthenic syndrome-14
Congenital Myasthenic Syndrome	GFPT1	Congenital myasthenic syndrome-12
Congenital Myasthenic Syndrome	MUSK	Congenital myasthenic syndrome-9
Congenital Myasthenic Syndrome	MYO9A	Congenital myasthenic syndrome-24
Congenital Myasthenic Syndrome	PREPL	Congenital myasthenic syndrome-22
Congenital Myasthenic Syndrome	RAPSN	Congenital myasthenic syndrome-11
Congenital Myasthenic Syndrome	SLC18A3	Congenital myasthenic syndrome-21
Congenital Myasthenic Syndrome	SLC25A1	Congenital myasthenic syndrome-23
Congenital Myasthenic Syndrome	SLC5A7	Congenital myasthenic syndrome-20
Congenital Myasthenic Syndrome	VAMP1	Congenital myasthenic syndrome-25
Congenital Myasthenic Syndrome	SCN4A	Congenital myasthenic syndrome-16
Congenital prothrombin deficiency	F2	Congenital Prothrombin deficiency
Congenital sucrase-isomaltase deficiency	SI	Congenital sucrase-isomaltase deficiency
Crigler-Najjar syndrome Type I	UGT1A1	Crigler-Najjar syndrome Type I
Cryopyrin associated periodic fever syndrome	NLRP3	Cryopyrin associated periodic fever syndrome
Cystic fibrosis	CFTR	Cystic fibrosis

Cystinosis	CTNS	Cystinosis, nephropathic
cytochrome P450 oxidoreductase deficiency	POR	cytochrome P450 oxidoreductase deficiency
Diabetes Insipidus	AVPR2	Diabetes insipidus, nephrogenic, 1
Diabetes Insipidus	AQP2	Recessive diabetes insipidus, nephrogenic, 2
Diabetes Insipidus	AQP2	Dominant diabetes insipidus, nephrogenic, 2
Diabetes Insipidus	AVP	Diabetes insipidus, neurohypophyseal
Diamond Blackfan Anaemia	RPS19	Diamond blackfan anaemia 1
Diamond Blackfan Anaemia	RPL5	Diamond blackfan anaemia 6
Diamond Blackfan Anaemia	RPL11	Diamond-Blackfan anemia 7
Diamond Blackfan Anaemia	RPS26	Diamond-Blackfan anemia 10
Diamond Blackfan Anaemia	RPL35A	Diamond-Blackfan anemia 5
Diamond Blackfan Anaemia	RPS10	Diamond-Blackfan anemia 9
Diamond Blackfan Anaemia	RPS17	Diamond-Blackfan anemia 4
Diamond Blackfan Anaemia	RPS24	Diamond-blackfan anemia 3
Diamond Blackfan Anaemia	RPL15	Diamond-Blackfan anemia 12
Diamond Blackfan Anaemia	RPL31	RPL31 associated Diamond-Blackfan anemia
Diamond Blackfan Anaemia	RPS7	Diamond-Blackfan anemia 8
Distal renal tubular acidosis	ATP6V0A4	Distal renal tubular acidosis 3, with or without sensorineural hearing loss
Distal renal tubular acidosis	ATP6V1B1	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss
Distal renal tubular acidosis	SLC4A1	Distal renal tubular acidosis 1
Distal renal tubular acidosis	SLC4A1	Distal renal tubular acidosis 4 with hemolytic anemia
Dopa responsive dystonia	TH	Dopa-responsive dystonia due to tyrosine hydroxylase deficiency
Early infantile epileptic encephalopathy	SCN1A	Early infantile epileptic encephalopathy-6
Early infantile epileptic encephalopathy	SCN2A	Early infantile epileptic encephalopathy-11
Early infantile epileptic encephalopathy	SCN8A	Early infantile epileptic encephalopathy-13
Early infantile epileptic encephalopathy	CAD	Early infantile epileptic encephalopathy-50
Early infantile epileptic encephalopathy	KCNQ2	Early infantile epileptic encephalopathy-7
Early infantile epileptic encephalopathy	KCNT1	Early infantile epileptic encephalopathy-14
Early onset inflammatory bowel disease	IL10	Interleukin-10 deficiency
Early onset inflammatory bowel disease	IL10RB	Inflammatory bowel disease 25
Early onset inflammatory bowel disease	IL10RA	Inflammatory bowel disease 28
Early onset osteoporosis	PLS3	Bone mineral density QTL18, osteoporosis
Early onset osteoporosis	LRP5	Osteoporosis-pseudoglioma syndrome
Ectodermal dysplasia and immunodeficiency 2	NFKBIA	Ectodermal dysplasia and immunodeficiency 2
Erythropoietic protoporphyria	FECH	Protoporphyria, erythropoietic, 1
Erythropoietic protoporphyria	ALAS2	Protoporphyria, erythropoietic, X-linked
Factor V deficiency	F5	Factor V deficiency
Factor VII deficiency	F7	Factor VII deficiency
Factor X deficiency	F10	Factor X deficiency
Factor XIII Deficiency	F13A1	Factor XIIIA deficiency
Factor XIII Deficiency	F13B	Factor XIIIB deficiency
Familial Chylomicronaemia Syndrome	LPL	Lipoprotein lipase deficiency
Familial Chylomicronaemia Syndrome	APOC2	Hyperlipoproteinemia, type Ib
Familial Chylomicronaemia Syndrome	LMF1	Lipase deficiency, combined
Familial Chylomicronaemia Syndrome	APOA5	apolipoprotein A-V deficiency
Familial Chylomicronaemia Syndrome	GPIHBP1	Hyperlipoproteinemia, type 1D
Familial Hemophagocytic lymphohistiocytosis	PRF1	Hemophagocytic lymphohistiocytosis, familial, 2
Familial Hemophagocytic lymphohistiocytosis	UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3
Familial Hemophagocytic lymphohistiocytosis	STX11	Hemophagocytic lymphohistiocytosis, familial, 4
Familial Hemophagocytic lymphohistiocytosis	STXBP2	Familial hemophagocytic lymphohistiocytosis-5
Familial hyperphosphataemic tumoral calcinosis	GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 1
Familial hyperphosphataemic tumoral calcinosis	FGF23	Tumoral calcinosis, hyperphosphatemic, familial, 2

Familial isolated hypoparathyroidism	GCM2	familial isolated hypoparathyroidism 2
Familial Mediterranean fever	MEFV	Familial Mediterranean fever, AR
Familial Mediterranean fever	MEFV	Familial Mediterranean fever, AD
Familial thrombotic thrombocytopenic purpura	ADAMTS13	Familial thrombotic thrombocytopenic purpura
Fructose-1,6-bisphosphatase deficiency	FBP1	Fructose-1,6-bisphosphatase deficiency
Galactokinase deficiency with cataracts	GALK1	Galactokinase deficiency with cataracts
Galactosaemia	GALT	Galactosaemia
Gastrointestinal defects and immunodeficiency syndrome	TTC7A	Gastrointestinal defects and immunodeficiency syndrome
Generalised arterial calcification of infancy	ENPP1	Arterial calcification, generalized, of infancy, 1
Generalised arterial calcification of infancy	ABCC6	Generalized arterial calcification of infancy 2
Glanzmann thrombasthenia	ITGA2B	Glanzmann thrombasthenia 1
Glanzmann thrombasthenia	ITGB3	Glanzmann thrombasthenia 2
Glucocorticoid deficiency	MC2R	Glucocorticoid deficiency, due to ACTH unresponsiveness
Glucocorticoid deficiency	MRAP	Glucocorticoid deficiency 2
Glucocorticoid deficiency	NNT	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency
Glucose/galactose malabsorption	SLC5A1	Glucose/galactose malabsorption
GLUT1 deficiency syndrome	SLC2A1	GLUT1 deficiency syndrome-1
GLUT1 deficiency syndrome	SLC2A1	GLUT1 deficiency syndrome-1
Glutaric acidaemia type I	GCDH	Glutaric aciduria, type I
Glycogen storage disease II (Pompe Disease)	GAA	Glycogen storage disease II
Glycogen storage disease type I	G6PC1	Glycogen storage disease Ia
Glycogen storage disease type I	SLC37A4	Glycogen storage disease Ib and 1c
Glycogen storage disease type III	AGL	Glycogen storage disease III
Griscelli Syndrome	RAB27A	Griscelli syndrome, type 2
Growth hormone receptor deficiency	GHR	Growth hormone receptor deficiency
Growth hormone-secreting pituitary adenoma-2	GPR101	Growth hormone-secreting pituitary adenoma-2
Haemophilia A	F8	Haemophilia A
Haemophilia B	F9	Haemophilia B
Hepatic venoocclusive disease with immunodeficiency	SP110	Hepatic venoocclusive disease with immunodeficiency
Hepatoerythropoietic porphyria	UROD	Porphyria, hepatoerythropoietic
Hereditary angioedema	SERPING1	Hereditary angioedema
Hereditary angioedema	SERPING1	Hereditary angioedema
Hereditary folate malabsorption	SLC46A1	Hereditary folate malabsorption
Hereditary fructose intolerance	ALDOB	Hereditary fructose intolerance
Hermansky-Pudlak syndrome	AP3B1	Hermansky-Pudlak syndrome 2
Hermansky-Pudlak syndrome	AP3D1	Hermansky-Pudlak syndrome 10
HMG-CoA lyase deficiency	HMGCL	HMG-CoA lyase deficiency
HMG-CoA synthase-2 deficiency	HMGCS2	HMG-CoA synthase-2 deficiency
Holocarboxylase synthetase deficiency	HLCS	Holocarboxylase synthetase deficiency
Homocystinuria	CBS	Homocystinuria, B6-responsive and nonresponsive types
Homocystinuria	MTHFR	Homocystinuria due to MTHFR deficiency
Homocystinuria-megaloblastic anaemia	MTRR	Homocystinuria-megaloblastic anemia, cbl E type
Homocystinuria-megaloblastic anaemia	MTR	Homocystinuria-megaloblastic anemia, cblG complementation type
Homozygous Familial hypercholesterolemia-1	LDLR	Homozygous Familial hypercholesterolemia-1
Hyper-IgD syndrome / mevalonate kinase deficiciency	MVK	Hyper-IgD syndrome / mevalonate kinase deficiciency
Hyper-IgE recurrent infection syndrome, autosomal recessive	DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive
Hyperinsulinism-hyperammonemia syndrome	GLUD1	Hyperinsulinism-hyperammonemia syndrome
Hypermanganesaemia with dystonia	SLC30A10	Hypermanganesemia with dystonia 1
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
Hyperphenyalaninaemia	PTS	Hyperphenylalaninemia due to 6-pyruvoyl-tetrahydropterin synthase deficiency
Hyperphenyalaninaemia	QDPR	Hyperphenylalaninemia due to dihydropteridine reductase deficiency

Hypobetalipoproteinaemia	APOB	Hypobetalipoproteinemia
Hypohidrotic ectodermal dysplasia	EDA	Ectodermal dysplasia 1, hypohidrotic, X-linked
Hypohidrotic ectodermal dysplasia	EDAR	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant
Hypohidrotic ectodermal dysplasia	EDAR	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive
Hypohidrotic ectodermal dysplasia	EDARADD	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant
Hypohidrotic ectodermal dysplasia	EDARADD	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive
Hypophosphataemic rickets	FGF23	Hypophosphatemic rickets, autosomal dominant
Hypophosphataemic rickets	SLC34A3	Hypophosphatemic rickets with hypercalciuria
Hypophosphataemic rickets	DMP1	Hypophosphatemic rickets, AR
Hypophosphatasia	ALPL	Autosomal recessive hypophosphatasia
Hypophosphatasia	ALPL	Autosomal dominant hypophosphatasia
IMAGE syndrome	CDKN1C	IMAGE syndrome
IMAGE-I syndrome	POLE	IMAGE-I syndrome
Imerslund-Grasbeck syndrome	CUBN	Imerslund-Grasbeck syndrome 1
Imerslund-Grasbeck syndrome	AMN	Imerslund-Grasbeck syndrome 2
Immunodeficiency	STAT1	Immunodeficiency 31B
Immunodeficiency	LCK	immunodeficiency 22
Immunodeficiency	FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy
Immunodeficiency	PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency
Immunodeficiency	PRKDC	Immunodeficiency 26, with or without neurologic abnormalities
Immunodeficiency	CD3E	Immunodeficiency 18
Immunodeficiency	CD3G	Immunodeficiency 17, CD3 gamma deficient
Immunodeficiency	CD247	Immunodeficiency 25
Immunodeficiency	LAT	Immunodeficiency 52
Immunodeficiency	CORO1A	immunodeficiency 8
Immunodeficiency	DOCK2	Immunodeficiency 40
Immunodeficiency	ZAP70	Immunodeficiency 48
Immunodeficiency	RAC2	Immunodeficiency 73B
Immunodeficiency	IFNGR1	Immunodeficiency 27A, mycobacteriosis
Immunodeficiency	IFNGR1	Immunodeficiency 27A, mycobacteriosis
Immunodeficiency	IFNGR2	Immunodeficiency 28, mycobacteriosis
Immunodeficiency	FCHO1	Immunodeficiency 76
Immunodeficiency	IKBKB	Immunodeficiency 15B
Immunodeficiency	CTPS1	Immunodeficiency 24
Immunodeficiency	IRAK4	IRAK4 deficiency
Immunodeficiency	MYD88	Immunodeficiency 68
Immunodeficiency	LIG1	LIG1 associated immunodeficiency
Immunodeficiency	MAGT1	X-linked Immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia
Immunodeficiency	RASGRP1	Immunodeficiency 64
Immunodeficiency	IRF8	Immunodeficiency 32B
Immunodeficiency	MCM4	Immunodeficiency 54
Immunodeficiency with hyper-IgM	CD40	Immunodeficiency with hyper-IgM, type 3
Immunodeficiency with hyper-IgM	CD40LG	X-linked immunodeficiency with hyper-IgM type 1
Immunodeficiency with lymphoproliferation and autoimmunity	IL2RA	Immunodeficiency 41 with lymphoproliferation and autoimmunity
Immunodeficiency with lymphoproliferation and autoimmunity	IL2RB	Immunodeficiency 63 with lymphoproliferation and autoimmunity
Immunodeficiency-centromeric instability-facial anomalies syndrome	DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1

Immunodeficiency-centromeric instability-facial anomalies syndrome	ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome 2
Immunodeficiency-centromeric instability-facial anomalies syndrome	CDCA7	Immunodeficiency-centromeric instability-facial anomalies syndrome 3
Immunodeficiency-centromeric instability-facial anomalies syndrome	HELLS	Immunodeficiency-centromeric instability-facial anomalies syndrome 4
Insulin-like growth factor I deficiency	IGF1	Insulin-like growth factor I deficiency
Intestinal hypomagnesaemia	TRPM6	Hypomagnesemia 1, intestinal
Intrinsic factor deficiency	CBLIF	Intrinsic factor deficiency
Isolated growth hormone deficiency	GH1	Isolated growth hormone deficiency type 1A
Isolated growth hormone deficiency	GH1	Isolated growth hormone deficiency type 1B
Isolated growth hormone deficiency	GH1	Growth hormone deficiency, isolated, type II
Isolated growth hormone deficiency	GHRHR	Isolated growth hormone deficiency type 4
Isolated growth hormone deficiency	RNPC3	RNPC3 associated growth hormone deficiency
Isolated methylmalonic acidaemia	MMUT	Methylmalonic acidemia, mut(0) type
Isolated methylmalonic acidaemia	MCEE	Methylmalonyl-CoA epimerase deficiency
Isolated methylmalonic acidaemia	MMAA	Methylmalonic aciduria, vitamin B12-responsive, cblA type
Isolated methylmalonic acidaemia	MMAB	Methylmalonic aciduria, vitamin B12-responsive, cblB type
Isolated methylmalonic acidaemia	MMADHC	Methylmalonic aciduria, cblD type, variant 2
Isovaleric acidemia	IVD	Isovaleric acidemia
Jervell and Lange-Nielsen syndrome	KCNQ1	Jervell and Lange-Nielsen syndrome
Kenny-Caffey syndrome, type 2	FAM111A	Kenny-Caffey syndrome, type 2
LCHAD deficiency	HADHA	LCHAD deficiency
Leptin deficiency	LEP	Leptin deficiency
Leptin receptor deficiency	LEPR	Leptin receptor deficiency
Leukocyte adhesion deficiency	FERMT3	Leukocyte adhesion deficiency, type III
Leukocyte adhesion deficiency	ITGB2	Leukocyte adhesion deficiency, type I
LIG4 Syndrome	LIG4	LIG4 Syndrome
Lipoid adrenal hyperplasia	STAR	Lipoid adrenal hyperplasia
Long QT syndrome	KCNQ1	Long QT Syndrome 1
Long QT syndrome	KCNH2	Long QT Syndrome 2
Long QT syndrome	SCN5A	Long QT Syndrome 3
Lymphoproliferative syndrome	SH2D1A	X-linked lymphoproliferative syndrome 1
Lymphoproliferative syndrome	XIAP	X-linked lymphoproliferative syndrome 2
Lymphoproliferative syndrome	ITK	Lymphoproliferative syndrome 2
Lymphoproliferative syndrome	CD70	Lymphoproliferative syndrome 3
Lysinuric protein intolerance	SLC7A7	Lysinuric protein intolerance
Maple Syrup Urine Disease	DBT	Maple syrup urine disease, type II
Maple Syrup Urine Disease	BCKDHA	Maple syrup urine disease, type Ia
Maple Syrup Urine Disease	BCKDHB	Maple syrup urine disease, type Ib
Medium-chain acyl-CoA dehydrogenase deficiency	ACADM	Medium-chain acyl-CoA dehydrogenase deficiency
Megaloblastic anaemia	SLC19A1	Megaloblastic anemia, folate-responsive
Megaloblastic anaemia	SLC19A2	Thiamine Repsonsive Megaloblastic Anaemia Syndrome
Megaloblastic anaemia	DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency
Metachromatic leukodystrophy	ARSA	Metachromatic leukodystrophy
Methylmalonic aciduria and homocystinuria	MMADHC	Methylmalonic aciduria and homocystinuria, cblD type
Methylmalonic aciduria and homocystinuria	MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
Methylmalonic aciduria and homocystinuria	LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type
MIRAGE syndrome	SAMD9	MIRAGE syndrome
Mucopolysaccharidosis I	IDUA	Mucopolysaccharidosis I
Mucopolysaccharidosis II	IDS	Mucopolysaccharidosis II
Mucopolysaccharidosis IVA	GALNS	Mucopolysaccharidosis IVA
Mucopolysaccharidosis VI	ARSB	Mucopolysaccharidosis VI
Multiple Acyl-CoA Dehydrogenase Deficiency	ETFA	Glutaric acidemia IIA
Multiple Acyl-CoA Dehydrogenase Deficiency	ETFB	Glutaric acidemia IIB

Multiple Acyl-CoA Dehydrogenase Deficiency	ETFDH	Glutaric acidemia IIC
Multiple endocrine neoplasia II	RET	Multiple endocrine neoplasia II
N-acetylglutamate synthase deficiency	NAGS	N-acetylglutamate synthase deficiency
Neonatal diabetes	ABCC8	Diabetes mellitus, permanent neonatal 3, with or without neurologic features
Neonatal diabetes	ABCC8	Diabetes mellitus, permanent neonatal 3, with or without neurologic features
Neonatal diabetes	KCNJ11	Diabetes, permanent neonatal 2, with or without neurologic features
Neonatal diabetes	GCK	Diabetes mellitus, permanent neonatal 1
Neonatal diabetes	INS	Autosomal dominant diabetes mellitus, permanent neonatal 4
Neonatal diabetes	INS	Autosomal recessive diabetes mellitus, permanent neonatal 4
Neonatal diabetes	GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism
Neonatal diabetes	NEUROD1	Maturity-onset diabetes of the young 6
Neonatal hyperparathyroidism	CASR	Neonatal hyperparathyroidism
Neonatal hyperparathyroidism	CASR	Neonatal hyperparathyroidism
Nephrotic syndrome due to primary coenzyme Q10 deficiency	COQ8B	Nephrotic syndrome, type 9
Nephrotic syndrome with or without adrenal insufficency	SGPL1	Nephrotic syndrome, type 14
Neurodegeneration due to cerebral folate transport deficiency	FOLR1	Neurodegeneration due to cerebral folate transport deficiency
Neuronal ceroid lipofuscinosis	TPP1	Ceroid lipofuscinosis, neuronal, 2
Nijmegen breakage syndrome	NBN	Nijmegen breakage syndrome
OAS1 associated polymorphic autoinflammatory immunodeficiency	OAS1	OAS1 associated polymorphic autoinflammatory immunodeficiency
Ornithine transcarbamylase deficiency	OTC	Ornithine transcarbamylase deficiency
Osteogenesis Imperfecta	COL1A1	Osteogenesis Imperfecta
Osteogenesis Imperfecta	COL1A2	Osteogenesis Imperfecta
Osteogenesis Imperfecta	IFITM5	Osteogenesis Imperfecta type V
Osteopetrosis	TCIRG1	Osteopetrosis type 1
Osteopetrosis	CLCN7	Osteopetrosis type 4
Osteopetrosis	CLCN7	Osteopetrosis type 4
Osteopetrosis	TNFRSF11A	Osteopetrosis type 7
Osteopetrosis	SNX10	Osteopetrosis type 8
Osteopetrosis	CA2	Osteopetrosis with renal tubular acidosis
OTOF related deafness	OTOF	Auditory neuropathy, autosomal recessive, 1
Otofaciocervical syndrome 2	PAX1	Otofaciocervical syndrome 2
Pancreatic agenesis	PTF1A	Pancreatic agenesis 2
Phenylketonuria	PAH	Phenylketonuria
Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease	ARPC1B	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease
POMC deficiency	POMC	Obesity, adrenal insufficiency, and red hair due to POMC deficiency
Porphyria variegata	PPOX	Porphyria variegata
Primary Ciliary Dyskinesia	DNAH5	Primary Ciliary Dyskinesia 3
Primary Ciliary Dyskinesia	CCDC39	Primary Ciliary Dyskinesia 14
Primary Ciliary Dyskinesia	DNAH11	Primary Ciliary Dyskinesia 15
Primary Ciliary Dyskinesia	CCDC40	Primary Ciliary Dyskinesia 7
Primary Ciliary Dyskinesia	DNAI1	Primary Ciliary Dyskinesia 1
Primary Ciliary Dyskinesia	SPAG1	Primary Ciliary Dyskinesia 28
Primary Ciliary Dyskinesia	ODAD2	Ciliary dyskinesia, primary, 23
Primary Ciliary Dyskinesia	CCDC103	Ciliary dyskinesia, primary, 17
Primary Ciliary Dyskinesia	ODAD3	Ciliary dyskinesia, primary, 30
Primary Ciliary Dyskinesia	CCNO	primary ciliary dyskinesia 29
Primary Ciliary Dyskinesia	CFAP300	ciliary dyskinesia, primary, 38
Primary Ciliary Dyskinesia	DNAAF3	primary ciliary dyskinesia 2
Primary Ciliary Dyskinesia	DNAI2	primary ciliary dyskinesia 9

Primary Ciliary Dyskinesia	FOXJ1	ciliary dyskinesia, primary, 43
Primary Ciliary Dyskinesia	HYDIN	primary ciliary dyskinesia 5
Primary Ciliary Dyskinesia	MCIDAS	ciliary dyskinesia, primary, 42
Primary Ciliary Dyskinesia	ODAD1	primary ciliary dyskinesia 20
Primary Ciliary Dyskinesia	ODAD4	primary ciliary dyskinesia 35
Primary Ciliary Dyskinesia	RSPH1	primary ciliary dyskinesia 24
Primary Ciliary Dyskinesia	RSPH3	Ciliary dyskinesia, primary, 32
Primary Ciliary Dyskinesia	RSPH4A	primary ciliary dyskinesia 11
Primary Ciliary Dyskinesia	RSPH9	Ciliary dyskinesia, primary, 12
Primary Ciliary Dyskinesia	ZMYND10	primary ciliary dyskinesia 22
Primary Ciliary Dyskinesia	CFAP298	Ciliary dyskinesia, primary, 26
Primary Ciliary Dyskinesia	CCDC65	Ciliary dyskinesia, primary, 27
Primary Ciliary Dyskinesia	DNAAF1	Ciliary dyskinesia, primary, 13
Primary Ciliary Dyskinesia	DNAAF11	Ciliary dyskinesia, primary, 19
Primary Ciliary Dyskinesia	DNAAF2	Ciliary dyskinesia, primary, 10
Primary Ciliary Dyskinesia	DNAAF4	Ciliary dyskinesia, primary, 25
Primary Ciliary Dyskinesia	DNAAF5	Ciliary dyskinesia, primary, 18
Primary Ciliary Dyskinesia	DNAAF6	Ciliary dyskinesia, primary, 36, X-linked
Primary Ciliary Dyskinesia	DNAH9	Ciliary dyskinesia, primary, 40
Primary Ciliary Dyskinesia	DNAL1	Ciliary dyskinesia, primary, 16
Primary Ciliary Dyskinesia	DRC1	Ciliary dyskinesia, primary, 21
Primary Ciliary Dyskinesia	GAS8	Ciliary dyskinesia, primary, 33
Primary Ciliary Dyskinesia	LRRC56	Ciliary dyskinesia, primary, 39
Primary coenzyme Q10 deficiency	COQ4	Coenzyme Q10 deficiency, primary, 7
Primary coenzyme Q10 deficiency	COQ6	Coenzyme Q10 deficiency, primary, 6
Primary coenzyme Q10 deficiency	COQ2	Coenzyme Q10 deficiency, primary, 1
Primary hyperoxaluria type I	AGXT	Hyperoxaluria, primary, type 1
Primary hyperoxaluria type II	GRHPR	Hyperoxaluria, primary, type II
Primary hyperoxaluria type III	HOGA1	Hyperoxaluria, primary, type III
Primary systemic carnitine deficiency	SLC22A5	Primary systemic carnitine deficiency
Properdin deficiency	CFP	Properdin deficiency, X-linked
Propionic acidaemia	PCCA	Propionic acidemia 1
Propionic acidaemia	PCCB	Propionic acidemia 2
Pseudohypoaldosteronism	SCNN1A	Pseudohypoaldosteronism Type 1A
Pseudohypoaldosteronism	SCNN1B	Pseudohypoaldosteronism Type 1A
Pseudohypoaldosteronism	SCNN1G	Pseudohypoaldosteronism Type 1A
Pseudohypoaldosteronism	NR3C2	Pseudohypoaldosteronism type I, autosomal dominant
Pseudohypoaldosteronism	WNK1	Pseudohypoaldosteronism, type IIC
Pseudohypoaldosteronism	KLHL3	Pseudohypoaldosteronism, type IID
Pseudohypoaldosteronism	KLHL3	Pseudohypoaldosteronism, type IID
Pseudohypoaldosteronism	CUL3	Pseudohypoaldosteronism, type IIE
PSTPIP1 associated inflammatory disease	PSTPIP1	PSTPIP1 associated inflammatory disease
Pyridoxamine 5-prime-phosphate oxidase deficiency	PNPO	Pyridoxamine 5-prime-phosphate oxidase deficiency
Pyridoxine dependent epilepsy	ALDH7A1	Pyridoxine-dependent epilepsy
Pyruvate kinase deficiency	PKLR	Pyruvate kinase deficiency
Resistance to thyroid hormone alpha	THRA	Resistance to thyroid hormone alpha (OMIM: Hypothyroidism, congenital, nongoitrous, 6)
Reticular Dysgenesis	AK2	Reticular Dysgenesis
Retinoblastoma	RB1	Retinoblastoma
Rickets due to defect in vitamin D 25-hydroxylation deficiency	CYP2R1	Rickets due to defect in vitamin D 25-hydroxylation deficiency
RPE65 associated Leber congenital amaurosis, early-onset severe retinal dystrophy	RPE65	RPE65 associated Leber congenital amaurosis, early-onset severe retinal dystrophy
SCID	ADA	SCID due to ADA deficiency
SCID	IL2RG	SCID X-Linked

SCID	JAK3	JAK3 associated T cell-negative, B cell-positive, natural killer cell-negative severe combined immunodeficiency
SCID	NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation
SCID	IL7R	Immunodeficiency 104, severe combined
SCID	PTPRC	Immunodeficiency 105, severe combined
SCID	RAG1	Severe combined immunodeficiency, B cell-negative
SCID	RAG2	Severe combined immunodeficiency, B cell-negative
SCID	DCLRE1C	Severe combined immunodeficiency with sensitivity to ionising radiation
SCID	CD3D	Immunodeficiency 19, severe combined
SCOT deficiency	OXCT1	Succinyl-CoA:3-ketoacid CoA transferase (SCOT) deficiency
Shwachman-Diamond syndrome	SBDS	Shwachman-Diamond syndrome 1
Shwachman-Diamond syndrome	EFL1	Shwachman-Diamond syndrome 2
Shwachman-Diamond syndrome	SRP54	SRP54 associated Shwachman-Diamond syndrome
Sickle Cell Disease	HBB	Sickle Cell Disease
SMA	SMN1	Spinal Muscular Atrophy
Specific complement deficiency	CFB	Complement factor B deficiency
Specific complement deficiency	CFH	Complement factor H deficiency
Specific complement deficiency	CFH	Complement factor H deficiency
Specific complement deficiency	CFI	Complement factor I deficiency
Specific complement deficiency	C3	C3 deficiency
Specific complement deficiency	C2	C2 deficiency
Specific complement deficiency	C5	C5 deficiency
Specific complement deficiency	C7	C7 deficiency
Specific complement deficiency	C8A	C8 deficiency, type I
Specific complement deficiency	C8B	C8 deficiency, type II
Specific complement deficiency	C6	C6 deficiency
Specific complement deficiency	C9	C9 deficiency
Specific complement deficiency	CFD	Complement factor D deficiency
Specific granule deficiency	SMARCD2	Specific granule deficiency 2
Specific granule deficiency	CEBPE	Specific granule deficiency 1
STK4 associated T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations	STK4	STK4 associated T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations
Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)
Thiamine metabolism dysfunction syndrome 5	TPK1	Thiamine metabolism dysfunction syndrome 5
Thyroid hormone resistance	THRB	Thyroid hormone resistance
Thyroid hormone resistance	THRB	Thyroid hormone resistance
Transcobalamin II deficiency	TCN2	Transcobalamin II deficiency
Trichohepatoenteric syndrome	SKIC3	Trichohepatoenteric syndrome 1
Trichohepatoenteric syndrome	SKIC2	Trichohepatoenteric syndrome 2
Trifunctional protein deficiency	HADHB	Trifunctional protein deficiency
Tumor necrosis factor receptor associated periodic fever syndrome	TNFRSF1A	Tumor necrosis factor receptor associated periodic fever syndrome
Tyrosinemia, type I	FAH	Tyrosinemia, type I
Tyrosinemia, type II	TAT	Tyrosinemia, type II
Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome	ADA2	Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome
Vitamin B6-dependent epilepsy	PLPBP	Vitamin B6-dependent epilepsy
Vitamin D dependent rickets	CYP27B1	Vitamin D-dependent rickets, type I
Vitamin D resistant rickets	VDR	Rickets, vitamin D-resistant, type IIA
VLCAD deficiency	ACADVL	VLCAD deficiency
Wilms Tumour predisposition syndrome	WT1	Wilms Tumour type 1/ Denys-Drash syndrome
Wilms Tumour predisposition syndrome	REST	Wilms tumour predisposition
Wilms Tumour predisposition syndrome	TRIM28	TRIM28 related Wilms tumor
Wiskott Aldrich syndrome	WAS	Wiskott Aldrich syndrome

Wolcot-Rallison syndrome	EIF2AK3	multiple epiphyseal dysplasia with early onset diabetes mellitus, Wolcot-Rallison syndrome
Xeroderma pigmentosum	DDB2	Xeroderma pigmentosum, group E, DDB-negative subtype
Xeroderma pigmentosum	ERCC2	Xeroderma pigmentosum, group D
Xeroderma pigmentosum	ERCC3	Xeroderma pigmentosum, group B
Xeroderma pigmentosum	ERCC4	Xeroderma pigmentosum, group F
Xeroderma pigmentosum	ERCC5	Xeroderma pigmentosum, group G
Xeroderma pigmentosum	POLH	Xeroderma pigmentosum, variant type
Xeroderma pigmentosum	XPA	Xeroderma pigmentosum, group A
Xeroderma pigmentosum	XPC	Xeroderma pigmentosum, group C
X-linked hypophosphataemic rickets	PHEX	Hypophosphatemic rickets, X-linked dominant
X-linked immunodysregulation, polyendocrinopathy, and enteropathy	FOXP3	X-linked immunodysregulation, polyendocrinopathy, and enteropathy