SecureCari®

Expanded Carrier Screening 21-Gene Panel

Screens for 4,032 pathogenic or likely pathogenic variants across 21 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
Spinal muscular atrophy	*SMN1*	AR
Duchenne muscular dystrophy;Becker muscular dystrophy	*DMD*	XLR
Thalassemias, alpha-	*HBA1/HBA2*	AR
Thalassemia, beta; Sickle cell disease	*HBB*	AR
Hemophilia A	*F8*	XLR
Hemophilia B	*F9*	XLR
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
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
Pompe disease	*GAA*	AR
Cystic fibrosis	*CFTR*	AR

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

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