NBSure™

Newborn Genetic Screening Inherited Metabolic Disorders Panel

Detects pathogenic or likely pathogenic variants in 57 genes associated with inherited metabolic diseases

Organic Acid Metabolism Disorders (21)

Disorders	Genes	Inheritance
Methylmalonic aciduria, vitamin B12-responsive, cblA type	*MMAA*	AR
Methylmalonic aciduria, vitamin B12-responsive, cblB type	*MMAB*	AR
Methylmalonic aciduria and homocystinuria, cblC type	*MMACHC*	AR
Homocystinuria-megaloblastic anemia, cblD type	*MMADHC*	AR
Methylmalonic aciduria and homocystinuria, cblD type		AR
Methylmalonic aciduria, cblD type		AR
Methylmalonic aciduria, mut(0) type	*MMUT*	AR
Propionicacidemia	*PCCA*	AR
Propionicacidemia	*PCCB*	AR
Isovaleric acidemia	*IVD*	AR
Glutaricaciduria, type I	*GCDH*	AR
Biotinidase deficiency	*BTD*	AR
Holocarboxylase synthetase deficiency	*HLCS*	AR
3-Methylcrotonyl-CoA carboxylase 1 deficiency	*MCCC1*	AR
3-Methylcrotonyl-CoA carboxylase 2 deficiency	*MCCC2*	AR
3-methylglutaconic aciduria, type I	*AUH*	AR
HMG-CoA lyase deficiency	*HMGCL*	AR
Alpha-methylacetoacetic aciduria	*ACAT1*	AR
Malonyl-CoA decarboxylase deficiency	*MLYCD*	AR
Ethylmalonic encephalopathy	*ETHE1*	AR
Isobutyryl-CoA dehydrogenase deficiency	*ACAD8*	AR

Amino Acid Metabolism Disorders (29)

Disorders	Genes	Inheritance
Phenylketonuria	*PAH*	AR
Hyperphenylalaninemia, BH4-deficient, A	*PTS*	AR
Hyperphenylalaninemia, BH4-deficient, B	*GCH1*	AR
Dystonia, DOPA-responsive		AD/ AR
Hyperphenylalaninemia, BH4-deficient, C	*QDPR*	AR
Hyperphenylalaninemia, BH4-deficient, D	*PCBD1*	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
Tyrosinemia, type I	*FAH*	AR
Tyrosinemia, type II	*TAT*	AR
Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency	*MAT1A*	AD/ AR
Methionine adenosyltransferase deficiency, autosomal recessive		AD/ AR
Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	*AHCY*	AR
Homocystinuria, B6-responsive and nonresponsive types	*CBS*	AR
Thrombosis, hyperhomocysteinemic		AR
Homocystinuria-megaloblastic anemia, cblG complementation type	*MTR*	AR
Homocystinuria-megaloblastic anemia, cbl E type	*MTRR*	AR
Homocystinuria due to MTHFR deficiency	*MTHFR*	AR
Glycine encephalopathy1	*GLDC*	AR
Glycine encephalopathy 2	*AMT*	AR
Citrullinemia	*ASS1*	AR
Citrullinemia, adult-onset type II	*SLC25A13*	AR
Citrullinemia, type II, neonatal-onset		AR
Argininemia	*ARG1*	AR
Argininosuccinic aciduria	*ASL*	AR
Carbamoylphosphate synthetase I deficiency	*CPS1*	AR
Ornithine transcarbamylase deficiency	*OTC*	XL
Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	*SLC25A15*	AR

Fatty Acid Metabolism Disorders (19)

Disorders	Genes	Inheritance
Carnitine deficiency, systemic primary	*SLC22A5*	AR
CPT deficiency, hepatic, type IA	*CPT1A*	AR
CPT II deficiency, infantile	*CPT2*	AR
CPT II deficiency, lethal neonatal		AR
CPT II deficiency, myopathic, stress-induced		AD/ AR
Carnitine-acylcarnitine translocase deficiency	*SLC25A20*	AR
Acyl-CoA dehydrogenase, medium chain, deficiency of	*ACADM*	AR
Acyl-CoA dehydrogenase, short-chain, deficiency of	*ACADS*	AR
VLCAD deficiency	*ACADVL*	AR
3-hydroxyacyl-CoA dehydrogenase deficiency	*HADH*	AR
Hyperinsulinemic hypoglycemia, familial, 4		AR
Glutaric acidemia IIA	*ETFA*	AR
Glutaric acidemia IIB	*ETFB*	AR
Glutaric acidemia IIC	*ETFDH*	AR
Mitochondrial trifunctional protein deficiency 1	*HADHA*	AR
LCHAD deficiency		AR
Fatty liver, acute, of pregnancy		AR
HELLP syndrome, maternal, of pregnancy		AR
Mitochondrial trifunctional protein deficiency 2	*HADHB*	AR

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

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