

CORE	Disorder	Gene
	Cystic fibrosis and other CFTR-related disorders	CFTR
	Fragile X syndrome*	FMR1
	Spinal muscular atrophy	SMN1

Disorder	Gene
Alpha-thalassemia	HBA1/HBA2
Bloom syndrome	BLM
Canavan disease	ASPA
Citrullinemia type 1	ASS1
Congenital disorder of glycosylation (PMM2-related)	PMM2
Dihydroliipoamide dehydrogenase deficiency (DLD)	DLD
DMD-related dystrophinopathy* (including Duchenne/Becker muscular dystrophy and dilated cardiomyopathy)	DMD
Familial dysautonomia	ELP1
Familial hyperinsulinism (ABCC8-related)	ABCC8
Fanconi anemia type C	FANCC
FKTN-related disorders (including Walker-Warburg syndrome)	FKTN
Galactosemia (GALT-related)	GALT
Gaucher disease	GBA
GJB2-related DFNB1 nonsyndromic hearing loss and deafness	GJB2
Glycogen storage disease type Ia	G6PC
Glycogen storage disease type II (Pompe disease)	GAA
HBB-related hemoglobinopathies (including beta-thalassemia and sickle cell disease)	HBB
Krabbe disease	GALC
Maple syrup urine disease (MSUD) type 1A	BCKDHA
Maple syrup urine disease (MSUD) type 1B	BCKDHB
Medium chain acyl-CoA dehydrogenase (MCAD) deficiency	ACADM
Mucopolidosis type IV	MCOLN1

Disorder	Gene
Mucopolysaccharidosis type I (including Hurler, Hurler-Scheie, and Scheie syndromes)	IDUA
Nemaline myopathy 2	NEB
Neuronal ceroid-lipofuscinosis (CLN3-related)	CLN3
Niemann-Pick disease type A/B	SMPD1
Ornithine transcarbamylase (OTC) deficiency*	OTC
Pendred syndrome	SLC26A4
Phenylalanine hydroxylase deficiency (including phenylketonuria (PKU))	PAH
Polycystic kidney disease (PKHD1-related)	PKHD1
Rhizomelic chondrodysplasia punctata type 1/Refsum disease (PEX7-related)	PEX7
Smith-Lemli-Opitz syndrome	DHCR7
Tay-Sachs disease/hexosaminidase A deficiency	HEXA
TMEM216-related disorders (including Joubert syndrome 2 and Meckel syndrome 2)	TMEM216
Tyrosinemia type I	FAH
Usher syndrome type IF/PCDH15-related disorders	PCDH15
Usher syndrome type IIA/USH2A-related disorders	USH2A
Usher syndrome type IIIA	CLRN1
X-linked adrenoleukodystrophy*	ABCD1
X-linked severe combined immunodeficiency (X-SCID)*	IL2RG
Zellweger spectrum disorder (PEX1-related)	PEX1
Zellweger spectrum disorder (PEX6-related)	PEX6

Disorder	Gene
3-beta-hydroxysteroid dehydrogenase type II deficiency (congenital adrenal hyperplasia)	HSD3B2
3-hydroxy-3-methylglutaryl-CoA (HMG-CoA) lyase deficiency	HMGCL
3-methylglutaconic aciduria type III (Costeff optic atrophy)	OPA3
11-beta-hydroxylase-deficient congenital adrenal hyperplasia	CYP11B1
17-alpha-hydroxylase-deficient congenital adrenal hyperplasia	CYP17A1
Abetalipoproteinemia	MTTP
ACAD9 deficiency	ACAD9
Achromatopsia (CNGB3-related)	CNGB3
Acrodermatitis enteropathica	SLC39A4
Adenosine deaminase deficiency	ADA

Disorder	Gene
Aicardi-Goutieres syndrome (SAMHD1-related)	SAMHD1
Aldosterone synthase deficiency	CYP11B2
Alpha-mannosidosis	MAN2B1
Alpha-thalassemia X-linked intellectual disability syndrome*	ATRX
Alport syndrome (COL4A3-related)	COL4A3
Alport syndrome (COL4A4-related)	COL4A4
Alport syndrome, X-linked (COL4A5-related)*	COL4A5
Alström syndrome	ALMS1
Andermann syndrome	SLC12A6
Arginase deficiency	ARG1

\*Indicates disorder with X-linked inheritance.

Disorder	Gene	Disorder	Gene
Argininosuccinic aciduria	ASL	Congenital amegakaryocytic thrombocytopenia	MPL
Aromatase deficiency	CYP19A1	Congenital disorder of glycosylation (ALG6-related)	ALG6
Asparagine synthetase deficiency	ASNS	Congenital disorder of glycosylation (MPI-related)	MPI
Aspartylglucosaminuria	AGA	Congenital ichthyosis (TGM1-related)	TGM1
Ataxia with vitamin E deficiency	TTPA	Congenital insensitivity to pain with anhidrosis	NTRK1
Ataxia-telangiectasia	ATM	Congenital myasthenic syndrome (CHRNE-related)	CHRNE
Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia	AIRE	Congenital myasthenic syndrome (RAPSN-related)	RAPSN
Autosomal recessive deafness 77 (DFNB77)	LOXHD1	Congenital neutropenia (HAX1-related)	HAX1
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)	SACS	Corneal dystrophy and perceptive deafness	SLC4A11
Bardet-Biedl syndrome (BBS1-related)	BBS1	Aldosterone synthase deficiency	CYP11B2
Bardet-Biedl syndrome (BBS2-related)	BBS2	Cystinosis	CTNS
Bardet-Biedl syndrome (BBS10-related)	BBS10	D-bifunctional protein deficiency	HSD17B4
Bardet-Biedl syndrome (BBS12-related)	BBS12	DHDDS-related disorders (including congenital disorder of glycosylation/retinitis pigmentosa 59)	DHDDS
Bartter syndrome type IV (BSND-related)	BSND	Dystrophic epidermolysis bullosa (COL7A1-related)	COL7A1
Beta-ketothiolase deficiency	ACAT1	Ehlers-Danlos syndrome, dermatosparaxis type	ADAMTS2
Carbamoylphosphate synthetase I deficiency	CPS1	Ellis-van Creveld syndrome (EVC-related)	EVC
Carnitine palmitoyltransferase I deficiency	CPT1A	Ellis-van Creveld syndrome (EVC2-related)	EVC2
Carnitine palmitoyltransferase II deficiency	CPT2	Emery-Dreifuss muscular dystrophy (EMD-related)*	EMD
Carpenter syndrome (RAB23-related)	RAB23	Enhanced S-cone syndrome/retinitis pigmentosa 37	NR2E3
Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders	RMRP	Ethylmalonic encephalopathy	ETHE1
Cerebrotendinous xanthomatosis	CYP27A1	Fabry disease*	GLA
Charcot-Marie-Tooth disease (NDRG1-related)	NDRG1	Factor IX deficiency (hemophilia B)*	F9
Charcot-Marie-Tooth disease, X-linked (GJB1-related)*	GJB1	Familial hypercholesterolemia (LDLR-related)	LDLR
Chorea-acanthocytosis	VPS13A	Familial hypercholesterolemia (LDLRAP1-related)	LDLRAP1
Choroideremia*	CHM	Familial hyperinsulinism (KCNJ11-related)	KCNJ11
Chronic granulomatous disease (CYBA-related)	CYBA	Fanconi anemia type A	FANCA
Chronic granulomatous disease (CYBB-related)*	CYBB	Fanconi anemia type G	FANCG
Citrin deficiency	SLC25A13	FKRP-related disorders (including Walker-Warburg syndrome)	FKRP
Cockayne syndrome type A	ERCC8	Fumarate hydratase deficiency	FH
Cockayne syndrome type B	ERCC6	Galactokinase deficiency galactosemia	GALK1
Cohen syndrome	VPS13B	Gitelman syndrome (SLC12A3-related)	SLC12A3
Combined malonic and methylmalonic aciduria (ACSF3-related)	ACSF3	Glutaric acidemia type I	GCDH
Combined oxidative phosphorylation deficiency (GFM1-related)	GFM1	Glutaric acidemia type II (ETFA-related)	ETFA
Combined oxidative phosphorylation deficiency (TSFM-related)	TSFM	Glutaric acidemia type II (ETFDH-related)	ETFDH
Combined pituitary hormone deficiency (LHX3-related)	LHX3	Glycine encephalopathy (AMT-related)	AMT
Combined pituitary hormone deficiency (PROPI-related)	PROPI	Glycine encephalopathy (GLDC-related)	GLDC
Combined SAP deficiency	PSAP	Glycogen storage disease type Ib	SLC37A4

COMPREHENSIVE CARRIER SCREEN (CONTINUED)

\*Indicates disorder with X-linked inheritance.

COMPREHENSIVE CARRIER SCREEN (CONTINUED)	Disorder	Gene	Disorder	Gene
		Glycogen storage disease type III	AGL	Limb-girdle muscular dystrophy type 2E
	Glycogen storage disease type IV/adult polyglucosan body disease	GBE1	Lipoid congenital adrenal hyperplasia (STAR-related)	STAR
	Glycogen storage disease type V	PYGM	Lipoprotein lipase deficiency	LPL
	Glycogen storage disease type VII	PFKM	Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	HADHA
	GRACILE syndrome/BCS1L-related disorders (including mitochondrial complex III deficiency, Bjornstad syndrome, Leigh syndrome)	BCS1L	Lysinuric protein intolerance	SLC7A7
	Guanidinoacetate methyltransferase deficiency	GAMT	Lysosomal acid lipase deficiency (includes Wolman disease and cholesterol ester storage disease)	LIPA
	Hereditary fructose intolerance	ALDOB	Major histocompatibility complex class II deficiency (CIITA-related)	CIITA
	Hereditary hemochromatosis (HJV-related)	HJV	Maple syrup urine disease (MSUD) type 2	DBT
	Hereditary hemochromatosis (TFR2-related)	TFR2	Megalencephalic leukoencephalopathy with subcortical cysts type 1	MLC1
	Hermansky-Pudlak syndrome (HPS1-related)	HPS1	Menkes disease/ATP7A-related disorders* (including occipital horn syndrome and distal hereditary motor neuropathy)	ATP7A
	Hermansky-Pudlak syndrome (HPS3-related)	HPS3	Metachromatic leukodystrophy (ARSA-related)	ARSA
	Holocarboxylase synthetase deficiency	HLCS	Methylmalonic acidemia (MMAA-related)	MMAA
	Homocystinuria (CBS-related)	CBS	Methylmalonic acidemia (MMAB-related)	MMAB
	Homocystinuria due to MTHFR deficiency	MTHFR	Methylmalonic acidemia (MUT-related)	MUT
	Homocystinuria, cobalamin E type	MTRR	Methylmalonic acidemia with homocystinuria, cobalamin C type	MMACHC
	Hydrolethalus syndrome type 1	HYLS1	Methylmalonic acidemia with homocystinuria, cobalamin D type	MMADHC
	Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome	SLC25A15	Microphthalmia/clinical anophthalmia (VSX2-related)	VSX2
	Hypohidrotic ectodermal dysplasia (EDA-related)*	EDA	Mitochondrial complex I deficiency/Leigh syndrome (NDUFAF5-related)	NDUFAF5
	Hypophosphatasia	ALPL	Mitochondrial complex I deficiency/Leigh syndrome (NDUFS6-related)	NDUFS6
	Inclusion body myopathy 2	GNE	Mitochondrial DNA depletion syndrome (MPV17-related)	MPV17
	Isovaleric acidemia	IVD	Mitochondrial myopathy and sideroblastic anemia 1	PUS1
	Junctional epidermolysis bullosa (LAMA3-related)	LAMA3	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	TYMP
	Junctional epidermolysis bullosa (LAMB3-related)	LAMB3	MKS1-related disorders	MKS1
	Junctional epidermolysis bullosa (LAMC2-related)	LAMC2	Mucopolidosis type II/III (GNPTAB-related)	GNPTAB
	LAMA2-related muscular dystrophy	LAMA2	Mucopolidosis type III (GNPTG-related)	GNPTG
	Leber congenital amaurosis 2	RPE65	Mucopolysaccharidosis type II (Hunter syndrome)*	IDS
	Leber congenital amaurosis 5	LCA5	Mucopolysaccharidosis type IIIA (Sanfilippo A syndrome)	SGSH
	Leber congenital amaurosis 8/CRB1-related disorders	CRB1	Mucopolysaccharidosis type IIIB	NAGLU
	Leber congenital amaurosis 10/CEP290-related disorders	CEP290	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome)/retinitis pigmentosa 73	HGSNAT
	Leber congenital amaurosis 13	RDH12	Mucopolysaccharidosis type IIID (Sanfilippo syndrome)	GNS
	Leigh syndrome, French Canadian type	LRPPRC	Mucopolysaccharidosis type IVB (Morquio B syndrome)/GM1 gangliosidosis	GLB1
	Lethal congenital contracture syndrome 1/lethal arthrogyriposis with anterior horn cell disease	GLE1	Mucopolysaccharidosis type IX	HYAL1
	Leukoencephalopathy with vanishing white matter (EIF2B5-related)	EIF2B5	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	ARSB
	Limb-girdle muscular dystrophy type 2A (calpainopathy)	CAPN3	Multiple sulfatase deficiency	SUMF1
	Limb-girdle muscular dystrophy type 2B (dysferlinopathy)	DYSF		
	Limb-girdle muscular dystrophy type 2C	SGCG		
	Limb-girdle muscular dystrophy type 2D	SGCA		

\*Indicates disorder with X-linked inheritance.

Disorder	Gene	Disorder	Gene
N-acetylglutamate synthase deficiency	NAGS	Renal tubular acidosis with deafness (ATP6V1B1-related)	ATP6V1B1
Nephrogenic diabetes insipidus (AQP2-related)	AQP2	Retinitis pigmentosa 25	EYS
Nephrotic syndrome/congenital Finnish nephrosis (NPHS1-related)	NPHS1	Retinitis pigmentosa 26	CERKL
Nephrotic syndrome/steroid-resistant nephrotic syndrome (NPHS2-related)	NPHS2	Retinitis pigmentosa 28	FAM161A
Neuronal ceroid-lipofuscinosis (MFSD8-related)	MFSD8	Rhizomelic chondrodysplasia punctata type 3	AGPS
Neuronal ceroid-lipofuscinosis (PPT1-related)	PPT1	Roberts syndrome	ESCO2
Neuronal ceroid-lipofuscinosis (TPP1-related)	TPP1	RPGRIP1L-related disorders (including Joubert syndrome 7, COACH syndrome and Meckel syndrome 5)	RPGRIP1L
Neuronal ceroid-lipofuscinosis (CLN5-related)	CLN5	RTEL1-related disorders (including dyskeratosis congenita)	RTEL1
Neuronal ceroid-lipofuscinosis (CLN6-related)	CLN6	Sandhoff disease	HEXB
Neuronal ceroid-lipofuscinosis/Northern epilepsy (CLN8-related)	CLN8	Schimke immuno-osseous dysplasia	SMARCAL1
Niemann-Pick disease type C (NPC1-related)	NPC1	Severe combined immune deficiency (DCLRE1C-related)	DCLRE1C
Niemann-Pick disease type C (NPC2-related)	NPC2	Severe combined immunodeficiency/Omenn syndrome (RAG2-related)	RAG2
Nijmegen breakage syndrome	NBN	Severe congenital neutropenia (VPS45-related)	VPS45
Ornithine aminotransferase deficiency	OAT	Sialic acid storage disorders	SLC17A5
Osteopetrosis (TCIRG1-related)	TCIRG1	Sjögren-Larsson syndrome	ALDH3A2
Peroxisomal acyl-CoA oxidase deficiency	ACOX1	SLC26A2-related disorders (including diatrophic dysplasia, atelosteogenesis type 2, achondrogenesis type 1B/multiple metaphyseal dysplasia)	SLC26A2
Phosphoglycerate dehydrogenase deficiency/Neu-Laxova syndrome	PHGDH	SLC35A3-related disorder	SLC35A3
Polymicrogyria (ADGRG1-related)	ADGRG1	Spastic paraplegia type 15	ZFYVE26
POMGNT1-related disorders (including muscle eye brain disease)	POMGNT1	Spastic paraplegia type 49	TECPR2
Pontocerebellar hypoplasia (RARS2-related)	RARS2	Spondylothoracic dysostosis	MESP2
Pontocerebellar hypoplasia (SEPSECS-related)	SEPSECS	Steel syndrome	COL27A1
Pontocerebellar hypoplasia (VRK1-related)	VRK1	Stüve-Wiedemann syndrome	LIFR
Postnatal progressive microcephaly with seizures and brain atrophy/infantile cerebral and cerebellar atrophy (MED17-related)	MED17	Tetrahydrobiopterin deficiency (PTS-related)	PTS
Primary carnitine deficiency	SLC22A5	Transient infantile liver failure (TRMU-related)	TRMU
Primary ciliary dyskinesia (DNAH5-related)	DNAH5	Tyrosine hydroxylase deficiency	TH
Primary ciliary dyskinesia (DNAI1-related)	DNAI1	Tyrosinemia type II	TAT
Primary ciliary dyskinesia (DNAI2-related)	DNAI2	Usher syndrome type IB/MYO7A-related disorders	MYO7A
Primary hyperoxaluria type 1	AGXT	Usher syndrome type IC/USH1C-related disorders	USH1C
Primary hyperoxaluria type 2	GRHPR	Usher syndrome type ID	CDH23
Primary hyperoxaluria type 3	HOGA1	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	ACADVL
Progressive familial intrahepatic cholestasis type 2	ABCB11	Wilson disease	ATP7B
Propionic acidemia (PCCA-related)	PCCA	WNT10A-related disorders (including odonto-onycho-dermal dysplasia and Schopf-Schulz-Passarge syndrome)	WNT10A
Propionic acidemia (PCCB-related)	PCCB	X-linked creatine transporter deficiency*	SLC6A8
PRPS1-related disorders* (including Charcot-Marie-Tooth disease type 5 and Arts syndrome)	PRPS1	X-linked juvenile retinoschisis*	RS1
Pycnodysostosis	CTSK	X-linked myotubular myopathy*	MTM1
Pyruvate carboxylase deficiency	PC	Xeroderma pigmentosum complementation group A	XPA
Pyruvate dehydrogenase deficiency (PDHA1-related)*	PDHA1		
Pyruvate dehydrogenase deficiency (PDHB-related)	PDHB		

COMPREHENSIVE CARRIER SCREEN (CONTINUED)

\*Indicates disorder with X-linked inheritance.

COMPREHENSIVE

Disorder	Gene
Xeroderma pigmentosum complementation group C	XPC
Zellweger spectrum disorder (PEX2-related)	PEX2
Zellweger spectrum disorder (PEX10-related)	PEX10
Zellweger spectrum disorder (PEX12-related)	PEX12

ADD-ON GENES (CAN BE ADDED TO ANY PANEL)

Disorder	Gene
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCCC1-related)	MCCC1
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCCC2-related)	MCCC2
Alkaptonuria	HGD
Alpha-1 antitrypsin deficiency	SERPINA1
Bernard-Soulier syndrome (GP1BA-related)	GP1BA
Bernard-Soulier syndrome (GP9-related)	GP9
Biotinidase deficiency	BTD
Factor V Leiden thrombophilia	F5
Factor XI deficiency (hemophilia C)	F11
Familial Mediterranean fever	MEFV
Glucose-6-phosphate dehydrogenase (G6PD) deficiency*	G6PD
Hereditary hemochromatosis (HFE-related)	HFE
Prothrombin-related thrombophilia	F2

\*Indicates disorder with X-linked inheritance.