SecureCari®

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	*GJB2*	AR/DD
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct; Pendred syndrome	*SLC26A4*	AR
Usher syndrome, type 2A	*USH2A*	AR
Spinal muscular atrophy	*SMN1*	AR
Duchenne muscular dystrophy; Becker muscular dystrophy	*DMD*	XLR
Muscular dystrophy, limb-girdle, autosomal recessive 1	*CAPN3*	AR
Thalassemias, alpha-	*HBA1/HBA2*	AR
Thalassemia, beta; Sickle cell disease	*HBB*	AR
Hemophilia A	*F8*	XLR
Hemophilia B	*F9*	XLR
Fanconi anemia, complementation group A	*FANCA*	AR
Fanconi anemia, complementation group C	*FANCC*	AR
Albinism, oculocutaneous, type IA; Albinism, oculocutaneous, type IB	*TYR*	AR
Albinism, oculocutaneous, type II	*OCA2*	AR
Phenylketonuria	*PAH*	AR
Hyperphenylalaninemia, BH4-deficient, A	*PTS*	AR
Methylmalonic aciduria and homocystinuria, cblC type	*MMACHC*	AR
Methylmalonic aciduria, mut(0) type	*MMUT*	AR
Methylmalonic aciduria, vitamin B12-responsive, cblA type	*MMAA*	AR
Methylmalonic aciduria, vitamin B12-responsive, cblB type	*MMAB*	AR
Homocystinuria-megaloblastic anemia, cblD type; Methylmalonic aciduria and homocystinuria, cblD type; Methylmalonic aciduria, cblD type	*MMADHC*	AR
Wilson disease	*ATP7B*	AR
Adrenal hypoplasia, congenital; 46XY sex reversal 2, dosage-sensitive	*NR0B1*	XL
Carnitine deficiency, systemic primary	*SLC22A5*	AR
Fabry disease	*GLA*	XL
Glycogen storage disease Ia	*G6PC1*	AR
Glycogen storage disease Ib; Glycogen storage disease Ic	*SLC37A4*	AR
Pompe disease	*GAA*	AR
Glycogen storage disease IIIa; Glycogen storage disease IIIb	*AGL*	AR
Glutaricaciduria, type I	*GCDH*	AR
Citrullinemia	*ASS1*	AR
Citrullinemia, type II	*SLC25A13*	AR
Biotinidase deficiency	*BTD*	AR
Sitosterolemia 2	*ABCG5*	AR
Sitosterolemia 1	*ABCG8*	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
Mucopolysaccharidosis Ih; Mucopolysaccharidosis Ih/s; Mucopolysaccharidosis Is	*IDUA*	AR
Mucopolysaccharidosis type IIIB (Sanfilippo B)	*NAGLU*	AR
Mucopolysaccharidosis II	*IDS*	XLR
Niemann-Pick disease, type A; Niemann-Pick disease, type B	*SMPD1*	AR
Niemann-Pick disease, type C1; Niemann-Pick disease, type D	*NPC1*	AR
Propionicacidemia	*PCCA*	AR
	PCCB	AR
Tyrosinemia, type I	*FAH*	AR
VLCAD deficiency	*ACADVL*	AR
Cystic fibrosis	*CFTR*	AR
Alport syndrome 3B, autosomal recessive	*COL4A3*	AR
Alport syndrome 2, autosomal recessive	*COL4A4*	AR

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

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