

Condição	Gene	Nome Técnico da Condição
Abetalipoproteinaemia	MTTP	Abetalipoproteinaemia
Achalasia-addisonianism-alacrimia syndrome	AAAS	Achalasia-addisonianism-alacrimia syndrome
Acrodermatitis enteropathica	SLC39A4	Acrodermatitis enteropathica
Adenine phosphoribosyltransferase deficiency	APRT	Adenine phosphoribosyltransferase deficiency
Adenosine deaminase 2 deficiency	ADA2	Deficiency of ADA2 (DADA2)
Adrenocorticotrophic hormone deficiency	TBX19	Adrenocorticotrophic hormone deficiency
Adrenoleukodystrophy	ABCD1	Adrenoleukodystrophy
Afibrinogenaemia	FGA	FGA related afibrinogenaemia
Afibrinogenaemia	FGB	FGB related afibrinogenaemia
Afibrinogenaemia	FGG	FGG related afibrinogenaemia
Agammaglobulinaemia	BTK	X-linked Agammaglobulinaemia
Agammaglobulinaemia	IGHM	Agammaglobulinaemia 1
Agammaglobulinaemia	IGLL1	Agammaglobulinaemia 2
Agammaglobulinaemia	CD79A	Agammaglobulinaemia 3
Agammaglobulinaemia	CD79B	Agammaglobulinaemia 6
Agammaglobulinaemia	BLNK	Agammaglobulinaemia 4
Agammaglobulinaemia	PIK3R1	Agammaglobulinaemia 7
Agammaglobulinaemia	TCF3	Agammaglobulinaemia 8, autosomal recessive
Agammaglobulinaemia	TCF3	Agammaglobulinaemia 8, autosomal dominant
Agammaglobulinaemia	SLC39A7	SLC39A7 associated Agammaglobulinaemia
Alpha-methylacetoacetic aciduria	ACAT1	Alpha-methylacetoacetic aciduria
Alport syndrome	COL4A5	COL4A5 related X-linked Alport syndrome
Alport syndrome	COL4A4	COL4A4 related autosomal recessive Alport syndrome
Alport syndrome	COL4A3	COL4A3 related autosomal recessive Alport syndrome
Apparent mineralocorticoid excess	HSD11B2	Apparent mineralocorticoid excess
Arginase deficiency	ARG1	Argininaemia
Argininosuccinic aciduria	ASL	Argininosuccinic aciduria
Aromatic L-amino acid decarboxylase deficiency	DDC	Aromatic L-amino acid decarboxylase deficiency
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
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	TFAFAZZIN	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	BTB	Biotinidase deficiency
Branched-chain ketoacid dehydrogenase kinase deficiency	BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency
Carbamoyl phosphate synthetase I deficiency	CPS1	Carbamoyl phosphate synthetase I deficiency
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
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
Citrullinaemia	ASS1	Citrullinaemia
Combined immunodeficiency and megaloblastic anaemia with or without hyperhomocysteinaemia	MTHFD1	Combined immunodeficiency and megaloblastic anaemia with or without hyperhomocysteinaemia
Combined pituitary hormone deficiency	PROP1	Combined Pituitary hormone deficiency 2
Combined pituitary hormone deficiency	POU1F1	Pituitary hormone deficiency, combined or isolated, 1, autosomal dominant

Combined pituitary hormone deficiency	POU1F1	Pituitary hormone deficiency, combined or isolated, 1, autosomal recessive
Combined pituitary hormone deficiency	HESX1	Pituitary hormone deficiency, combined, 5
Combined pituitary hormone deficiency	LHX3	Pituitary hormone deficiency, combined, 3
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 hyperplasia	CYP17A1	17-alpha-hydroxylase/17,20-lyase deficiency
Congenital adrenal hypoplasia	NR0B1	Adrenal hypoplasia, congenital
Congenital adrenal insufficiency with 46XY sex reversal	CYP11A1	Congenital adrenal insufficiency with 46XY DSD
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	Diarrhoea 7, protein-losing enteropathy type
Congenital Diarrhoea	SLC26A3	Diarrhoea 1, secretory chloride, congenital
Congenital Diarrhoea	SLC9A3	Diarrhoea 8, secretory sodium, congenital
Congenital Diarrhoea	EPCAM	Diarrhoea 5, with tufting enteropathy, congenital
Congenital Diarrhoea	SPINT2	Diarrhoea 3, secretory sodium, congenital, syndromic
Congenital Diarrhoea	NEUROG3	Diarrhoea 4, malabsorptive, congenital
Congenital disorder of glycosylation	PGM1	Congenital disorder of glycosylation, type It
Congenital erythropoietic porphyria	UROS	Congenital erythropoietic porphyria
Congenital generalised lipodystrophy	AGPAT2	Congenital generalized lipodystrophy type 1
Congenital generalised lipodystrophy	BSCL2	Lipodystrophy, congenital generalized, type 2
Congenital generalised lipodystrophy	CAV1	Lipodystrophy, congenital generalized, type 3
Congenital generalised lipodystrophy	CAVIN1	Lipodystrophy, congenital generalized, type 4
Congenital hyperinsulinism	HADH	Familial hyperinsulinemic hypoglycemia-4
Congenital hyperinsulinism	HK1	HK1 related 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, autosomal recessive
Congenital hypoaldosteronism	CYP11B2	Hypoaldosteronism, congenital, due to CMO I deficiency
Congenital hypothyroidism	DUOX2	Thyroid dysmorphogenesis 6
Congenital hypothyroidism	DUOX2	Thyroid dysmorphogenesis 5
Congenital hypothyroidism	TPO	Thyroid dysmorphogenesis 2A
Congenital hypothyroidism	TG	Thyroid dysmorphogenesis 3
Congenital hypothyroidism	SLC5A5	Thyroid dysmorphogenesis 1
Congenital hypothyroidism	SLC26A7	Thyroid dysmorphogenesis (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 dysmorphogenesis 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 Myasthenic Syndrome	CHRNE	Myasthenic syndrome, congenital, 4, autosomal recessive
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, autosomal recessive
Congenital Myasthenic Syndrome	SYT2	Congenital myasthenic syndrome 7, autosomal dominant
Congenital Myasthenic Syndrome	CHAT	Congenital myasthenic syndrome-6
Congenital Myasthenic Syndrome	CHRNA1	Congenital myasthenic syndrome-1, autosomal recessive
Congenital Myasthenic Syndrome	CHRNA1	Congenital myasthenic syndrome-1, autosomal dominant
Congenital Myasthenic Syndrome	CHRNA1	Congenital myasthenic syndrome-2, autosomal recessive
Congenital Myasthenic Syndrome	CHRNA1	Congenital myasthenic syndrome-2, autosomal dominant

Congenital Myasthenic Syndrome	CHRND	Congenital myasthenic syndrome-3, autosomal recessive
Congenital Myasthenic Syndrome	CHRND	Congenital myasthenic syndrome-3, autosomal dominant
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	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
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 anaemia 7
Diamond Blackfan Anaemia	RPS26	Diamond-Blackfan anaemia 10
Diamond Blackfan Anaemia	RPL35A	Diamond-Blackfan anaemia 5
Diamond Blackfan Anaemia	RPS10	Diamond-Blackfan anaemia 9
Diamond Blackfan Anaemia	RPS17	Diamond-Blackfan anaemia 4
Diamond Blackfan Anaemia	RPS24	Diamond-blackfan anaemia 3
Diamond Blackfan Anaemia	RPL15	Diamond-Blackfan anaemia 12
Diamond Blackfan Anaemia	RPL31	RPL31 associated Diamond-Blackfan anaemia
Diamond Blackfan Anaemia	RPS7	Diamond-Blackfan anaemia 8
Distal renal tubular acidosis	ATP6V0A4	Distal renal tubular acidosis type 3 with or without sensorineural hearing loss

Distal renal tubular acidosis	ATP6V1B1	Distal renal tubular acidosis type 2 with progressive sensorineural hearing loss
Distal renal tubular acidosis	SLC4A1	Distal renal tubular acidosis type 1, autosomal dominant
Distal renal tubular acidosis	SLC4A1	Distal renal tubular acidosis type 4 with hemolytic anaemia, autosomal recessive
DOCK8 Deficiency	DOCK8	DOCK8 Deficiency
Dopa responsive dystonia	TH	Dopa-responsive dystonia due to tyrosine hydroxylase deficiency
Early onset osteoporosis	PLS3	Bone mineral density QTL18, osteoporosis
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 XIII A deficiency
Factor XIII Deficiency	F13B	Factor XIII B deficiency
Familial Chylomicronaemia Syndrome	GPIHBP1	Hyperlipoproteinemia, type 1D
Familial Chylomicronaemia Syndrome	APOC2	Hyperlipoproteinemia, type 1b
Familial Chylomicronaemia Syndrome	LMF1	Lipase deficiency, combined
Familial Chylomicronaemia Syndrome	LPL	Lipoprotein lipase deficiency
Familial Chylomicronaemia Syndrome	APOA5	apolipoprotein A-V deficiency
Familial Haemophagocytic lymphohistiocytosis	PRF1	Haemophagocytic lymphohistiocytosis, familial, 2
Familial Haemophagocytic lymphohistiocytosis	UNC13D	Haemophagocytic lymphohistiocytosis, familial, 3
Familial Haemophagocytic lymphohistiocytosis	STX11	Haemophagocytic lymphohistiocytosis, familial, 4
Familial Haemophagocytic lymphohistiocytosis	STXBP2	Familial haemophagocytic 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, Autosomal Recessive
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	MC2R familial glucocorticoid deficiency
Glucocorticoid deficiency	MRAP	MRAP familial glucocorticoid deficiency type 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, Autosomal Dominant
GLUT1 deficiency syndrome	SLC2A1	GLUT1 deficiency syndrome-1, Autosomal Recessive
Glutaric aciduria type I	GCDH	Glutaric aciduria, type I
Glycogen storage disease type I	G6PC1	Glycogen storage disease type Ia
Glycogen storage disease type I	SLC37A4	Glycogen storage disease type Ib and 1c
Glycogen storage disease type II (Pompe Disease)	GAA	Glycogen storage disease type II
Glycogen storage disease type III	AGL	Glycogen storage disease type 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 folate malabsorption	SLC46A1	Hereditary folate malabsorption
Hereditary fructose intolerance	ALDOB	Hereditary fructose intolerance
Heritable Retinoblastoma	RB1	Retinoblastoma
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-megaloblastic anaemia	MTRR	Homocystinuria-megaloblastic anaemia, cbl E type
Homocystinuria-megaloblastic anaemia	MTR	Homocystinuria-megaloblastic anaemia, cblG complementation type
Homozygous Familial hypercholesterolaemia	LDLR	Homozygous Familial hypercholesterolaemia-1

Homozygous Familial hypercholesterolaemia	LDLRAP1	Homozygous Familial hypercholesterolaemia-4
Homozygous Familial hypercholesterolaemia	APOB	Hypercholesterolaemia, familial, 2 autosomal recessive
Homozygous Familial hypercholesterolaemia	PCSK9	Familial hypercholesterolaemia-3 Autosomal Recessive
Homozygous Variegate Porphyria	PPOX	Homozygous Variegate Porphyria
Hyperinsulinism-hyperammonaemia syndrome	GLUD1	Hyperinsulinism-hyperammonaemia syndrome
Hypermanganesaemia with dystonia	SLC30A10	Hypermanganesaemia with dystonia 1
Hyperornithinaemia-hyperammonaemia-homocitrullinuria syndrome	SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
Hyperphenylalaninaemia	PTS	Hyperphenylalaninemia due to 6-pyruvoyl-tetrahydropterin synthase deficiency
Hyperphenylalaninaemia	QDPR	Hyperphenylalaninemia due to dihydropteridine reductase deficiency
Hypobetalipoproteinaemia	APOB	Hypobetalipoproteinaemia
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	PHEX	Hypophosphatemic rickets, X-linked dominant
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
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
	SH2D1A	X-Linked Lymphoproliferative Syndrome 1
	XIAP	X-Linked inhibitor of apoptosis protein
	ITK	Lymphoproliferative syndrome 1

	CD70	Lymphoproliferative syndrome 3
Immunodeficiency	STAT1	Immunodeficiency 31B
Immunodeficiency	LCK	immunodeficiency 22
		T-cell immunodeficiency, congenital alopecia, and nail dystrophy
Immunodeficiency	FOXN1	Immunodeficiency due to purine nucleoside phosphorylase deficiency
Immunodeficiency	PNP	Immunodeficiency 26, with or without neurologic abnormalities
Immunodeficiency	PRKDC	Immunodeficiency 52
Immunodeficiency	LAT	Immunodeficiency 40
Immunodeficiency	DOCK2	Immunodeficiency 73B
Immunodeficiency	RAC2	Immunodeficiency 27A, mycobacteriosis, Autosomal dominant
		Immunodeficiency 27A, mycobacteriosis, autosomal recessive
Immunodeficiency	IFNGR1	Immunodeficiency 28, mycobacteriosis
Immunodeficiency	IFNGR2	Immunodeficiency 76
Immunodeficiency	FCHO1	Immunodeficiency 15B
Immunodeficiency	IKBKB	Immunodeficiency 24
Immunodeficiency	CTPS1	IRAK4 deficiency
Immunodeficiency	IRAK4	Immunodeficiency 68
Immunodeficiency	MYD88	STK4 associated T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations
Immunodeficiency	STK4	LIG1 associated immunodeficiency
Immunodeficiency	LIG1	X-linked Immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia
Immunodeficiency	MAGT1	Immunodeficiency 64
Immunodeficiency	RASGRP1	Immunodeficiency 32B
Immunodeficiency	IRF8	Immunodeficiency 54
Immunodeficiency	MCM4	
Immunodeficiency	ARPC1B	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease
Immunodeficiency with hyper-IgM	CD40	Immunodeficiency with hyper-IgM, type 3
		X-linked immunodeficiency with hyper-IgM type 1
Immunodeficiency with hyper-IgM	CD40LG	Immunodeficiency 41 with lymphoproliferation and autoimmunity
Immunodeficiency with lymphoproliferation and autoimmunity	IL2RA	Immunodeficiency 63 with lymphoproliferation and autoimmunity
Immunodeficiency with lymphoproliferation and autoimmunity	IL2RB	Immunodeficiency-centromeric instability-facial anomalies syndrome 1
Immunodeficiency-centromeric instability-facial anomalies syndrome	DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 2
Immunodeficiency-centromeric instability-facial anomalies syndrome	ZBTB24	

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, autosomal recessive
Isolated growth hormone deficiency	GH1	Isolated growth hormone deficiency type 1B, autosomal recessive
Isolated growth hormone deficiency	GH1	Isolated growth hormone deficiency type II, autosomal dominant
Isolated growth hormone deficiency	GHRHR	Isolated growth hormone deficiency type 4
Isolated growth hormone deficiency	RNPC3	RNPC3 associated growth hormone deficiency
Isolated methylmalonic acidemia	MMAA	Methylmalonic aciduria, vitamin B12-responsive, cblA type
Isolated methylmalonic acidemia	MMUT	Methylmalonic acidemia, mut(0) type
Isolated methylmalonic acidemia	MMADHC	Methylmalonic aciduria, cblD type, variant 2
Isolated methylmalonic acidemia	MMAB	Methylmalonic aciduria, vitamin B12-responsive, cblB type
Isolated methylmalonic acidemia	MCEE	Methylmalonyl-CoA epimerase deficiency
Isovaleric acidemia	IVD	Isovaleric acidemia
Kenny-Caffey syndrome, type 2	FAM111A	Kenny-Caffey syndrome, type 2
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	STAR deficiency (congenital lipoid hyperplasia)
Long Chain Hydroxyacyl CoA Dehydrogenase Deficiency, LCHAD	HADHA	LCHAD deficiency
Lysinuric protein intolerance	SLC7A7	Lysinuric protein intolerance
Mannose Phosphate Isomerase (MPI) - Congenital Disorders of Glycosylation	MPI	Mannose Phosphate Isomerase (MPI) - Congenital Disorders of Glycosylation
Maple Syrup Urine Disease	BCKDHA	Maple syrup urine disease, type Ia
Maple Syrup Urine Disease	BCKDHB	Maple syrup urine disease, type Ib
Maple Syrup Urine Disease	DBT	Maple syrup urine disease, type II
Medium-chain acyl-CoA dehydrogenase deficiency	ACADM	Medium-chain acyl-CoA dehydrogenase deficiency
Megaloblastic anaemia	SLC19A1	Folate dependent megaloblastic anaemia
Megaloblastic anaemia	SLC19A2	Thiamine Responsive Megaloblastic Anaemia Syndrome

Megaloblastic anaemia	DHFR	Megaloblastic anaemia due to dihydrofolate reductase deficiency
Metachromatic leukodystrophy	ARSA	Metachromatic leukodystrophy
Methylenetetrahydrofolate Reductase Deficiency	MTHFR	Homocystinuria due to MTHFR deficiency
Methylmalonic acidemia and homocystinuria	MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
Methylmalonic acidemia and homocystinuria	MMADHC	Methylmalonic aciduria and homocystinuria, cblD type
Methylmalonic acidemia and homocystinuria	LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type
MIRAGE syndrome	SAMD9	MIRAGE syndrome
Mucopolysaccharidosis Type I	IDUA	Mucopolysaccharidosis Type I
Mucopolysaccharidosis Type II	IDS	Mucopolysaccharidosis Type II
Mucopolysaccharidosis Type IVA	GALNS	Mucopolysaccharidosis Type IVA
Mucopolysaccharidosis Type VI	ARSB	Mucopolysaccharidosis Type VI
Multiple Acyl-CoA Dehydrogenase Deficiency	ETFB	Glutaric acidemia type IIB
Multiple Acyl-CoA Dehydrogenase Deficiency	ETFDH	Glutaric acidemia type IIC
Multiple Acyl-CoA Dehydrogenase Deficiency	ETFA	Glutaric acidemia type IIA
N-acetylglutamate synthase deficiency	NAGS	N-acetylglutamate synthase deficiency
Neonatal diabetes	ABCC8	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, autosomal recessive
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	NEUROD1 related autosomal recessive neonatal diabetes
Neonatal hyperparathyroidism	CASR	Neonatal hyperparathyroidism, Autosomal Recessive
Neonatal hyperparathyroidism	CASR	Neonatal hyperparathyroidism, Autosomal Dominant
Nephrotic syndrome with or without adrenal insufficiency	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 related polymorphic autoinflammatory immunodeficiency
Ornithine transcarbamylase deficiency	OTC	Ornithine transcarbamylase deficiency
Osteogenesis Imperfecta	COL1A1	COL1A1 related Osteogenesis Imperfecta
Osteogenesis Imperfecta	COL1A2	COL1A2 related Osteogenesis Imperfecta
Osteogenesis Imperfecta	IFITM5	Osteogenesis Imperfecta type V
Osteopetrosis	TCIRG1	Osteopetrosis type 1
Osteopetrosis	CLCN7	Osteopetrosis type 4, autosomal recessive
Osteopetrosis	CLCN7	Osteopetrosis type 4, autosomal dominant
Osteopetrosis	TNFRSF11A	Osteopetrosis type 7
Osteopetrosis	SNX10	Osteopetrosis type 8
Osteopetrosis	CA2	Osteopetrosis with renal tubular acidosis
Osteoporosis-pseudoglioma syndrome	LRP5	Osteoporosis-pseudoglioma syndrome
OTOF related deafness	OTOF	Auditory neuropathy, autosomal recessive, 1
Otofaciocervical syndrome	PAX1	Otofaciocervical syndrome 2
Pancreatic agenesis	PTF1A	Pancreatic agenesis 2
Periodic fever syndrome	NLRP3	Cryopyrin associated periodic fever syndrome
Phenylketonuria	PAH	Phenylketonuria
POMC deficiency	POMC	Obesity, adrenal insufficiency, and red hair due to POMC deficiency
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	DNAH9	Ciliary dyskinesia, primary, 40
Primary Ciliary Dyskinesia	DRC1	Ciliary dyskinesia, primary, 21
Primary Ciliary Dyskinesia	GAS8	Ciliary dyskinesia, primary, 33
Primary coenzyme Q10 deficiency	COQ4	COQ4 related primary coenzyme Q ₁₀ deficiency
Primary coenzyme Q10 deficiency	COQ8B	COQ8B related primary coenzyme Q10 deficiency with nephrotic syndrome
Primary coenzyme Q10 deficiency	COQ2	COQ2 related primary coenzyme Q ₁₀ deficiency
Primary coenzyme Q10 deficiency	COQ6	COQ6 related primary coenzyme Q ₁₀ deficiency
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	SCNN1A related pseudohypoaldosteronism Type 1A
Pseudohypoaldosteronism	SCNN1B	SCNN1B related pseudohypoaldosteronism Type 1A
Pseudohypoaldosteronism	SCNN1G	SCNN1G related pseudohypoaldosteronism Type 1A
Pseudohypoaldosteronism	NR3C2	Pseudohypoaldosteronism type I, autosomal dominant
Pseudohypoaldosteronism	WNK1	Pseudohypoaldosteronism, type IIC
Pseudohypoaldosteronism	KLHL3	Pseudohypoaldosteronism, type IID, autosomal recessive
Pseudohypoaldosteronism	KLHL3	Pseudohypoaldosteronism, type IID, autosomal dominant
Pseudohypoaldosteronism	CUL3	Pseudohypoaldosteronism, type IIE
PSTPIP1 related inflammatory disease	PSTPIP1	PSTPIP1 associated inflammatory disease
Pyridoxamine 5-prime-phosphate oxidase deficiency	PNPO	Pyridoxamine 5'-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)

Riboflavin transporter deficiency	SLC52A3	Riboflavin transporter deficiency (Brown-Vialetto-Van Laere syndrome 1)
Riboflavin transporter deficiency	SLC52A2	Riboflavin transporter deficiency (Brown-Vialetto-Van Laere syndrome 2)
RPE65 related Leber congenital amaurosis/early-onset severe retinal dystrophy	RPE65	RPE65 associated Leber congenital amaurosis , early-onset severe retinal dystrophy
SCID	ADA	Severe combined immunodeficiency due to ADA deficiency
SCID	IL2RG	SCID X-Linked
		JAK3 related T cell-negative, B cell-positive, natural killer cell-negative severe combined immunodeficiency
SCID	JAK3	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation
SCID	NHEJ1	Immunodeficiency 104, severe combined
SCID	IL7R	Immunodeficiency 105, severe combined
SCID	PTPRC	RAG1 related severe combined immunodeficiency, B cell-negative
SCID	RAG1	RAG2 related severe combined immunodeficiency, B cell-negative
SCID	RAG2	Severe combined immunodeficiency with sensitivity to ionising radiation
SCID	DCLRE1C	Immunodeficiency 19, severe combined
SCID	CD3D	Immunodeficiency 18
SCID	CD3E	
SCID	CD3G	Immunodeficiency 17, CD3 gamma deficient
SCID	CD247	Immunodeficiency 25, severe combined
SCID	AK2	Reticular Dysgenesis
SCID	CORO1A	Immunodeficiency 8
SCID	ZAP70	Immunodeficiency 48, 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
		DNAJC21 related Shwachman-Diamond syndrome
Shwachman-Diamond syndrome	DNAJC21	
Shwachman-Diamond syndrome	SRP54	SRP54 related Shwachman-Diamond syndrome
Sickle Cell Disease	HBB	Sickle Cell Disease
Specific complement deficiency	CFB	Complement factor B deficiency
		Complement factor H deficiency Autosomal recessive
Specific complement deficiency	CFH	
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
Spinal muscular atrophy	SMN1	Spinal Muscular Atrophy
Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	SLC19A3	Sodium-dependent multivitamin transporter deficiency
Thiamine metabolism dysfunction syndrome 5	TPK1	Thiamine Pyrophosphokinase Deficiency
Thyroid hormone resistance	THRB	Thyroid hormone resistance, autosomal dominant
Thyroid hormone resistance	THRB	Thyroid hormone resistance, autosomal recessive
Transcobalamin II deficiency	TCN2	Transcobalamin II deficiency
Trichohepatoenteric syndrome	SKIC3	Trichohepatoenteric syndrome 1
Trichohepatoenteric syndrome	SKIC2	Trichohepatoenteric syndrome 2
Trifunctional protein deficiency	HADHB	Mitochondrial trifunctional protein deficiency 2
Tyrosinaemia, type I	FAH	Tyrosinemia, type I
Tyrosinaemia, type II	TAT	Tyrosinemia, type II
Uridine-responsive epileptic encephalopathy	CAD	Uridine-responsive epileptic encephalopathy
Very early onset inflammatory bowel disease	IL10	Interleukin-10 deficiency
Very early onset inflammatory bowel disease	IL10RB	Inflammatory bowel disease 25
Very early onset inflammatory bowel disease	IL10RA	Inflammatory bowel disease 28
Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency	ACADVL	VLCAD deficiency
Vitamin B6-dependent epilepsy	PLPBP	Vitamin B6-dependent epilepsy
Vitamin D dependent rickets	CYP27B1	Vitamin D-dependent rickets, type I
Vitamin D dependent rickets	CYP2R1	Rickets due to defect in vitamin D 25-hydroxylation deficiency
Vitamin D resistant rickets	VDR	Rickets, vitamin D-resistant, type IIA
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 tumour predisposition

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 immunodysregulation, polyendocrinopathy, and enteropathy	FOXP3	X-linked immunodysregulation, polyendocrinopathy, and enteropathy