

Expanded Carrier Screening 51-Gene Panel

Screens for 7,343 pathogenic or likely pathogenic variants across 51 genes associated with selected X-linked and autosomal recessive disorders

Disorders	Genes	Inheritance
Deafness, autosomal recessive 1A	<i>GJB2</i>	AR/DD
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct; Pendred syndrome	<i>SLC26A4</i>	AR
Usher syndrome, type 2A	<i>USH2A</i>	AR
Spinal muscular atrophy	<i>SMN1</i>	AR
Duchenne muscular dystrophy; Becker muscular dystrophy	<i>DMD</i>	XLR
Muscular dystrophy, limb-girdle, autosomal recessive 1	<i>CAPN3</i>	AR
Thalassemias, alpha-	<i>HBA1/HBA2</i>	AR
Thalassemia, beta; Sickle cell disease	<i>HBB</i>	AR
Hemophilia A	<i>F8</i>	XLR
Hemophilia B	<i>F9</i>	XLR
Fanconi anemia, complementation group A	<i>FANCA</i>	AR
Fanconi anemia, complementation group C	<i>FANCC</i>	AR
Albinism, oculocutaneous, type IA; Albinism, oculocutaneous, type IB	<i>TYR</i>	AR
Albinism, oculocutaneous, type II	<i>OCA2</i>	AR
Phenylketonuria	<i>PAH</i>	AR
Hyperphenylalaninemia, BH4-deficient, A	<i>PTS</i>	AR
Methylmalonic aciduria and homocystinuria, cblC type	<i>MMACHC</i>	AR
Methylmalonic aciduria, mut(0) type	<i>MMUT</i>	AR
Methylmalonic aciduria, vitamin B12-responsive, cblA type	<i>MMAA</i>	AR
Methylmalonic aciduria, vitamin B12-responsive, cblB type	<i>MMAB</i>	AR
Homocystinuria-megaloblastic anemia, cblD type; Methylmalonic aciduria and homocystinuria, cblD type; Methylmalonic aciduria, cblD type	<i>MMADHC</i>	AR
Wilson disease	<i>ATP7B</i>	AR
Adrenal hypoplasia, congenital; 46XY sex reversal 2, dosage-sensitive	<i>NR0B1</i>	XL
Carnitine deficiency, systemic primary	<i>SLC22A5</i>	AR
Fabry disease	<i>GLA</i>	XL
Glycogen storage disease Ia	<i>G6PC1</i>	AR
Glycogen storage disease Ib; Glycogen storage disease Ic	<i>SLC37A4</i>	AR
Pompe disease	<i>GAA</i>	AR
Glycogen storage disease IIIa; Glycogen storage disease IIIb	<i>AGL</i>	AR
Glutaricaciduria, type I	<i>GCDH</i>	AR

Disorders	Genes	Inheritance
Citrullinemia	<i>ASS1</i>	AR
Citrullinemia, type II	<i>SLC25A13</i>	AR
Biotinidase deficiency	<i>BTD</i>	AR
Sitosterolemia 2	<i>ABCG5</i>	AR
Sitosterolemia 1	<i>ABCG8</i>	AR
Maple syrup urine disease, type Ia	<i>BCKDHA</i>	AR
Maple syrup urine disease, type Ib	<i>BCKDHB</i>	AR
Maple syrup urine disease, type II	<i>DBT</i>	AR
Mucopolysaccharidosis Ih; Mucopolysaccharidosis Ih/s; Mucopolysaccharidosis Is	<i>IDUA</i>	AR
Mucopolysaccharidosis type IIIB (Sanfilippo B)	<i>NAGLU</i>	AR
Mucopolysaccharidosis II	<i>IDS</i>	XLR
Niemann-Pick disease, type A; Niemann-Pick disease, type B	<i>SMPD1</i>	AR
Niemann-Pick disease, type C1; Niemann-Pick disease, type D	<i>NPC1</i>	AR
Propionicacidemia	<i>PCCA</i>	AR
	<i>PCCB</i>	AR
Tyrosinemia, type I	<i>FAH</i>	AR
VLCAD deficiency	<i>ACADVL</i>	AR
Cystic fibrosis	<i>CFTR</i>	AR
Alport syndrome 3B, autosomal recessive	<i>COL4A3</i>	AR
Alport syndrome 2, autosomal recessive	<i>COL4A4</i>	AR

Note: AD: Autosomal dominant; AR: Autosomal recessive; XL: X-linked; XLR: X-linked recessive; DD: Digenic dominant