Gene and Condition List

What conditions are being screened in BabyScreen+?

The focus of genomic newborn screening is to identify newborns who have severe, childhood-onset conditions for which treatment is available. Early diagnosis and treatment provide these babies with the best health outcomes. Screening is designed to provide genetic information that is relevant and useful, while reducing uncertain and unclear information.

Genomic newborn screening through BabyScreen+ looks at over 500 genes. The conditions included can affect babies in different ways and can involve one or many different parts of the body. Some types of conditions screened include:

Metabolic conditions
Conditions that impact the child's ability to convert food into specific nutrients and chemicals that the body requires. These are often managed with diet modification and enzyme replacement therapy.

Endocrine conditions
Conditions that occur due to a change in the function or quantity of hormones produced in the body. These are often treated with hormone replacements.

Immunological conditions
Conditions caused by the immune system not functioning as expected. Sometimes treatment is with medications or a bone marrow transplant.

The impact of these conditions on a baby depends on early access to treatment and management. Other options for treatment and management depend on the condition and can include:

- Drugs and supplements
- Organ transplant
- Diet modification
- Gene therapy

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How are the conditions selected?

To select the conditions and genes to be screened, a group of experts was established. This included clinical geneticists, genetic scientists, genetic counsellors, specialist physicians and a patient support organisation.

For a gene to be included in the screening, it must meet the following criteria:

- The gene is known to cause a condition
- Screening the gene is technically possible with confidence using currently available technology
- The condition associated with the gene has a serious impact on a person's health and/or is life limiting
- The condition associated with the gene usually affects children under 5 years of age
- There is available treatment that prevents, cures or significantly reduces the severity of the condition

What conditions are not screened?

Genomic newborn screening through BabyScreen+ does not look for:

- Mild conditions
- Conditions that do not usually start in childhood
- Conditions where treatment is not available in early childhood
- Genetic changes that have no known health impacts for the newborn (e.g. being a carrier for a genetic condition)

What are the limitations of screening?

There are many different types of genetic changes that can cause genetic conditions. The genetic testing offered through this study is called 'screening', because the technology used will detect many, but not all genetic changes.

This means newborns with a 'low chance' result still have a small chance of having the genetic conditions screened. This is because:

- Screening may not cover all the genes linked with a condition. This may be because a gene is associated with a mild form of the condition, or there are technical challenges in screening that gene.
- The test cannot detect all types of changes in the genes screened.

Newborns who receive a 'high chance' result also have a small chance of not having the condition. All high chance results will be confirmed through additional testing.

Knowledge about our genes is changing every day. Results from genomic newborn screening performed through this study are being analysed and interpreted by experienced laboratory scientists. Interpretation of genetic information is based on currently available information.

What should I do if I have a family history of a genetic condition?

Genomic newborn screening is relevant to all newborns, regardless of whether there is a family history of a genetic condition. If you have a family history of a genetic condition and are participating in BabyScreen+, please provide this information during enrolment. If you have already completed enrolment, you can contact our study team to tell us about your family history. We can use this information to check whether BabyScreen+ is able to detect the genetic change or condition in your family.

Is BabyScreen+ relevant for my baby if I have had reproductive carrier screening and/or non-invasive prenatal testing (NIPT)?

Yes, BabyScreen+ is still relevant for your baby even if you have had reproductive carrier screening, NIPT, and/or any other genetic or chromosome test. Even if you have returned a 'low risk' result in these tests, it is still possible that your baby may have one of the conditions screened in BabyScreen+.

If you would like to talk about reproductive carrier screening or NIPT, please speak with your healthcare team or contact our study team.

How can I contact the study team?

Our study team includes experienced genetic counsellors, clinical geneticists and laboratory scientists. Please don't hesitate to contact us if you have any queries about the conditions screened through BabyScreen+.

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List of genes and conditions screened in BabyScreen+

Please note that some genes are associated with more than one condition.

Gene	Condition
AAAS	Achalasia-addisonianism-alacrimia syndrome
ABCC6	Generalized arterial calcification of infancy, type 2
ABCC8	Hyperinsulinemic hypoglycemia, familial
ABCD1	Adrenoleukodystrophy
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type
ABCG5	Sitosterolaemia 2
ACAD9	Mitochondrial complex I deficiency, nuclear type 20
ACADM	Medium chain acyl CoA dehydrogenase deficiency
ACADVL	VLCAD deficiency
ACAT1	Alpha-methylacetoacetic aciduria
ACTA2	Aortic aneurysm, familial thoracic 6
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2
ADA	Severe combined immunodeficiency due to ADA deficiency
ADA2	Vasculitis, autoinflammation, immunodeficiency, and haematologic defects syndrome
ADAMTS13	Thrombotic thrombocytopenic purpura, familial
ADGRV1	Usher syndrome, type 2C
AGL	Glycogen storage disease IIIa
AGRN	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects
AGXT	Hyperoxaluria, primary, type 1
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
AICDA	Immunodeficiency with hyper-IgM, type 2
AIRE	Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia
AK2	Reticular dysgenesis
AKR1D1	Bile acid synthesis defect, congenital, 2
ALDH4A1	Hyperprolinemia, type II
ALDH7A1	Epilepsy, pyridoxine-dependent
ALDOB	Fructose intolerance, hereditary
ALPL	Hypophosphatasia, childhood
	Hypophosphatasia, infantile
AMACR	Bile acid synthesis defect, congenital, 4
AMN	Megaloblastic anaemia-1, Norwegian type
AP3B1	Hermansky-Pudlak syndrome 2
AQP2	Diabetes insipidus, nephrogenic, 2
ARG1	Arginase deficiency
ARPC1B	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia
ARSA	Metachromatic leukodystrophy
ARSB	Mucopolysaccharidosis VI

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Gene	Condition
ASL	Argininosuccinic aciduria
ASS1	Citrullinaemia
ATP6V0A4	Distal renal tubular acidosis 3, with or without sensorineural hearing loss
ATP6V1B1	Distal renal tubular acidosis 2, with progressive sensorineural hearing loss
ATP7A	Menkes disease
ATP7B	Wilson disease
AVP	Diabetes insipidus, neurohypophyseal
AVPR2	Diabetes insipidus, nephrogenic
BCHE	Butyrylcholinesterase deficiency
BCKDHA	Maple syrup urine disease, type Ia
BCKDHB	Maple syrup urine disease, type Ib
BCKDK	Branched-chain keto acid dehydrogenase kinase deficiency
BLNK	Agammaglobulinaemia 4
BMP1	Osteogenesis imperfecta, type XIII
BRCA1	Fanconi anaemia, complementation group S
BRCA2	Fanconi anaemia, complementation group D1
BRIP1	Fanconi anaemia, complementation group J
BSCL2	Lipodystrophy, congenital generalized, type 2
	Berardinelli-Seip lipodystrophy
BSND	Bartter syndrome, type 4a
BTD	Biotinidase deficiency
BTK	Agammaglobulinemia, X-linked 1
C17orf62 (CYBC1)	Chronic granulomatous disease 5, autosomal recessive
C2	C2 deficiency
C3	C3 deficiency
C5	C5 deficiency
C6	C6 deficiency
C7	C7 deficiency
C8B	C8 deficiency, type II
C9	C9 deficiency
CA12	Hyperchlorhidrosis, isolated
CA2	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis
CA5A	Hyperammonaemia due to carbonic anhydrase VA deficiency
CABP2	Deafness, autosomal recessive 93
CACNA1S	Malignant hyperthermia susceptibility 5
CAD	Developmental and epileptic encephalopathy 50
CALM3	Long QT syndrome 16
CARD11	Immunodeficiency 11A, autosomal recessive
	Immunodeficiency 11B with atopic dermatitis, autosomal dominant
CASR	Hypocalcemia, autosomal dominant
	Hyperparathyroidism, neonatal
CAV1	Lipodystrophy, congenital generalized, type 3
CAVIN1	Lipodystrophy, congenital generalized, type 4

Gene	Condition
CBS	Homocystinuria
CD19	Immunodeficiency, common variable, 3
CD247	Immunodeficiency 25
CD27	CD27-deficiency
CD3D	Immunodeficiency 19
CD3E	Immunodeficiency 18
CD3G	Immunodeficiency 17, CD3 gamma deficient
CD40	Immunodeficiency with hyper-IgM, type 3
CD40LG	Immunodeficiency, X-linked, with hyper-IgM
CD55	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy
CD70	Lymphoproliferative syndrome 3
CD79A	Agammaglobulinaemia 3
CD79B	Agammaglobulinaemia 6
CDC14A	Deafness, autosomal recessive 32, with or without immotile sperm
CDCA7	Immunodeficiency-centromeric instability-facial anomalies syndrome 3
CDCA8	Congenital hypothyroidism
CDH23	Usher syndrome, type 1D
	Deafness, autosomal recessive 12
	Usher syndrome, type 1D/F digenic
CDKN1C	IMAGe syndrome
CEBPE	Specific granule deficiency
CFD	Complement factor D deficiency
CFH	Complement factor H deficiency
CFI	Complement factor I deficiency
CFP	Properdin deficiency, X-linked
CFTR	Cystic fibrosis
CHAT	Congenital myasthenic syndrome
CHRNA1	Myasthenic syndrome, congenital, 1A, slow-channel
	Myasthenic syndrome, congenital, 1B, fast-channel
CHRNB1	Myasthenic syndrome, congenital, 2A, slow-channel
	Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency
CHRND	Myasthenic syndrome, congenital, 3B, fast-channel
	Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency
	Myasthenic syndrome, congenital, 3A, slow-channel
	Multiple pterygium syndrome, lethal type
CHRNE	Myasthenic syndrome, congenital, 4B, fast-channel
	Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency
	Myasthenic syndrome, slow-channel congenital
	Myasthenic syndrome, congenital, 4A, slow-channel
CIB2	Deafness, autosomal recessive 48

Gene	Condition
CIITA	Bare Lymphocyte Syndrome, type II, complementation group A
CLCN7	Osteopetrosis, autosomal recessive 4
CLDN14	Deafness, autosomal recessive 29
CLPP	Perrault syndrome 3
COCH	Deafness, autosomal recessive 110
COL11A1	Stickler syndrome, type II
COL11A2	Deafness, autosomal recessive 53
COL13A1	Myasthenic syndrome, congenital, 19
COL1A1	Osteogenesis imperfecta, type I
COL1A2	Osteogenesis imperfecta, type II
COL2A1	Stickler syndrome, type I
COL4A3	Alport syndrome 2, autosomal recessive
COL4A4	Alport syndrome 2, autosomal recessive
COL4A5	Alport syndrome 1, X-linked
COL9A1	Stickler syndrome, type IV
COL9A2	Stickler syndrome, type V
COL9A3	Stickler syndrome, type VI
COLQ	Congenital myasthenic syndrome
COQ2	Coenzyme Q10 deficiency, primary, 1
COQ4	Coenzyme Q10 deficiency, primary, 7
COQ6	Coenzyme Q10 deficiency, primary, 6
COQ8A	Coenzyme Q10 deficiency, primary, 4
CORO1A	Immunodeficiency 8
CPS1	Carbamoylphosphate synthetase I deficiency
CPT1A	Carnitine palmitoyltransferase I deficiency
CPT2	CPT II deficiency, infantile
	CPT II deficiency, lethal neonatal
	CPT II deficiency, myopathic, stress-induced
CRTAP	Osteogenesis imperfecta, type VII
CSF3R	Neutropenia, severe congenital, 7, autosomal recessive
CTNS	Cystinosis, nephropathic
CTPS1	Immunodeficiency 24
CUBN	Megaloblastic anaemia-1, Finnish type
CUL3	Pseudohypoaldosteronism, type IIE
CXCR4	WHIM syndrome 1
CYB561	Orthostatic hypotension 2
CYBA	Chronic granulomatous disease
CYBB	Chronic granulomatous disease
CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
CYP11B2	Hypoaldosteronism, congenital, due to CMO I deficiency
	Hypoaldosteronism, congenital, due to CMO II deficiency

Gene	Condition
CYP17A1	17,20-lyase deficiency, isolated
CYP21A2	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency
CYP27A1	Cerebrotendinous xanthomatosis
CYP27B1	Vitamin D-dependent rickets, type I
CYP2R1	Rickets due to defect in vitamin D 25-hydroxylation deficiency
CYP7B1	Bile acid synthesis defect, congenital, 3
DBT	Maple syrup urine disease
DCLRE1C	Severe combined immunodeficiency, Athabascan type
	Omenn syndrome
DDC	Aromatic L-amino acid decarboxylase deficiency
DFNB59	Deafness, autosomal recessive 59
DGAT1	Diarrhea 7, protein-losing enteropathy type
DHCR7	Smith-Lemli-Opitz syndrome
DHFR	Megaloblastic anaemia due to dihydrofolate reductase deficiency
DICER1	DICER1 syndrome
DLAT	Pyruvate dehydrogenase E2 deficiency
DMP1	Hypophosphatemic rickets
DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient
DNAJC21	Bone marrow failure syndrome 3
DNASE2	Autoinflammatory-pancytopenia syndrome
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1
DOCK2	Immunodeficiency 40
DOCK8	Hyper-IgE syndrome
DOK7	Congenital myasthenic syndrome
DPAGT1	Congenital disorder of glycosylation, type Ij
	DPAGT1-CDG
	Myasthenic syndrome, congenital, 13, with tubular aggregates
DUOX2	Thyroid dyshormonogenesis 6
DUOXA2	Thyroid dyshormonogenesis 5
ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency
EDN3	Waardenburg syndrome, type 4B
EDNRB	Waardenburg syndrome, type 4A
EFL1	Shwachman-Diamond syndrome 2
EIF2AK3	Wolcott-Rallison syndrome
ELANE	Neutropenia, congenital
ENG	Telangiectasia, hereditary hemorrhagic, type 1
ENPP1	Generalised arterial calcification of infancy, type 1
	Hypophosphatemic rickets, autosomal recessive, 2
EPS8	Autosomal recessive nonsyndromic hearing loss 102
ERCC4	Fanconi anaemia, complementation group Q
ESPN	Deafness, autosomal recessive 36
ESRRB	Deafness, autosomal recessive 35
ETFA	Glutaric acidaemia IIA

Gene	Condition
ETFB	Glutaric acidemia IIB
ETFDH	Glutaric acidemia IIC
ETHE1	Ethylmalonic encephalopathy
F10	Factor X deficiency
F13A1	Factor XIIIA deficiency
F13B	Factor XIIIB deficiency
F7	Factor VII deficiency
F9	Haemophilia B
FAH	Tyrosinaemia, type I
FAM111A	Kenny-Caffey syndrome, type 2
FANCA	Fanconi anaemia, complementation group A
FANCB	Fanconi anaemia, complementation group B
FANCC	Fanconi anaemia, complementation group C
FANCD2	Fanconi anaemia, complementation group D2
FANCG	Fanconi anaemia
FANCI	Fanconi anaemia
FBN1	Marfan syndrome
FBP1	Fructose-1,6-bisphosphatase deficiency
FCHO1	Immunodeficiency 76
FECH	Protoporphyria, erythropoietic, 1
FERMT3	Leukocyte adhesion deficiency, type III
FGA	Afibrinogenemia, congenital
FGB	Afibrinogenemia, congenital
FGF23	Autosomal dominant hypophosphatemic rickets
	Familial hyperphosphatemic tumoral calcinosis/hyperphosphatemic hyperostosis syndrome
FGF3	Deafness, congenital with inner ear agenesis, microtia, and microdontia
FGFR3	Achondroplasia
FGG	Afibrinogenemia, congenital
FH	Fumurase deficiency
FKBP10	Osteogenesis imperfecta, type XI
FLAD1	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency
FOLR1	Neurodegeneration due to cerebral folate transport deficiency
FOXA2	Hyperinsulinism
FOXE1	Bamforth-Lazarus syndrome
FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, autosomal recessive
	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant
FOXP3	IPEX syndrome
FUCA1	Fucosidosis
G6PC	Glycogen storage disease Ia
G6PC3	Neutropaenia, congenital

Gene	Condition
G6PD	Glucose-6-phosphate dehydrogenase deficiency
GAA	Glycogen storage disease II, Pompe disease
GALC	Krabbe disease
GALE	Galactose epimerase deficiency
GALK1	Galactokinase deficiency with cataracts
GALM	Galactosemia IV
GALNS	Mucopolysaccharidosis IVA
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 1
GALT	Galactosaemia
GAMT	Cerebral creatine deficiency syndrome 2
GATA2	Immunodeficiency 21
	Emberger syndrome
GATA3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia
GATA4	Neonatal diabetes mellitus, GATA4-related
GATM	Cerebral creatine deficiency syndrome 3
GBA	Gaucher disease type 1
GCDH	Glutaric aciduria, type I
GCH1	Hyperphenylalaninemia, BH4-deficient, B
	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia
GCK	Hyperinsulinemic hypoglycemia, familial
GCM2	Hyperparathyroidism 4
	Hypoparathyroidism, familial isolated 2
GFI1	Neutropenia, severe congenital 2, autosomal dominant
GGCX	Combined vitamin K-dependent clotting factors deficiency, type 1
GH1	Growth hormone deficiency, isolated, type IA
	Growth hormone deficiency, isolated, type II
	Kowarski syndrome
GHR	Growth hormone insensitivity, partial
	Laron dwarfism
GHRHR	Growth hormone deficiency, isolated, type IV
GIF	Intrinsic factor deficiency
GIPC3	Deafness, autosomal recessive 15
GJB2	Deafness, autosomal recessive 1A
GLA	Fabry disease
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism
GLRA1	Hyperekplexia, hereditary 1, autosomal dominant or recessive
GLUD1	Hyperinsulinism
GNAS	Pseudopseudohypoparathyroidism
	Pseudohypoparathyroidism
GOT2	Developmental and epileptic encephalopathy 82
GPIHBP1	Hyperlipoproteinemia, type 1D
	Familial chylomicronemia syndrome
GREB1L	Deafness, autosomal dominant 80

Gene	Condition
GRHPR	Hyperoxaluria, primary, type II
GRXCR1	Deafness, autosomal recessive 25
GUSB	Mucopolysaccharidosis VII
GYS2	Glycogen storage disease 0, liver
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency
HADHA	Mitochondrial trifunctional protein deficiency
	LCHAD deficiency
HADHB	Mitochondrial trifunctional protein deficiency
HAX1	Neutropenia, severe congenital 3, autosomal recessive
	Kostmann syndrome
HBB	Sickle cell anaemia
HELLS	Immunodeficiency-centromeric instability-facial anomalies syndrome 4
HESX1	Pituitary hormone deficiency, combined, 5
HGF	Deafness, autosomal recessive 39
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency
HK1	Hyperinsulinism, HK1-related
HLCS	Holocarboxylase synthetase deficiency
HMGCL	3-hydroxy-3-methylglutaric aciduria
HOGA1	Hyperoxaluria, primary, type III
HSD11B2	Apparent mineralocorticoid excess
HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency
HSD3B7	Bile acid synthesis defect, congenital, 1
ICOS	Immunodeficiency, common variable, 1
IDS	Mucopolysaccharidosis II (MPS2, Hunter syndrome)
IDUA	Mucopolysaccharidosis type I (MPS1)
IFITM5	Osteogenesis imperfecta, type V
IGF1	Insulin-like growth factor I deficiency
IGHM	Agammaglobulinaemia 1
IGLL1	Agammaglobulinaemia 2
IGSF1	Hypothyroidism, central, and testicular enlargement
IKBKB	Immunodeficiency 15B
IKZF1	Immunodeficiency, common variable, 13
IL10	Autoinflammatory syndrome, IL10-related
IL10RA	Inflammatory bowel disease 28, early onset, autosomal recessive
IL10RB	Inflammatory bowel disease 25, early onset, autosomal recessive
IL1RN	Interleukin 1 receptor antagonist deficiency
IL21R	Immunodeficiency 56
IL2RA	Immunodeficiency 41 with lymphoproliferation and autoimmunity
IL2RB	Immunodeficiency 63 with lymphoproliferation and autoimmunity
IL2RG	Severe combined immunodeficiency, X-linked
IL36RN	Psoriasis 14, pustular

Gene	Condition
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type
ILDR1	Deafness, autosomal recessive 42
INS	Diabetes mellitus, insulin-dependent, 2
	Diabetes mellitus, permanent neonatal 4
	Maturity-onset diabetes of the young, type 10
IRAK4	Immunodeficiency 67
IRF8	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive
IRS4	Hypothyroidism, congenital, nongoitrous, 9
ITGA2B	Glanzmann thrombasthaenia 1
ITGB2	Leukocyte adhesion deficiency
ITGB3	Glanzmann thrombasthenia 2
ITK	Lymphoproliferative syndrome 1
IVD	Isovaleric acidemia
IYD	Thyroid dyshormonogenesis 4
JAGN1	Neutropenia, severe congenital, 6, autosomal recessive
JAK3	SCID, autosomal recessive, T-negative/B-positive type
KCNH2	Long QT syndrome 2
KCNJ1	Bartter syndrome, type 2
KCNJ11	Diabetes mellitus, transient neonatal, 3
	Diabetes, permanent neonatal, with or without neurologic features
	Hyperinsulinemic hypoglycemia, familial, 2
KCNJ2	Andersen syndrome
KCNQ1	Long QT syndrome 1
	Jervell and Lange-Nielsen syndrome
KDELR2	Osteogenesis imperfecta 21
KLHL3	Pseudohypoaldosteronism, type IID
LAMA2	Muscular dystrophy, congenital, merosin deficient or partially deficient
LAT	Immunodeficiency 52
LDLR	Hypercholesterolemia, familial, 1
LEP	Obesity, morbid, due to leptin deficiency
LEPR	Obesity, morbid, due to leptin receptor deficiency
LHFPL5	Deafness, autosomal recessive 67
LHX3	Pituitary hormone deficiency, combined
LHX4	Pituitary hormone deficiency, combined, 4
LIG1	Immunodeficiency 96
LIG4	LIG4 syndrome
LIPA	Wolman syndrome
LMBRD1	Methylmalonic aciduria and homocystinuria
LOXHD1	Deafness, autosomal recessive 77
LPL	Lipoprotein lipase deficiency
LRBA	Immunodeficiency, common variable, 8, with autoimmunity

Gene	Condition
LRP5	Osteoporosis-pseudoglioma syndrome
LRTOMT	Deafness, autosomal recessive 63
LYST	Chediak-Higashi syndrome
MAFB	Multicentric carpotarsal osteolysis syndrome
MAGT1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia
MALT1	Immunodeficiency 12
MAN2B1	Mannosidosis, alpha-, types I and II
MARVELD2	Deafness, autosomal recessive 49
MC2R	Glucocorticoid deficiency, due to ACTH unresponsiveness
MCEE	Methylmalonyl-CoA epimerase deficiency
MEFV	Familial Mediterranean fever
MESD	Osteogenesis imperfecta, type XX
MITF	Waardenburg syndrome, type 2A
	Deafness
MLH1	Mismatch repair cancer syndrome 1
MLYCD	Malonyl-CoA decarboxylase deficiency
MMAA	Methylmalonic aciduria, vitamin B12-responsive
MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type
MNX1	Permanent neonatal diabetes mellitus, MNX1-related
MOCS1	Molybdenum cofactor deficiency
MPI	Congenital disorder of glycosylation, type Ib
MPL	Thrombocytopenia, congenital amegakaryocytic
MRAP	Glucocorticoid deficiency 2
MSH2	Mismatch repair cancer syndrome 2
MSH6	Mismatch repair cancer syndrome 3
MTHFD1	Combined immunodeficiency and megaloblastic anaemia with or without hyperhomocysteinaemia
MTR	Homocystinuria-megaloblastic anaemia, cblG complementation type
MT-RNR1	Aminoglycoside sensitivity
MTRR	Methylmalonic aciduria and homocystinuria
MTTP	Abetalipoproteinemia
MUSK	Congenital myasthenic syndrome
MUT	Methylmalonic aciduria, mut(0) type
MVK	Mevalonic aciduria
MYD88	Immunodeficiency 68
MYH7	Cardiomyopathy, hypertrophic, 1
MYO15A	Deafness, autosomal recessive 3
MYO3A	Deafness, autosomal recessive 30
MYO6	Deafness, autosomal recessive 37

Gene	Condition
MYO7A	Deafness, autosomal recessive 2
	Usher syndrome, type 1B
MYSM1	Bone marrow failure syndrome 4
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B)
NAGS	N-acetylglutamate synthetase deficiency
NCF2	Chronic granulomatous disease
NCF4	Chronic granulomatous disease, autosomal recessive, cytochrome b-positive, type III
NEUROG3	Diarrhoea 4, malabsorptive, congenital
NFKBIA	Ectodermal dysplasia and immunodeficiency 2
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation
NIPAL4	Ichthyosis, congenital, autosomal recessive 6
NKX2-1	Choreoathetosis, hypothyroidism, and neonatal respiratory distress
NKX2-5	Atrial septal defect 7, with or without AV conduction defects
NNT	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency
NPC1	Niemann-Pick disease type C1
NPC2	Niemann-Pick disease type C2
NR0B1	Adrenal hypoplasia, congenital
NR3C2	Pseudohypoaldosteronism type I, autosomal dominant
NR5A1	Adrenocortical insufficiency
OAS1	Immunodeficiency 100 with pulmonary alveolar proteinosis and hypogammaglobulinaemia
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia
ORAI1	Immunodeficiency 9
OTC	Ornithine transcarbamylase deficiency
OTOA	Deafness, autosomal recessive 22
OTOF	Deafness, autosomal recessive 9
OTOG	Deafness, autosomal recessive 18B
OTOGL	Deafness, autosomal recessive 84B
OTULIN	Autoinflammation, panniculitis, and dermatosis syndrome
OTX2	Pituitary hormone deficiency, combined, 6
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency
P3H1	Osteogenesis imperfecta, type VIII
PAH	Phenylketonuria
PALB2	Fanconi anaemia, complementation group N
PAX3	Waardenburg syndrome, type 1
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia
PC	Pyruvate carboxylase deficiency
PCBD1	Hyperphenylalaninemia, BH4-deficient, D
PCCA	Propionic acidaemia
PCCB	Propionicacidaemia
PCDH15	Usher syndrome, type 1F
	Deafness, autosomal recessive 23

Gene	Condition
PCSK9	Hypercholesterolaemia, familial, 3
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency
PDHB	Pyruvate dehydrogenase E1-beta deficiency
PDHX	Lactic acidaemia due to PDX1 deficiency
PDP1	Pyruvate dehydrogenase phosphatase deficiency
PDX1	Pancreatic agenesis
PDZD7	Deafness, autosomal recessive 57
	Usher syndrome, type 2C, GPR98/PDZD7 digenic
PGM1	Congenital disorder of glycosylation, type It
PGM3	Immunodeficiency 23
PHEX	Hypophosphatemic rickets, X-linked dominant
PHGDH	Phosphoglycerate dehydrogenase deficiency
PHKA2	Glycogen storage disease, type IXa1 and a2
PHKB	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive
	Glycogen storage disease Ixb
PHKG2	Glycogen storage disease Ixc
PIK3CD	Immunodeficiency 14B, autosomal recessive
	Immunodeficiency 14A, autosomal dominant
PIK3R1	Agammaglobulinemia 7, autosomal recessive
	Immunodeficiency 36
PKLR	Pyruvate kinase deficiency
PLG	Plasminogen deficiency, type I
PLPBP	Epilepsy, early-onset, vitamin B6-dependent
PLS3	Bone mineral density QTL18, osteoporosis
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency
POLE	IMAGE-I syndrome
POMC	Obesity and adrenal insufficiency due to POMC deficiency
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
	Disordered steroidogenesis due to cytochrome P450 oxidoreductase
POU1F1	Pituitary hormone deficiency, combined, 1
POU3F4	Deafness, X-linked 2
PPOX	Variegate porphyria, childhood-onset
PRDX1	Methylmalonic aciduria and homocystinuria, cblC type, digenic
PRF1	Haemophagocytic lymphohistiocytosis, familial, 2
PRKAR1A	Carney complex, type 1,
PRKDC	Immunodeficiency 26, with or without neurologic abnormalities
PROP1	Pituitary hormone deficiency, combined, 2
PSTPIP1	Pyogenic sterile arthritis, pyoderma gangrenosum and acne
PTCH1	Basal cell nevus syndrome
PTF1A	Pancreatic and cerebellar agenesis
	Pancreatic agenesis 2

Gene	Condition
PTPRC	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive
PTPRQ	Deafness, autosomal recessive 84A
	Deafness, autosomal dominant 73
PTS	Hyperphenylalaninemia, BH4-deficient, A
PYGL	Glycogen storage disease VI
QDPR	Dihydropteridine reductase deficiency
RAB27A	Griscelli syndrome
RAC2	Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia
RAG1	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity
	Combined cellular and humoral immune defects with granulomas
	Omenn syndrome
	Severe combined immunodeficiency, B cell-negative
RAG2	Combined cellular and humoral immune defects with granulomas
	Omenn syndrome
	Severe combined immunodeficiency, B cell-negative
RAPSN	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency
RASGRP1	Immunodeficiency 64
RB1	Retinoblastoma
RDX	Deafness, autosomal recessive 24
REST	Susceptibility to Wilms tumor, type 6
RET	Multiple endocrine neoplasia IIB
	Multiple endocrine neoplasia IIA
RFX5	Bare lymphocyte syndrome, type II, complementation group C
	Bare lymphocyte syndrome, type II, complementation group E
RFXANK	MHC class II deficiency, complementation group B
RFXAP	Bare lymphocyte syndrome, type II, complementation group D
RMRP	Cartilage-hair hypoplasia
RNPC3	Pituitary hormone deficiency, combined or isolated, 7
RPE65	Leber congenital amaurosis 2
	Retinitis pigmentosa 20
RPL11	Diamond-Blackfan anaemia
RPL15	Diamond-Blackfan anaemia 12
RPL35A	Diamond-Blackfan anaemia 5
RPL5	Diamond-Blackfan anaemia
RPS10	Diamond-Blackfan anaemia 9
RPS17	Diamond-Blackfan anaemia
RPS19	Diamond-Blackfan anaemia
RPS24	Diamond-Blackfan anaemia
RPS26	Diamond-Blackfan anaemia
RPS7	Diamond-Blackfan anaemia 8
RUNX1	Platelet disorder, familial, with associated myeloid malignancy

Gene	Condition
RYR1	Malignant hyperthermia susceptibility 1
RYR2	Arrhythmogenic right ventricular dysplasia 2
	Ventricular tachycardia, catecholaminergic polymorphic
S1PR2	Deafness, autosomal recessive 68
SAMD9	MIRAGE syndrome
SAMD9L	Ataxia-pancytopenia syndrome
SAR1B	Chylomicron retention disease
SBDS	Shwachman-Diamond syndrome
SCNN1A	Pseudohypoaldosteronism, type I
SCNN1B	Pseudohypoaldosteronism, type I
SCNN1G	Pseudohypoaldosteronism, type I
SERPINF1	Osteogenesis imperfecta, type VI
SERPINH1	Osteogenesis imperfecta, type X
SGPL1	Nephrotic syndrome, type 14
SH2D1A	Lymphoproliferative syndrome, X-linked, 1
SI	Sucrase-isomaltase deficiency, congenital
SLC12A1	Bartter syndrome, type 1
SLC18A2	Parkinsonism-dystonia, infantile, 2
SLC18A3	Myasthenic syndrome, congenital, 21, presynaptic
SLC19A2	Thiamine-responsive megaloblastic anaemia syndrome
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)
SLC22A5	Carnitine deficiency, systemic primary
SLC25A13	Citrullinemia, type II, neonatal-onset
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome
SLC25A19	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)
SLC25A20	Carnitine-acylcarnitine translocase deficiency
SLC25A38	Anaemia, sideroblastic, 2, pyridoxine-refractory
SLC26A3	Diarrhoea 1, secretory chloride, congenital
SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct
	Pendred syndrome
SLC26A7	Congenital hypothyroidism, SLC26A7-related
SLC2A1	GLUT1 deficiency syndrome 2, childhood onset
	Susceptibility to idiopathic generalized epilepsy 12
	GLUT1 deficiency syndrome 1, infantile onset, severe
SLC30A10	Hypermanganesemia with dystonia 1
SLC34A3	Hypophosphatemic rickets with hypercalciuria
SLC35A2	Congenital disorder of glycosylation, type IIm
SLC37A4	Glycogen storage disease Ib
	Glycogen storage disease Ic
	Congenital disorder of glycosylation, type IIw
SLC39A4	Acrodermatitis enteropathica
SLC39A7	Agammaglobulinaemia 9, autosomal recessive

Gene	Condition
SLC39A8	Congenital disorder of glycosylation, type IIn
SLC46A1	Folate malabsorption, hereditary
SLC4A1	Distal renal tubular acidosis 4 with haemolytic anaemia
SLC52A2	Brown-Vialetto-Van Laere syndrome 2
SLC52A3	Brown-Vialetto-Van Laere syndrome 1
SLC5A1	Glucose/galactose malabsorption
SLC5A5	Thyroid dyshormonogenesis 1
SLC5A6	Neurodegeneration, infantile-onset, biotin-responsive
SLC5A7	Myasthenic syndrome, congenital, 20, presynaptic
SLC7A7	Lysinuric protein intolerance
SLITRK6	Deafness and myopia
SLX4	Fanconi anaemia, complementation group P
SMAD2	Loeys-Dietz syndrome 6
SMAD3	Loeys-Dietz syndrome 3
SMARCD2	Specific granule deficiency 2
SMN1	Spinal muscular atrophy type 1
SMPD1	Niemann-Pick disease, type A
	Niemann-Pick disease, type B
SNX10	Osteopetrosis, autosomal recessive 8
SP110	Hepatic veno-occlusive disease with immunodeficiency
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency
SRP54	Neutropenia, severe congenital, 8, autosomal dominant
STAR	Congenital lipoid adrenal hyperplasia
STAT1	Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive
STAT3	Autoimmune disease, multisystem, infantile-onset, 1
STIM1	Immunodeficiency 10
STK4	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations
STX11	Haemophagocytic lymphohistiocytosis, familial, 4,
STX16	Pseudohypoparathyroidism, type IB
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5
SYT2	Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive
TANGO2	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration
TAT	Tyrosinemia, type II
TBL1X	Hypothyroidism, congenital, nongoitrous, 8
TBX19	Adrenocorticotropic hormone deficiency
TCF3	Agammaglobulinaemia 8, autosomal dominant
	Agammaglobulinaemia 8B, autosomal recessive
TCIRG1	Osteopetrosis, autosomal recessive 1
TCN2	Transcobalamin II deficiency
TECTA	Deafness, autosomal recessive 21
	Deafness, autosomal dominant 8/12

Gene	Condition
TF	Atransferrinemia
TG	Thyroid dyshormonogenesis 3
TGFB2	Loeys-Dietz syndrome 4
TGFB3	Loeys-Dietz syndrome 5
TGFBR1	Loeys-Dietz syndrome 1
TGFBR2	Loeys-Dietz syndrome 2
TH	Tyrosine hydroxylase deficiency
THRA	Hypothyroidism, congenital, nongoitrous, 6
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type)
TMC1	Deafness, autosomal recessive 7
TMEM38B	Osteogenesis imperfecta, type XIV
TMIE	Deafness, autosomal recessive 6
TMPRSS3	Deafness, autosomal recessive
TNFRSF11A	Osteopetrosis, autosomal recessive 7
TP53	Li-Fraumeni syndrome
TPK1	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)
TPO	Thyroid dyshormonogenesis 2A
TPP1	Ceroid lipofuscinosis, neuronal, 2 (Batten disease)
TPRN	Deafness, autosomal recessive 79
TRHR	Hypothyroidism, congenital, nongoitrous, 7
TRIM28	Wilms tumour, TRIM28-related
TRIOBP	Deafness, autosomal recessive 28
TRMU	Liver failure, transient infantile
TRPM6	Hypomagnesemia 1, intestinal
TSHB	Hypothyroidism, congenital, nongoitrous, 4
TSHR	Hypothyroidism, congenital, nongoitrous, 1
	Hyperthyroidism, familial gestational hyperthyroidism
TTPA	Ataxia with isolated vitamin E deficiency
TUBB1	Congenital hypothyroidism, TUBB1-related
	Macrothrombocytopenia, autosomal dominant, TUBB1-related
UBE2T	Fanconi anaemia, complementation group T
UGT1A1	Crigler-Najjar syndrome, type I
UMPS	Orotic aciduria
UNC13D	Haemophagocytic lymphohistiocytosis, familial, 3
UROS	Porphyria, congenital erythropoietic
USH1C	Usher syndrome type 1
USH1G	Usher syndrome type 1
USH2A	Usher syndrome type 2
VAMP1	Myasthenic syndrome, congenital, 25
VDR	Rickets, vitamin D-resistant, type IIA
VHL	von Hippel-Lindau syndrome
VKORC1	Combined vitamin K-dependent clotting factors deficiency, type 2
VPS45	Neutropenia, severe congenital, 5, autosomal recessive

Gene	Condition
WAS	Neutropenia, severe congenital, X-linked
	Thrombocytopaenia, X-linked
	Wiskott-Aldrich syndrome
WDR1	Periodic fever, immunodeficiency, and thrombocytopenia syndrome
WDR72	Amelogenesis imperfecta, type IIA3
	Distal RTA
WHRN	Usher syndrome, type 2D
	Deafness, autosomal recessive 31
WIPF1	Wiskott-Aldrich syndrome 2
WNK1	Pseudohypoaldosteronism, type 2C
WNK4	Pseudohypoaldosteronism, type 2B
WT1	Wilms tumor, type 1
XIAP	Lymphoproliferative syndrome, X-linked, 2
XPA	Xeroderma pigmentosum, group A
XPC	Xeroderma pigmentosum, group C
ZAP70	Immunodeficiency