NEWBORN SCREENING ONTARIO
DÉPISTAGE NÉONATAL ONTARIO

INFANT Study Test Description and Gene List

Gene List Version: V1.1

Effective Date: October 17th, 2025

GLOSSARY OF TERMS

Inheritance

The mode by which a genetic condition is transmitted across generations.

AR – Autosomal Recessive

A genetic condition that occurs only when an individual inherits two altered copies of a gene, one from each parent. Any condition on this panel that is documented as only AR will only be reported as such. No heterozygotes will be reported for these conditions.

AD – Autosomal Dominant

A genetic condition that occurs when an individual inherits one altered copy of a gene. A***n affected person has a 50% chance of passing it to their children.***

XL –X-linked

A genetic condition caused by an alteration on the X chromosome. Males are more often affected because they have only one X chromosome, while females may be carriers or have milder symptoms

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INFANT Study Gene List (223 Genes)

Gene List Version 1.1

See footnotes below for more details

Gene	Condition Name	Clinical Category	Inheritance[1,2,3]
ABCC6	Generalized arterial calcification of infancy	Cardiac	AR
ABCC8	Diabetes mellitus, transient and permanent neonatal	Endocrine	AR
ABCD1	Adrenoleukodystrophy	Metabolic	XL
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type	Metabolic	AR
ACADM	Medium chain acyl-CoA dehydrogenase deficiency	Metabolic	AR
ACADVL	Very long chain acyl-CoA dehydrogenase deficiency	Metabolic	AR
ACAT1	Beta-ketothiolase deficiency	Metabolic	AR
ACVR1	Fibrodysplasia ossificans progressiva	Bone/Connective Tissue	AD
ADA	Severe Combined Immunodeficiency due to ADA Deficiency	Immunology	AR
ADAMTS13	Thrombotic thrombocytopenic purpura	Blood	AR
AGL	Cori Disease (GSD Type 2)	Metabolic	AR
AGXT	Primary hyperoxaluria type 1	Metabolic	AR
AKR1D1	Congenital bile acid synthesis defect type 2	Metabolic	AR
ALDH7A1	Pyridoxine-dependent epilepsy	Metabolic	AR
ALDOB	Hereditary fructose intolerance	Metabolic	AR
ALPL	Hypophosphatasia	Metabolic	AR
AQP2	Nephrogenic diabetes insipidus type 2	Endocrine	AD/AR
ARG1	Hyperargininemia	Metabolic	AR
ARSA	Metachromatic leukodystrophy	Metabolic	AR
ARSB	Maroteaux-Lamy syndrome (MPS type 6)	Metabolic	AR
ASL	Argininosuccinic aciduria	Metabolic	AR
ASS1	Citrullinemia type I	Metabolic	AR
ATM	Ataxia-telangiectasia	Multisystem Genetic Syndrome	AR
ATP6V0A4	Distal renal tubular acidosis type 3	Metabolic/ Hearing Loss	AR
ATP6V1B1	Distal renal tubular acidosis type 4	Metabolic/ Hearing Loss	AR
AVPR2	Nephrogenic Diabetes insipidus type 1	Endocrine	AR
BCKDHA	Maple syrup urine disease, type 1a	Metabolic	AR

BCKDHB	Maple syrup urine disease, type 1b	Metabolic	AR
BTD	Biotinidase deficiency	Metabolic	AR
BTK	Agammaglobulinemia type 1	Immunology	XL
CALM1	Long QT syndrome type 14/ catecholaminergic polymorphic ventricular tachycardia (CPVT) type 4	Cardiac	AD
CALM2	Long QT syndrome type 15	Cardiac	AD
CALM3	Long QT Syndrome type 16/ catecholaminergic polymorphic ventricular tachycardia (CPVT) type 6	Cardiac	AD
CASR	Neonatal severe primary hyperparathyroidism	Endocrine	AD/AR
CBLIF	Intrinsic factor deficiency	Blood	AR
CBS	Classic homocystinuria	Metabolic	AR
CD3D	Severe combined immunodeficiency type 19	Immunology	AR
CD3E	Severe combined immunodeficiency type 19	Immunology	AR
CD40LG	X-linked hyper IgM syndrome	Immunology	XL
CDH23	Usher syndrome, type 1D/F	Multisystem (Hearing/Vision)	
CFTR	Cystic Fibrosis	Respiratory	AR
COL4A3	Alport syndrome type 3B, autosomal recessive	Multisystem (hearing, vision, kidney)	AR
COL4A4	Alport syndrome type 2 autosomal recessive	Multisystem (hearing, vision, kidney)	AR
COL4A5	Alport syndrome Type 1, X-linked	Multisystem (hearing, vision, kidney)	XL
CPS1	Carbamoyl phosphate synthetase I deficiency disease	Metabolic	AR
CPT1A	Carnitine palmitoyl transferase 1A deficiency	Metabolic	AR
CPT2	Carnitine palmitoyltransferase II deficiency	Metabolic	AR
CTNS	Cystinosis	Metabolic	AR
CYBA	Chronic granulomatous disease	Immunology	AR
CYBB	Chronic granulomatous disease	Immunology	AR
CYP11B1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	Endocrine	AR
CYP17A1	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	Endocrine	AR
CYP27A1	Cerebrotendinous xanthomatosis	Metabolic	AR
CYP27B1	Vitamin D-dependent rickets, type I	Skeletal System	AR
DBT	Maple syrup urine disease Type 2	Metabolic	AR
DCLRE1C	Severe Combined Immunodeficiency due to DCLRE1C Deficiency	Immunology	AR
DICER1	DICER1 tumour predisposition syndrome	Cancer Predisposition Syndrome	AD
DMD	Duchenne muscular dystrophy	Musculoskeletal	XL
DOCK8	DOCK8 immunodeficiency syndrome	Immunology	AR

DOK7	Congenital myasthenic syndrome type 10	Neuromuscular	AR
DUOXA2	Thyroid dyshormonogenesis 5	Endocrine	AR
ELANE	Neutropenia, severe congenital type 1	Immunology	AD
ENPP1	Generalized arterial calcification of infancy, type 1	Cardiac	AR
ETFA	Glutaric acidemia type 2A	Metabolic	AR
ETFB	Glutaric acidemia type 2B	Metabolic	AR
ETFDH	Glutaric acidemia type 2C	Metabolic	AR
F8	Hemophilia A	Blood	XL
F9	Hemophilia B	Blood	XL
FAH	Tyrosinemia, type I	Metabolic	AR
FBP1	Fructose-1,6-bisphosphatase deficiency	Metabolic	AR
FOLR1	Neurodegeneration due to cerebral folate transport deficiency	Neuromuscular	AR
FZD4	Exudative vitreoretinopathy 1/Retinopathy of prematurity	Vision Loss	AD
G6PC1	Glycogen storage disease Type Ia (Von Gierke disease)	Genetic Syndrome	AR
G6PC3	Neutropenia, severe congenital 4/Dursun Syndrome	Immunology	AR
GAA	Glycogen storage disease Type II (Pompe)	Genetic Syndrome	AR
GALE	Galactose epimerase deficiency	Metabolic	AR
GALNS	Mucopolysaccharidosis Type IVA (Morquio Syndrome)	Genetic Syndrome	AR
GALT	Galactosemia	Metabolic	AR
GAMT	Guanidinoacetate Methyltransferase (GAMT) Deficiency	Metabolic	AR
GATA3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia	Endocrinology/Hearing Loss	AR
GATM	AGAT deficiency	Metabolic	AR
GCDH	Glutaric aciduria type I	Metabolic	AR
GCH1	GTP cyclohydrolase I deficiency	Metabolic	AR
GCK	Diabetes mellitus, permanent neonatal	Endocrine	AR
GJB2	Autosomal recessive non-syndromic hearing loss 1A	Hearing Loss	AR
GLUD1	Hyperinsulinism hyperammonemia syndrome	Metabolic	AD
GRHPR	Primary hyperoxaluria type 2	Metabolic	AR
GUSB	Mucopolysaccharidosis VII	Metabolic	AR
HADH	3-Hydroxyacyl-CoA Dehydrogenase Deficiency	Metabolic	AR
HADHA	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	Metabolic	AR
HADHB	Mitochondrial trifunctional protein deficiency	Metabolic	AR
HAX1	Neutropenia, severe congenital 3/Kostmann syndrome	Immunology	AR

HBB	Sickle Cell Disease/Beta Thalassemia	Blood	AR
HLCS	Holocarboxylase synthetase deficiency	Metabolic	AR
HMGCL	3-hydroxy-3-methylglutaric aciduria	Metabolic	AR
HRAS	Costello Syndrome	Genetic Syndrome	AD
HSD11B2	Apparent mineralocorticoid excess	Endocrine	AR
HSD3B2	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency	Endocrine	AR
HSD3B7	Congenital bile acid synthesis defect type 1	Metabolic	AR
IDS	Mucopolysaccharidosis Type II (Hunter syndrome)	Metabolic	XL
IDUA	Mucopolysaccharidosis Type I (Hurler syndrome)	Metabolic	AR
IFNAR2	Immunodeficiency 45	Immunology	AR
IGSF1	Central Congenital Hypothyroidism	Endocrine	XL
IL2RG	Severe Combined Immunodeficiency, X-linked	Immunology	XL
IL7R	Severe Combined Immunodeficiency due to IL7Ralpha Deficiency	Immunology	AR
INS	Diabetes mellitus, permanent neonatal	Endocrine	AD
IRS4	Central Congenital Hypothyroidism	Endocrine	AR
IVD	Isovaleric acidemia	Metabolic	AR
JAG1	Alagille syndrome 1	Genetic Syndrome	AD
JAK3	T-B+ SCID due to JAK3 deficiency	Immunology	AR
KCNH2	Long QT syndrome 2	Cardiac	AD/AR
KCNJ11	Hyperinsulinemic hypoglycemia, familial 2	Metabolic	AR
KCNQ1	LongQT/Jervell and Lange-Nielsen syndrome type 1	Cardiac Condition/ Hearing Loss	AD/AR
LDLR	Familial Hypercholesterolemia (HoFH) type 1	Lipid	AR
LHX3	Pituitary hormone deficiency, combined, 3	Endocrine	AR
LIG4	LIG4 syndrome	Genetic Syndrome	AR
LIPA	Wolman disease	Metabolic	AR
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	Metabolic	AR
LPL	Lipoprotein lipase deficiency	Lipid	AR
LYST	Chediak-Higashi syndrome	Genetic syndrome	AR
MC2R	Glucocorticoid deficiency, due to ACTH unresponsiveness	Endocrine	AR
MLYCD	Malonyl-CoA decarboxylase deficiency	Metabolic	AR
MMAA	Methylmalonic aciduria, vitamin B12-responsive, cblA type	Metabolic	AR
MMAB	Methylmalonic aciduria, vitamin B12-responsive, cblB type	Metabolic	AR

MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	Metabolic	AR
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	Metabolic	AR
MMUT	Methylmalonic aciduria, mut(0) type	Metabolic	AR
MPI	Congenital of glycosylation type 1b	Metabolic	AR
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type	Metabolic	AR
MTRR	Homocystinuria-megaloblastic anemia, cbl E type	Metabolic	AR
MYBPC3	DCM/HCM/Left ventricular non-compaction	Cardiac Condition	AR
MYH7	Hypertrophic cardiomyopathy Type 1	Cardiac Condition	AD/AR
MYO7A	Usher syndrome, type 1B	Hearing Loss	AR
NAGS	Hyperammonemia due to N-acetylglutamate synthase deficiency	Metabolic	AR
NCF2	Chronic granulomatous disease type 2	Immunology	AR
NDP	Norrie disease/Exudative vitreoretinopathy 2, X-linked	Vision Loss	XL
NHEJ1	Severe combined Immunodeficiency 124	Immunology	AR
NOTCH2[4]	Alagille syndrome type 2	Genetic Syndrome	AD
NR0B1	X-linked adrenal hypoplasia congenita	Endocrine	XL
OTC	Ornithine transcarbamylase deficiency	Metabolic	XL
OTOF	Non-Syndromic Hearing Loss type 9	Hearing Loss	AR
PAH	Phenylketonuria	Metabolic	AR
PAX8	Congenital hypothyroidism	Endocrine	AD
PCBD1	Tetrahydrobiopterin deficiency	Metabolic	AR
PCCA	Propionic acidemia	Metabolic	AR
PCCB	Propionic acidemia	Metabolic	AR
PCDH15	Usher syndrome, type 1F	Hearing Loss	AR
PHEX	X-Linked Hypophosphatemia	Endocrine	XL
PHKB	Glycogen storage disease IXb	Metabolic	AR
PNPO	Pyridoxal phosphate-responsive seizures	Metabolic	AR
POU1F1	Pituitary hormone deficiency, combined, 1	Endocrine	AR
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2	Immunology	AR
PROP1	Pituitary hormone deficiency, combined, 2	Endocrine	AR
PTPRC	Severe Combined Immunodeficiency 105	Immunology	AR
PTS	BH4-deficient hyperphenylalaninemia A	Metabolic	AR
PYGL	Glycogen storage disease VI	Metabolic	AR
QDPR	Dihydropteridine reductase deficiency	Metabolic	AR
RAG1	Severe Combined Immunodeficiency due to RAG1 Deficiency	Immunology	AR

RAG2	Severe Combined Immunodeficiency due to RAG2 Deficiency	Immunology	AR
RB1	Retinoblastoma	Cancer Predisposition Syndrome	AD
RET	Multiple endocrine neoplasia IIB	Cancer Predisposition Syndrome	AD
RFXANK	MHC class II deficiency 2	Immunology	AR
RPE65	Leber congenital amaurosis 2	Vision Loss	AR
RPL11	Diamond-Blackfan anemia 7	Blood	AD
RPL15	Diamond-Blackfan anemia 12	Blood	AD
RPL35A	Diamond-Blackfan anemia 5	Blood	AD
RPL5	Diamond-Blackfan anemia 6	Blood	AD
RPS10	Diamond-Blackfan anemia 9	Blood	AD
RPS17	Diamond-Blackfan anemia 4	Blood	AD
RPS19	Diamond-Blackfan anemia 1	Blood	AD
RPS24	Diamond-blackfan anemia 3	Blood	AD
RPS26	Diamond-Blackfan anemia 10	Blood	AD
RPS29	Diamond-Blackfan anemia 13	Blood	AD
RPS7	Diamond-Blackfan anemia 8	Blood	AD
RYR2	Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)	Cardiac	AD
SCN5A	Long QT syndrome 3	Cardiac	AD/AR
SCNN1A	Pseudohypoaldosteronism type IB1	Endocrine	AR
SCNN1B	Pseudohypoaldosteronism type IB2/Liddle Syndrome	Endocrine	AR
SCNN1G	Pseudohypoaldosteronism type IB3/Liddle Syndrome	Endocrine	AR
SH2D1A	Lymphoproliferative syndrome type 1	Immunology	XL
SLC22A5	Systemic primary carnitine deficiency disease	Metabolic	AR
SLC25A13	Citrin deficiency	Metabolic	AR
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	Metabolic	AR
SLC25A20	Carnitine-acylcarnitine translocase deficiency	Metabolic	AR
SLC26A3	Congenital secretory chloride diarrhea	Gastrointestinal	AR
SLC26A4	Pendred Syndrome	Genetic Syndrome	AR
SLC2A1	GLUT1 deficiency syndrome type 1/2	Metabolic	AD/AR
SLC37A4	Glycogen Storage Disease Type Ib (GSD-Ib)	Metabolic	AR
SLC39A4	Acrodermatitis enteropathica	Metabolic	AR
SLC52A2	Riboflavin transporter deficiency	Multisystem (neurologic, hearing, breathing)	AR
SLC52A3	Riboflavin transporter deficiency	Multisystem (neurologic, hearing, breathing)	AR
SLC5A5	Thyroid dyshormonogenesis 1	Endocrine	AR
SLC6A8	Cerebral creatine deficiency syndrome 1	Metabolic	XL
SLC7A7	Lysinuric protein intolerance	Metabolic	AR

SMARCB1	Rhabdoid tumour predisposition syndrome	Cancer Predisposition Syndrome	AD
SMN1	Spinal muscular atrophy	Neuromuscular	AR
SPI1	Agammaglobulinemia	Immunology	AD
SPR	Dopa responsive due to sepiapterin reductase deficiency	Metabolic	AR
STAR	Congenital lipoid adrenal hyperplasia due to STAR deficiency	Endocrine	AR
STX11	Hemophagocytic lymphohistiocytosis, familial, type 4	Immunology	AR
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease	Immunology	AR
TAFAZZIN	Barth syndrome	Metabolic	XL
TAT	Tyrosinemia, type II	Metabolic	AR
TBL1X	Central Congenital Hypothyroidism	Endocrine	XL
TCIRG1	Osteopetrosis, autosomal recessive 1	Endocrine/ Skeletal	AR
TCN2	Transcobalamin II deficiency	Metabolic	AR
TG	Thyroid dyshormonogenesis 3	Endocrine	AR
TH	TH-deficient dopa-responsive dystonia	Metabolic	AR
THRA	Hypothyroidism, congenital, nongoitrous, 6	Endocrine	AD
TPO	Thyroid dyshormonogenesis 2A	Endocrine	AR
TPP1	Ceroid lipofuscinosis, neuronal, 2	Neurology/ Neuromuscular	AR
TRHR	Central Congenital Hyothyroidism	Endocrine	AR
TSHB	Central Congenital Hyothyroidism	Endocrine	AR
TSHR	Central Congenital Hypothyroidism	Endocrine	AR
TSPAN12	Exudative vitreoretinopathy 5	Vision Loss	AD
TTPA	Ataxia with isolated vitamin E deficiency	Metabolic	AR
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3	Immunology	AR
USH1C	Usher syndrome, type 1C	Hearing Loss	AR
USH1G	Usher syndrome, type 1G	Hearing Loss	AR
VDR	Hypocalcemic vitamin D-resistant rickets	Endocrine	AR
WAS	WAS-Related severe congenital neutropenia/thrombocytopenia/Wiskott Aldrich Syndrome	Immunology	XL
WT1	Wilms tumor, type 1/WAGR Syndrome	Cancer Predisposition Syndrome	AD
ZAP70	Combined immunodeficiency due to ZAP70 deficiency	Immunology	AR

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IMPORTANT NOTES

[1] For all but one of the X-Linked (XL) conditions on this gene list, **only males will be reported because of the expected risk of disease**. The sole exception is X-Linked Hypophosphatemia (*PHEX* gene), where males and females will be reported.

[2] For conditions listed as autosomal recessive (AR), only homozygous or compound heterozygous pathogenic/likely pathogenic variants will be reported; heterozygotes will not be reported. For conditions with variable inheritance patterns, the dominant phenotype will not be included, as it is not a target of our inclusion criteria.

[3]For conditions reported as AD/AR, both heterozygous and homozygous/compound heterozygous pathogenic/likely pathogenic variants will be reported

[4] Pathogenic/likely pathogenic variants in exon 34 of the *NOTCH2* gene are excluded, as they are known to cause Hajdu-Cheney Syndrome, a condition for which no established clinical management exists in infancy.

[5] **Only the following variants in the *RET* gene are being reported:** c.2753T>C, p.Met918Thr, c.1900T>C, (p.Cys634Arg), c.1900T>A, (p.Cys634Ser), c.1900T>G, (p.Cys634Gly), c.1901G>C, (p.Cys634Ser), c.1901G>A, (p.Cys634Tyr), c.1901G>T, (p.Cys634Phe), c.1902C>G, (p.Cys634Trp), c.2647_2648delinsTT, p.Ala883Phe

TEST DESCRIPTION

Coming soon

TEST LIMITATIONS

Coming soon