Expanded Carrier Screening

LifeLabs GENETICS®

Disorder	Gene
17-beta hydroxysteroid dehydrogenase 3 deficiency	HSD17B3
2-methyl-3-hydroxybutyric aciduria	HSD17B10
3-hydroxy-3-methylglutarayl-CoA lyase deficiency	HMGCL
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCC1-related)	MCCC1
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCC2-related)	MCCC2
ABCA3-related conditions	ABCA3
ABCA4-related conditions	ABCA4
ABCB11-related conditions	ABCB11
ABCC8-related conditions	ABCC8
Abetalipoproteinemia	MTTP
Achromatopsia	CNGB3
ACOX1-related conditions	ACOX1
Acrodermatitis enteropathica	SLC39A4
Adenosine deaminase deficiency	ADA
ADGRV1-related conditions	ADGRV1
Agenesis of the corpus callosum with peripheral neuropathy FCP	SLC12A6
AHI1-related conditions	AHI1
Aicardi-Goutieres syndrome 2	RNASEH2B
Aicardi-Goutieres syndrome 3	RNASEH2C
Aicardi-Goutieres syndrome 4	RNASEH2A
Aicardi-Goutieres syndrome 5	SAMHD1
AIPL1-related conditions	AIPL1
Aldosterone synthase deficiency	CYP11B2
ALG13-related conditions	ALG13
Alkaptonuria	HGD
Alpha-1 antitrypsin deficiency	SERPINA1
Alpha-mannosidosis	MAN2B1
Alpha-N-acetylgalactosaminidase deficiency	NAGA
Alpha-thalassemia	HBA1/ HBA2
Alpha-thalassemia X-linked intellectual disability syndrome	ATRX
Alport syndrome	COL4A3
Alport syndrome	COL4A4

Disorder	Gene
Alport syndrome	COL4A5
Alström syndrome	ALMS1
Androgen insensitivity syndrome	AR
Arginase deficiency	ARG1
Arginine:glycine amidinotransferase deficiency	GATM
Argininosuccinate lyase deficiency	ASL
ARL6-related conditions	ARL6
Aromatase deficiency	CYP19A1
ARX-related conditions	ARX
Asparagine synthetase deficiency	ASNS
Aspartylglucosaminuria	AGA
Ataxia with vitamin E deficiency	TTPA
Ataxia-telangiectasia-like disorder	MRE11
ATM-related cancers	ATM
ATP7A-related conditions	ATP7A
ATP8B1-related conditions	ATP8B1
Atransferrinemia	TF
Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia	AIRE
Autosomal recessive congenital ichthyosis	ABCA12
Autosomal recessive congenital ichthyosis	TGM1
Autosomal recessive spastic ataxia of Charlevoix-Saguenay FCP	SACS
AVPR2-related conditions	AVPR2
Bardet-Biedl syndrome	BBS10
Bardet-Biedl syndrome	BBS12
Bardet-Biedl syndrome	BBS7
Bardet-Biedl syndrome	BBS9
Barth syndrome	TAZ
Bartter syndrome type 1	SLC12A1
Bartter syndrome type 2	KCNJ1
BBS1-related conditions	BBS1
BBS2-related conditions	BBS2
BBS4-related conditions	BBS4

Disorder	Gene
BBS5-related conditions	BBS5
BCS1L-related conditions	BCS1L
Bernard-Soulier syndrome (GP9-related)	GP9
Beta-ketothiolase deficiency	ACAT1
Beta-mannosidosis	MANBA
Biopterin-deficient hyperphenylalaninemia	PCBD1
Biopterin-deficient hyperphenylalaninemia	PTS
Biopterin-deficient hyperphenylalaninemia	QDPR
Biotin-responsive basal ganglia disease	SLC19A3
Biotinidase deficiency	BTD
Bloom syndrome	BLM
BRIP1-related conditions	BRIP1
Brittle cornea syndrome	PRDM5
Brittle cornea syndrome	ZNF469
BSND-related conditions	BSND
Canavan disease	ASPA
Carbamoyl phosphate synthetase I deficiency	CPS1
Cardioencephalomyopathy	SCO2
Carnitine palmitoyltransferase I deficiency	CPT1A
Carnitine palmitoyltransferase II deficiency	CPT2
Carnitine-acylcarnitine translocase deficiency	SLC25A20
Carpenter syndrome	RAB23
Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders	RMRP
Catecholaminergic polymorphic ventricular tachycardia	CASQ2
CC2D2A-related conditions	CC2D2A
CDH23-related conditions	CDH23
CEP290-related conditions	CEP290
Cerebellar ataxia, intellectual disability, and dysequilibrium syndrome 1	VLDLR
Cerebral dysgenesis, neuropathy, ichthyosis, and keratoderma	SNAP29
Cerebrotendinous xanthomatosis	CYP27A1
CERKL-related conditions	CERKL
CFTR-related conditions FCP	CFTR
Charcot-Marie-Tooth disease type 1X	GJB1
Charcot-Marie-Tooth disease type 4D	NDRG1
Chediak-Higashi syndrome	LYST

Disorder	Gene
Childhood-onset dystonia with optic atrophy and basal ganglia abnormalities	MECR
Chorea-acanthocytosis	VPS13A
Choroideremia	CHM
Chronic granulomatous disease	CYBA
Chronic granulomatous disease	CYBB
Chronic granulomatous disease	NCF2
Citrin deficiency	SLC25A13
Citrullinemia type 1	ASS1
CLN3-related conditions	CLN3
CLRN1-related conditions	CLRN1
Cobalamin C deficiency	MMACHC
Cobalamin D deficiency	MMADHC
Cobalamin F deficiency	LMBRD1
Cobalamin X deficiency	HCFC1
Cockayne syndrome A	ERCC8
Cockayne syndrome B	ERCC6
Cohen syndrome	VPS13B
COL11A2-related conditions	COL11A2
COL17A1-related conditions	COL17A1
Combined immunodeficiency due to IKBKB deficiency	IKBKB
Combined malonic and methylmalonic aciduria	ACSF3
Combined oxidative phosphorylation deficiency 1	GFM1
Combined oxidative phosphorylation deficiency 3	TSFM
Combined pituitary hormone deficiency	LHX3
Combined pituitary hormone deficiency	POU1F1
Combined pituitary hormone deficiency	PROP1
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2
Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency	HSD3B2
Congenital adrenal insufficiency	CYP11A1
Congenital amegakaryocytic thrombocytopenia	MPL
Congenital chronic diarrhea	DGAT1
Congenital disorder of glycosylation	SLC35A3
Congenital disorder of glycosylation type Ia	PMM2
Congenital disorder of glycosylation type Ib FCP	MPI
Congenital disorder of glycosylation type Ic	ALG6

Disorder	Gene
Congenital disorder of glycosylation type Ik	ALG1
Congenital disorder of glycosylation type Iv	NGLY1
Congenital dyserythropoietic anemia type II	SEC23B
Congenital hydrocephalus-1	CCDC88C
Congenital hypothyroidism	TSHB
Congenital insensitivity to pain with anhidrosis	NTRK1
Congenital myasthenic syndrome	CHAT
Congenital myasthenic syndrome	CHRNE
Congenital nephrotic syndrome type 1	NPHS1
Congenital nephrotic syndrome type 2	NPHS2
Congenital secretory chloride diarrhea	SLC26A3
Corneal dystrophy and perceptive deafness	SLC4A11
CRB1-related conditions	CRB1
CTSC-related conditions	CTSC
CYP17A1-related conditions	CYP17A1
CYP1B1-related conditions	CYP1B1
CYP7B1-related conditions	CYP7B1
Cystinosis	CTNS
Cytochrome P450 oxidoreductase deficiency	POR
Desbuquois dysplasia type 1	CANT1
Developmental and epileptic encephalopathy	CAD
DGUOK-related conditions	DGUOK
DHDDS-related conditions	DHDDS
Dihydrolipoamide dehydrogenase deficiency	DLD
Distal renal tubular acidosis with deafness	ATP6V1B1
DMD-related conditions	DMD
DOK7-related conditions	DOK7
Donnai-Barrow syndrome	LRP2
Dubin-Johnson syndrome	ABCC2
DUOX2-related conditions	DUOX2
DYNC2H1-related conditions	DYNC2H1
DYSF-related conditions	DYSF
Dyskeratosis congenita spectrum disorders	DKC1
Dyskeratosis congenita spectrum disorders	RTEL1
Dyskeratosis congenita spectrum disorders	TERT
Dystrophic epidermolysis bullosa	COL7A1

Disorder	Gene
EDA-related conditions	EDA
Ehlers-Danlos syndrome, dermatosparaxis type	ADAMTS2
Ehlers-Danlos syndrome, kyphoscoliotic type	PLOD1
Ellis-van Creveld syndrome	EVC
Emery-Dreifuss muscular dystrophy	EMD
Epimerase deficiency galactosemia	GALE
ERCC2-related conditions	ERCC2
Ethylmalonic encephalopathy	ETHE1
Ellis-van Creveld syndrome EVC2-related conditions	EVC2
F2-related conditions, including prothrombin-related thrombophilia	F2
F5-related conditions, including Factor V Leiden thrombophilia	F5
Fabry disease	GLA
Factor IX deficiency (hemophilia B)	F9
Factor XI deficiency (Hemophilia C)	F11
Familial chylomicronemia syndrome	LPL
Familial dysautonomia	ELP1
Familial hemophagocytic lymphohistiocytosis type 2	PRF1
Familial hemophagocytic lymphohistiocytosis type 3	UNC13D
Familial hemophagocytic lymphohistiocytosis type 4	STX11
Familial hemophagocytic lymphohistiocytosis type 5	STXBP2
Familial hyperaldosteronism type I	CYP11B1
Familial hypercholesterolemia	LDLR
Familial hypercholesterolemia	LDLRAP1
Familial mediterranean fever	MEFV
Fanconi anemia type A	FANCA
Fanconi anemia type B	FANCB
Fanconi anemia type C	FANCC
Fanconi anemia type D2	FANCD2
Fanconi anemia type E	FANCE
Fanconi anemia type G	FANCG
Fanconi anemia type I	FANCI
Fanconi anemia type L	FANCL
FHL1-related conditions	FHL1

Disorder	Gene
FKBP10-related conditions	FKBP10
FMR1-related conditions including fragile X syndrome	FMR1
Foveal hypoplasia	SLC38A8
Fraser syndrome	FRAS1
Fraser syndrome	FREM2
Fraser syndrome	GRIP1
Fructose-1,6-bisphosphatase deficiency	FBP1
Fucosidosis	FUCA1
Galactokinase deficiency galactosemia	GALK1
Galactosemia	GALT
Galactosialidosis	CTSA
GBA-related conditions	GBA
GBE1-related conditions	GBE1
GCH1-related conditions	GCH1
GDF5-related conditions	GDF5
Geroderma osteodysplastica	GORAB
GHR-related conditions	GHR
Gitelman syndrome	SLC12A3
GJB2-related conditions	GJB2
GLB1-related conditions	GLB1
GLE1-related conditions	GLE1
Glucose-6-phosphate dehydrogenase (G6PD) deficiency	G6PD
Glutaric acidemia type I	GCDH
Glutaric acidemia type IIA	ETFA
Glutaric acidemia type IIB	ETFB
Glutaric acidemia type IIC	ETFDH
Glutathione synthetase deficiency	GSS
Glycine encephalopathy	AMT
Glycine encephalopathy	GLDC
Glycogen storage disease type Ia	G6PC
Glycogen storage disease type II (Pompe disease)	GAA
Glycogen storage disease type III	AGL
Glycogen storage disease type IXb	PHKB
Glycogen storage disease type IXc	PHKG2
Glycogen storage disease type V	PYGM
Glycogen storage disease type VII	PFKM

Disorder	Gene
GM3 synthase deficiency	ST3GAL5
GNE-related conditions	GNE
GNPTAB-related conditions FCP	GNPTAB
Golocarboxylase synthetase deficiency	HLCS
GP1BA-related conditions	GP1BA
Guanidinoacetate methyltransferase deficiency	GAMT
GUCY2D-related conditions	GUCY2D
Gyrate atrophy of the choroid and retina	OAT
HADHA-related conditions	HADHA
HBB-related hemoglobinopathies	HBB
Heme oxygenase 1 deficiency	HMOX1
Hemolytic anemia, CD59-mediated	CD59
Hereditary fructose intolerance	ALDOB
Hereditary hemochromatosis type 1	HFE
Hereditary hemochromatosis type 2	HAMP
Hereditary hemochromatosis type 2	HJV
Hereditary hemochromatosis type 3	TFR2
Hereditary leiomyomatosis and renal cell cancer	FH
Hermansky-Pudlak syndrome type 1	HPS1
Hermansky-Pudlak syndrome type 3	HPS3
Hermansky-Pudlak syndrome type 4	HPS4
Hermansky-Pudlak syndrome type 5	HPS5
Hermansky-Pudlak syndrome type 6	HPS6
Hermansky-Pudlak syndrome type 8	BLOC1S3
Hermansky-Pudlak syndrome type 9	BLOC1S6
HGSNAT-related conditions	HGSNAT
Homocystinuria due to cobalamin E deficiency	MTRR
Homocystinuria due to cobalamin G deficiency	MTR
Homocystinuria due to cystathionine beta-synthase deficiency	CBS
Homocystinuria due to MTHFR deficiency	MTHFR
HPRT1-related conditions	HPRT1
HSD17B4-related conditions	HSD17B4
Hydrolethalus syndrome type 1	HYLS1
Hyper-IgM immunodeficiency	CD40
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	SLC25A15
Hyperphosphatemic familial tumoral calcinosis	GALNT3

Disorder	Gene
Hypomyelinating leukodystrophy-12	VPS11
Hypophosphatasia	ALPL
Ichthyosis prematurity syndrome	SLC27A4
IGHMBP2-related conditions	IGHMBP2
Imerslund-Gräsbeck syndrome	AMN
Immunodeficiency-centromeric instability-facial anomalies syndrome 1	DNMT3B
Immunodeficiency-centromeric instability-facial anomalies syndrome 2	ZBTB24
Isolated ectopia lentis	ADAMTSL4
Isovaleric acidemia	IVD
ITGB3-related conditions	ITGB3
Johanson-Blizzard syndrome	UBR1
Joubert syndrome and related disorders	MKS1
Joubert syndrome and related disorders	RPGRIP1L
Joubert syndrome and related disorders	TMEM216
Junctional epidermolysis bullosa	LAMC2
Junctional epidermolysis bullosa with pyloric atresia	ITGA6
Junctional epidermolysis bullosa with pyloric atresia	ITGB4
KCNJ11-related conditions	KCNJ11
Krabbe disease	GALC
L1 syndrome	L1CAM
LAMA2-related muscular dystrophy	LAMA2
LAMA3-related conditions	LAMA3
LAMB3-related conditions	LAMB3
Leber congenital amaurosis 5	LCA5
Leukoencephalopathy with vanishing white matter	EIF2B1
Leukoencephalopathy with vanishing white matter	EIF2B2
Leukoencephalopathy with vanishing white matter	EIF2B3
Leukoencephalopathy with vanishing white matter	EIF2B4
Leukoencephalopathy with vanishing white matter	EIF2B5
LIG4 syndrome	LIG4
Limb-girdle muscular dystrophy	CAPN3
Limb-girdle muscular dystrophy type 2	SGCD

Disorder	Gene
Limb-girdle muscular dystrophy type 2C	SGCG
Limb-girdle muscular dystrophy type 2D	SGCA
Limb-girdle muscular dystrophy type 2E	SGCB
Lipoid congenital adrenal hyperplasia	STAR
LRAT-related conditions	LRAT
Lysinuric protein intolerance	SLC7A7
Lysosomal acid lipase deficiency	LIPA
Major histocompatibility complex class II deficiency	CIITA
Malonyl-CoA decarboxylase deficiency	MLYCD
Maple syrup urine disease type 1A	BCKDHA
Maple syrup urine disease type 1B	BCKDHB
Maple syrup urine disease type 2	DBT
MECP2-related conditions	MECP2
Medium-chain acyl-CoA dehydrogenase deficiency	ACADM
Medium/short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	HADH
MEDNIK syndrome	AP1S1
Megalencephalic leukoencephalopathy with subcortical cysts 1	MLC1
Metabolic crises with rhabdomyolysis, cardiac arrhythmias and neurodegeneration	TANGO2
Metachromatic leukodystrophy	ARSA
Methylmalonic acidemia	MCEE
Methylmalonic acidemia	MMAA
Methylmalonic acidemia	MMAB
Methylmalonic acidemia	MUT
MFSD8-related conditions	MFSD8
Microcephalic osteodysplastic primordial dwarfism type II	PCNT
Microcephaly, postnatal progressive, with seizures and brain atrophy	MED17
Mitochondrial complex I deficiency 1	NDUFS4
Mitochondrial complex I deficiency 10	NDUFAF2
Mitochondrial complex I deficiency 16	NDUFAF5
Mitochondrial complex I deficiency 19	FOXRED1
Mitochondrial complex I deficiency 20/ ACAD9 deficiency	ACAD9
Mitochondrial complex I deficiency 3	NDUFS7

Disorder	Gene
Mitochondrial complex I deficiency 4	NDUFV1
Mitochondrial complex I deficiency 9	NDUFS6
Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type FCP	LRPPRC
Mitochondrial complex IV deficiency 12	PET100
Mitochondrial complex IV deficiency 6	COX15
Mitochondrial DNA depletion syndrome-2	TK2
Mitochondrial neurogastrointestinal encephalomyopathy	TYMP
Mitochondrial trifunctional protein deficiency	HADHB
MKKS-related conditions	MKKS
Molybdenum cofactor deficiency	MOCS1
Molybdenum cofactor deficiency	MOCS2
MPV17-related conditions	MPV17
Mucolipidosis type III gamma	GNPTG
Mucolipidosis type IV	MCOLN1
Mucopolysaccharidosis type I	IDUA
Mucopolysaccharidosis type II	IDS
Mucopolysaccharidosis type IIIA	SGSH
Mucopolysaccharidosis type IIIB	NAGLU
Mucopolysaccharidosis type IIID	GNS
Mucopolysaccharidosis type IVA	GALNS
Mucopolysaccharidosis type IX	HYAL1
Mucopolysaccharidosis type VI	ARSB
Mucopolysaccharidosis type VII	GUSB
Mulibrey nanism	TRIM37
Multiple pterygium syndrome	CHRNG
Multiple sulfatase deficiency	SUMF1
Muscular dystrophy-dystroglycanopathy	FKRP
Muscular dystrophy-dystroglycanopathy	FKTN
Muscular dystrophy-dystroglycanopathy	LARGE1
Muscular dystrophy-dystroglycanopathy	POMT1
Muscular dystrophy-dystroglycanopathy	POMT2
Muscular dystrophy-dystroglycanopathy	RXYLT1
MUSK-related conditions	MUSK
MVK-related conditions	MVK
Myopathy, lactic acidosis, and sideroblastic anemia 1	PUS1

Disorder	Gene
Myotonia congenita	CLCN1
N-acetylglutamate synthase deficiency	NAGS
NBN-related cancers	NBN
Nemaline myopathy 2	NEB
Nephrogenic diabetes insipidus	AQP2
Nephronophthisis	INVS
Nephronophthisis	NPHP1
Neuronal ceroid lipofuscinosis type 1	PPT1
Neuronal ceroid lipofuscinosis type 10	CTSD
Neuronal ceroid lipofuscinosis type 2	TPP1
Neuronal ceroid lipofuscinosis type 5	CLN5
Neuronal ceroid lipofuscinosis type 6	CLN6
Neuronal ceroid lipofuscinosis type 8	CLN8
Niemann-Pick disease type C	NPC1
Niemann-Pick disease type C	NPC2
Niemann-Pick disease types A and B	SMPD1
Nonsyndromic deafness	LOXHD1
Nonsyndromic deafness	MYO15A
Nonsyndromic deafness	MYO7A
Nonsyndromic deafness	OTOA
Nonsyndromic deafness	SYNE4
Nonsyndromic deafness	TMC1
Nonsyndromic deafness	TMPRSS3
Nonsyndromic intellectual disability	CC2D1A
NR0B1-related conditions	NR0B1
NR2E3-related conditions	NR2E3
NSMCE3 deficiency	NSMCE3
OCRL-related conditions	OCRL
Oculocutaneous albinism type 2	OCA2
Oculocutaneous albinism type 3	TYRP1
Oculocutaneous albinism type 4	SLC45A2
Oculocutaneous albinism types 1A and 1B	TYR
OPA3-related conditions	OPA3
Opitz GBBB syndrome	MID1
Ornithine transcarbamylase deficiency	OTC
Osteogenesis imperfecta	BMP1
Osteogenesis imperfecta	CRTAP

Disorder	Gene
Osteogenesis imperfecta	P3H1
Osteopetrosis	TCIRG1
OSTM1 deficiency associated osteopetrosis	OSTM1
OTOF-related conditions	OTOF
Pantothenate kinase-associated neurodegeneration	PANK2
Parkinson disease 15	FBXO7
PCDH15-related conditions	PCDH15
PEX5-related conditions	PEX5
PEX7-related conditions	PEX7
PGM3-congenital disorder of glycosylation	PGM3
Phenylalanine hydroxylase deficiency	PAH
Phosphoglycerate dehydrogenase deficiency	PHGDH
PIGN-congenital disorder of glycosylation	PIGN
PJVK-related conditions	DFNB59 aka PJVK
PLA2G6-related conditions	PLA2G6
PLEKHG5-related conditions	PLEKHG5
PLP1-related conditions	PLP1
POLG-related conditions	POLG
Polycystic kidney disease	PKHD1
Polymicrogyria	ADGRG1
POMGNT1-related conditions	POMGNT1
Pontocerebellar hypoplasia	TSEN54
Pontocerebellar hypoplasia type 1B	EXOSC3
Pontocerebellar hypoplasia type 2D	SEPSECS
Pontocerebellar hypoplasia type 2E	VPS53
Pontocerebellar hypoplasia type 6	RARS2
Primary carnitine deficiency	SLC22A5
Primary ciliary dyskinesia	CCDC103
Primary ciliary dyskinesia	CCDC39
Primary ciliary dyskinesia	DNAH11
Primary ciliary dyskinesia	DNAH5
Primary ciliary dyskinesia	DNAI1
Primary ciliary dyskinesia	DNAI2
Primary hyperoxaluria type 1	AGXT
Primary hyperoxaluria type 2	GRHPR
Primary hyperoxaluria type 3	HOGA1

Disorder	Gene
Primary microcephaly	MCPH1
Progressive early-onset encepahlopathy with brain atrophy and thin corpus callosum	PEBAT/ TBCD
Progressive familial intrahepatic cholestasis 3	ABCB4
Progressive pseudorheumatoid dysplasia	WISP3
Prolidase deficiency	PEPD
Propionic acidemia	PCCA
Propionic acidemia	PCCB
PRPS1-related conditions	PRPS1
PSAP-related conditions	PSAP
Pycnodysostosis	CTSK
Pyridoxal 5′-phosphate-dependent epilepsy	PNPO
Pyridoxine-dependent epilepsy	ALDH7A1
Pyruvate carboxylase deficiency	PC
Pyruvate dehydrogenase complex deficiency	PDHA1
Pyruvate dehydrogenase complex deficiency	PDHB
RAPSN-related conditions	RAPSN
RDH12-related conditions	RDH12
Refsum disease	PHYH
Retinitis pigmentosa 2	RP2
Retinitis pigmentosa 25	EYS
Retinitis pigmentosa 28	FAM161A
Retinitis pigmentosa 36	PRCD
Retinitis pigmentosa 62	MAK
Rhizomelic chondrodysplasia punctata type 2	GNPAT
Rhizomelic chondrodysplasia punctata type 3	AGPS
RLBP1-related conditions	RLBP1
Roberts syndrome	ESCO2
RPE65-related conditions	RPE65
RYR1-related conditions	RYR1
SAMD9-related conditions	SAMD9
Sandhoff disease	HEXB
Schimke immuno-osseous dysplasia	SMARCAL1
Seckel syndrome	CEP152
Sepiapterin reductase deficiency	SPR
Severe combined immunodeficiency due to CD3-delta deficiency	CD3D

Disorder	Gene
Severe combined immunodeficiency due to CD3-epsilon deficiency	CD3E
Severe combined immunodeficiency due to CD45 deficiency	PTPRC
Severe combined immunodeficiency due to DCLRE1C (Artemis) deficiency	DCLRE1C
Severe combined immunodeficiency due to FOXN1 deficiency	FOXN1
Severe combined immunodeficiency due to IL7R-alpha deficiency	IL7R
Severe combined immunodeficiency due to JAK3 deficiency	JAK3
Severe combined immunodeficiency due to RAG1 deficiency	RAG1
Severe combined immunodeficiency due to RAG2 deficiency	RAG2
Severe congenital neutropenia due to G6PC3 deficiency	G6PC3
Severe congenital neutropenia due to HAX1 deficiency	HAX1
Severe congenital neutropenia type 5	VPS45
Sialic acid storage diseases	SLC17A5
Sialidosis	NEU1
Sjögren-Larsson syndrome	ALDH3A2
SLC26A2-related conditions	SLC26A2
SLC26A4-related conditions	SLC26A4
SLC37A4-related conditions	SLC37A4
Smith-Lemli-Opitz syndrome	DHCR7
Spastic paraplegia type 15	ZFYVE26
Spastic paraplegia type 49	TECPR2
Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	SLC1A4
SPG11-related conditions	SPG11
Spinal muscular atrophy	SMN1
Spinocerebellar ataxia	ANO10
Spondylocostal dysostosis	DLL3
Spondylocostal dysostosis	MESP2
Steel syndrome	COL27A1
Steroid 5-alpha-reductase deficiency	SRD5A2
Stüve-Wiedemann syndrome	LIFR
Sulfite oxidase deficiency	SUOX
SURF1-related conditions	SURF1

Disorder	Gene
Tay-Sachs disease FCP	HEXA
TBCE-related conditions	TBCE
Thiamine-responsive megaloblastic anemia	SLC19A2
Thyroid dyshormonogenesis	SLC5A5
Thyroid dyshormonogenesis	TG
Thyroid dyshormonogenesis	TPO
TMEM67-related conditions	TMEM67
Transcobalamin II deficiency	TCN2
Transient infantile liver failure	TRMU
TREX1-related conditions	TREX1
Trichohepatoenteric syndrome	SKIV2L
Trichohepatoenteric syndrome	TTC37
TRIM32-related conditions	TRIM32
Trimethylaminuria	FMO3
Triple A syndrome	AAAS
TSHR-related conditions	TSHR
TULP1-related conditions	TULP1
Tyrosine hydroxylase deficiency	TH
Tyrosinemia type I FCP	FAH
Tyrosinemia type II	TAT
Tyrosinemia type III	HPD
USH1C-related conditions	USH1C
USH2A-related conditions	USH2A
Very long-chain acyl-CoA dehydrogenase deficiency	ACADVL
Vici syndrome	EPG5
Vitamin D-dependent rickets type 1A	CYP27B1
Vitamin D-dependent rickets type 2A	VDR
VRK1-related conditions	VRK1
VSX2-related conditions	VSX2
Warsaw syndrome	DDX11
WAS-related conditions	WAS
Werner syndrome	WRN
Wilson disease	ATP7B
WNT10A-related conditions	WNT10A
Wolcott-Rallison syndrome	EIF2AK3
Woodhouse-Sakati syndrome	DCAF17

Disorder	Gene
X-linked adrenoleukodystrophy	ABCD1
X-linked agammaglobulinemia	BTK
X-linked chondrodysplasia punctata type 1	ARSE
X-linked creatine transporter deficiency	SLC6A8
X-linked hyper-IgM immunodeficiency	CD40LG
X-linked juvenile retinoschisis	RS1
X-linked myotubular myopathy	MTM1
X-linked severe combined immunodeficiency	IL2RG
Xeroderma pigmentosum complementation group A	XPA
Xeroderma pigmentosum complementation group C	XPC
Xeroderma pigmentosum, variant type	POLH
Zellweger spectrum disorder	PEX1
Zellweger spectrum disorder	PEX10
Zellweger spectrum disorder	PEX12
Zellweger spectrum disorder	PEX13
Zellweger spectrum disorder	PEX16
Zellweger spectrum disorder	PEX2
Zellweger spectrum disorder	PEX26
Zellweger spectrum disorder	PEX6

FRENCH CANADIAN PANELS
Genes on the French Canadian panel are also included in the Expanded panel

Disorder		Gene
Agenesis of the corpus callosum with peripheral neuropathy	FCP	SLC12A6
Autosomal recessive spastic ataxia of Charlevoix-Saguenay	FCP	SACS
CFTR-related conditions	FCP	CFTR
Congenital disorder of glycosylation type Ib	FCP	MPI
GNPTAB-related conditions	FCP	GNPTAB
Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type	FCP	LRPPRC
Tay-Sachs disease	FCP	HEXA
Tyrosinemia type I	FCP	FAH

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