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)
Adrenocorticotropic hormone deficiency	**TBX19**	Adrenocorticotropic 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	**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
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 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 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	**CHRNB1**	Congenital myasthenic syndrome-2, autosomal recessive
Congenital Myasthenic Syndrome	**CHRNB1**	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 XIIIA deficiency
Factor XIII Deficiency	**F13B**	Factor XIIIB deficiency
Familial Chylomicronaemia Syndrome	**GPIHBP1**	Hyperlipoproteinemia, type 1D
Familial Chylomicronaemia Syndrome	**APOC2**	Hyperlipoproteinemia, type Ib
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
Hyperphenyalaninaemia	**PTS**	Hyperphenylalaninemia due to 6-pyruvoyl-tetrahydropterin synthase deficiency
Hyperphenyalaninaemia	**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
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	**LAT**	Immunodeficiency 52
Immunodeficiency	**DOCK2**	Immunodeficiency 40
Immunodeficiency	**RAC2**	Immunodeficiency 73B
Immunodeficiency	**IFNGR1**	Immunodeficiency 27A, mycobacteriosis, Autosomal dominant
Immunodeficiency	**IFNGR1**	Immunodeficiency 27A, mycobacteriosis, autosomal recessive
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	**STK4**	STK4 associated T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations
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	**ARPC1B**	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease
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, 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 acidaemia	**MMAA**	Methylmalonic aciduria, vitamin B12-responsive, cblA type
Isolated methylmalonic acidaemia	**MMUT**	Methylmalonic acidemia, mut(0) type
Isolated methylmalonic acidaemia	**MMADHC**	Methylmalonic aciduria, cblD type, variant 2
Isolated methylmalonic acidaemia	**MMAB**	Methylmalonic aciduria, vitamin B12-responsive, cblB type
Isolated methylmalonic acidaemia	**MCEE**	Methylmalonyl-CoA epimerase deficiency
Isovaleric acidaemia	**IVD**	Isovaleric acidaemia
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 Repsonsive 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 acidaemia and homocystinuria	**MMACHC**	Methylmalonic aciduria and homocystinuria, cblC type
Methylmalonic acidaemia and homocystinuria	**MMADHC**	Methylmalonic aciduria and homocystinuria, cblD type
Methylmalonic acidaemia 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 acidaemia type IIB
Multiple Acyl-CoA Dehydrogenase Deficiency	**ETFDH**	Glutaric acidaemia type IIC
Multiple Acyl-CoA Dehydrogenase Deficiency	**ETFA**	Glutaric acidaemia 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 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 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
SCID	**JAK3**	JAK3 related 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**	RAG1 related severe combined immunodeficiency, B cell-negative
SCID	**RAG2**	RAG2 related severe combined immunodeficiency, B cell-negative
SCID	**DCLRE1C**	Severe combined immunodeficiency with sensitivity to ionising radiation
SCID	**CD3D**	Immunodeficiency 19, severe combined
SCID	**CD3E**	Immunodeficiency 18
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
Shwachman-Diamond syndrome	**DNAJC21**	DNAJC21 related Shwachman-Diamond syndrome
Shwachman-Diamond syndrome	**SRP54**	SRP54 related Shwachman-Diamond syndrome
Sickle Cell Disease	**HBB**	Sickle Cell Disease
Specific complement deficiency	**CFB**	Complement factor B deficiency
Specific complement deficiency	**CFH**	Complement factor H deficiency Autosomal recessive
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