SecureCari®

Expanded Carrier Screening 129-Gene Panel

Screens for 11,280 pathogenic/ likely pathogenic variants in 129 genes associated with 155 X-linked and autosomal recessive disorders

Metabolic and Endocrine Disorders (76)

Disorders	Genes	Inheritance
Smith-Lemli-Opitz syndrome	*DHCR7*	AR
Tay-Sachs disease	*HEXA*	AR
Phenylketonuria	*PAH*	AR
Wilson disease	*ATP7B*	AR
Acyl-CoA dehydrogenase, medium chain, deficiency of	*ACADM*	AR
Schindler disease, type I	*NAGA*	AR
Schindler disease, type III	*NAGA*	AR
Methylmalonic aciduria, mut(0) type	*MMUT*	AR
Maple syrup urine disease, type Ia	*BCKDHA*	AR
Maple syrup urine disease, type Ib	*BCKDHB*	AR
Maple syrup urine disease, type II	*DBT*	AR
Diabetes mellitus, permanent neonatal 3, with or without neurologic features	*ABCC8*	AR
Niemann-Pick disease, type A	*SMPD1*	AR
Niemann-Pick disease, type B	*SMPD1*	AR
Niemann-Pick disease, type C1	*NPC1*	AR
Glycogen storage disease Ia	*G6PC1*	AR
Glycogen storage disease II	*GAA*	AR
Glycogen storage disease IV	*GBE1*	AR
Fructose intolerance, hereditary	*ALDOB*	AR
CPT II deficiency, infantile	*CPT2*	AR
CPT II deficiency, lethal neonatal	*CPT2*	AR
Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine- ponsive basal ganglia disease type)	*SLC19A3*	AR
Adrenal insufficiency, congenital, with 46,XY sex reversal, partial or complete	*CYP11A1*	AR
Atransferrinemia	*TF*	AR
Trimethylaminuria	*FMO3*	AR
Mitochondrial complex IV deficiency, nuclear type 2	*SCO2*	AR
Galactosemia	*GALT*	AR
Methylmalonic aciduria and homocystinuria, cblC type	*MMACHC*	AR
Tyrosinemia, type I	*FAH*	AR
Cerebrotendinous xanthomatosis	*CYP27A1*	AR
Mucolipidosis IV	*MCOLN1*	AR
Mucolipidosis II alpha/beta	*GNPTAB*	AR
Mucolipidosis III alpha/beta	*GNPTAB*	AR
Glycogen storage disease Ib	*SLC37A4*	AR
Glycogen storage disease Ic	*SLC37A4*	AR
Aspartylglucosaminuria	*AGA*	AR
Hyperoxaluria, primary, type I	*AGXT*	AR
Hyper-IgD syndrome	*MVK*	AR
Mevalonic aciduria	*MVK*	AR
3-Methylcrotonyl-CoA carboxylase 1 deficiency	*MCCC1*	AR
3-Methylcrotonyl-CoA carboxylase 2 deficiency	*MCCC2*	AR
VLCAD deficiency	*ACADVL*	AR
Mucopolysaccharidosis Ih	*IDUA*	AR
Mucopolysaccharidosis Ih/s	*IDUA*	AR
Mucopolysaccharidosis type IIIA (Sanfilippo A)	*SGSH*	AR
Mucopolysaccharidosis type IIIB (Sanfilippo B)	*NAGLU*	AR
Mucopolysaccharidosis type IVB (Morquio)	*GLB1*	AR
GM1-gangliosidosis, type I	*GLB1*	AR
GM1-gangliosidosis, type II	*GLB1*	AR
Biotinidase deficiency	*BTD*	AR
Homocystinuria, B6-responsive and nonresponsive types	*CBS*	AR
Mitochondrial DNA depletion syndrome 4A (Alpers type)	*POLG*	AR
Mitochondrial DNA depletion syndrome 4B (MNGIE type)	*POLG*	AR
Alpha-methylacetoacetic aciduria	*ACAT1*	AR
Hypophosphatasia, childhood	*ALPL*	AR
Hypophosphatasia, infantile	*ALPL*	AR
Glutaric acidemia IIC	*ETFDH*	AR
Glutaricaciduria, type I	*GCDH*	AR
Isovaleric acidemia	*IVD*	AR
Carnitine deficiency, systemic primary	*SLC22A5*	AR
LCHAD deficiency	*HADHA*	AR
Hyperphenylalaninemia, BH4-deficient, A	*PTS*	AR
Acyl-CoA dehydrogenase, short-chain, deficiency of	*ACADS*	AR
Krabbe disease	*GALC*	AR
Citrullinemia, type II, neonatal-onset	*SLC25A13*	AR
Pituitary hormone deficiency, combined, 2	*PROP1*	AR
Glycine encephalopathy 2	*AMT*	AR
Glycogen storage disease IIIa	*AGL*	AR
Glycogen storage disease IIIb	*AGL*	AR
Holocarboxylase synthetase deficiency	*HLCS*	AR
Propionic acidemia	*PCCA*	AR
	PCCB	AR
Cystinosis, nephropathic	*CTNS*	AR
Combined malonic and methylmalonic aciduria	*ACSF3*	AR
Adrenal hypoplasia, congenital	*NR0B1*	XL
Fabry disease	*GLA*	XL
Ornithine transcarbamylase deficiency	*OTC*	XL

Hematological and Immunological Disorders (12)

Disorders	Genes	Inheritance
Thalassemia, beta	*HBB*	AR
Sickle cell disease	*HBB*	AR
Fanconi anemia, complementation group A	*FANCA*	AR
Fanconi anemia, complementation group C	*FANCC*	AR
Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia	*AIRE*	AR
Hemophagocytic lymphohistiocytosis, familial, 2	*PRF1*	AR
Thalassemia, alpha-	*HBA1*	AR
	HBA2	AR
Hemophagocytic lymphohistiocytosis, familial, 3	*UNC13D*	AR
Ataxia-telangiectasia	*ATM*	AR
Hemophilia A	*F8*	XL
Hemophilia B	*F9*	XL
Severe combined immunodeficiency, X-linked	*IL2RG*	XL

Respiratory, visual, and auditory systems disorders (12)

Disorders	Genes	Inheritance
Deafness, autosomal recessive 1A	*GJB2*	AR
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	*SLC26A4*	AR
Pendred syndrome	*SLC26A4*	AR
Cystic fibrosis	*CFTR*	AR
Leber congenital amaurosis 10	*CEP290*	AR
Retinitis pigmentosa 59	*DHDDS*	AR
Retinitis pigmentosa 74	*BBS2*	AR
Surfactant metabolism dysfunction, pulmonary, 3	*ABCA3*	AR
Achromatopsia 3	*CNGB3*	AR
Retinoschisis	*RS1*	XL
Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness	*RPGR*	XL
Macular degeneration, X-linked atrophic	*RPGR*	XL

Multi-System Disorders (10)

Disorders	Genes	Inheritance
Cerebrooculofacioskeletal syndrome 2	*ERCC2*	AR
Trichothiodystrophy 1, photosensitive	*ERCC2*	AR
Fraser syndrome 3	*GRIP1*	AR
Bardet-Biedl syndrome 2	*BBS2*	AR
Meckel syndrome, type 2	*TMEM216*	AR
Donnai-Barrow syndrome	*LRP2*	AR
Bardet-Biedl syndrome 1	*BBS1*	AR
Meckel syndrome 6	*CC2D2A*	AR
Ehlers-Danlos syndrome, kyphoscoliotic type, 1	*PLOD1*	AR
Opitz GBBB syndrome	*MID1*	XL

Nervous System and Neuromuscular Disorders (30)

Disorders	Genes	Inheritance
Canavan disease	*ASPA*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	*FKTN*	AR
Spinocerebellar ataxia, autosomal recessive 10	*ANO10*	AR
Joubert syndrome 3	*AHI1*	AR
Joubert syndrome 5	*CEP290*	AR
Myasthenic syndrome, congenital, 4A, slow-channel	*CHRNE*	AR
Myasthenic syndrome, congenital, 4B, fast-channel	*CHRNE*	AR
Microcephaly 1, primary, autosomal recessive	*MCPH1*	AR
Joubert syndrome 2	*TMEM216*	AR
Hydrocephalus, congenital, 1	*CCDC88C*	AR
Myotonia congenita, recessive	*CLCN1*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5	*FKRP*	AR
Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5	*FKRP*	AR
Aicardi-Goutieres syndrome 2	*RNASEH2B*	AR
Joubert syndrome 9	*CC2D2A*	AR
Megalencephalic leukoencephalopathy with subcortical cysts 1	*MLC1*	AR
Pontocerebellar hypoplasia, type 6	*RARS2*	AR
Metachromatic leukodystrophy	*ARSA*	AR
Spinal muscular atrophy-1	*SMN1*	AR
Spinal muscular atrophy-II	*SMN1*	AR
Spinal muscular atrophy-III	*SMN1*	AR
Spinal muscular atrophy-IV	*SMN1*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 1	*CAPN3*	AR
Developmental and epileptic encephalopathy 1	*ARX*	XL
Hydrocephalus, congenital, X-linked	*L1CAM*	XL
Becker muscular dystrophy	*DMD*	XL
Duchenne muscular dystrophy	*DMD*	XL
Spastic paraplegia 2, X-linked	*PLP1*	XL
Intellectual developmental disorder, X-linked 109	*AFF2*	XL
Myopathy, centronuclear, X-linked	*MTM1*	XL

Skin Disorders (6)

Disorders	Genes	Inheritance
Albinism, oculocutaneous, type IA	*TYR*	AR
Albinism, oculocutaneous, type IB	*TYR*	AR
Xeroderma pigmentosum, group A	*XPA*	AR
Xeroderma pigmentosum, group C	*XPC*	AR
Epidermolysis bullosa dystrophica inversa	*COL7A1*	AR
Albinism, oculocutaneous, type II	*OCA2*	AR

Skeletal System Disorders (5)

Disorders	Genes	Inheritance
Short-rib thoracic dysplasia type 3 with or without polydactyly	*DYNC2H1*	AR
Vitamin D-dependent rickets, type I	*CYP27B1*	AR
Ellis-van Creveld syndrome	*EVC2*	AR
Epiphyseal dysplasia, multiple, 4	*SLC26A2*	AR
Achondrogenesis Ib	*SLC26A2*	AR

Digestive and Renal System Disorders (4)

Disorders	Genes	Inheritance
Nephrotic syndrome, type 1	*NPHS1*	AR
Polycystic kidney disease 4, with or without hepatic disease	*PKHD1*	AR
Cholestasis, progressive familial intrahepatic, 1	*ATP8B1*	AR
Cholestasis, progressive familial intrahepatic, 2	*ABCB11*	AR

Note: AR: Autosomal recessive; XL: X-linked