LABSOLUTIONS

1451 NORTHSIDE DRIVE NW | SUITE A | ATLANTA, GA 30318
(tel) 404.228.5027 | (fax) 404.343.0087 | cs@labsolutions.com

REVEAL™ Test Menu

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GENE	ASSOCIATED CONDITIONS
ABCD1	Adrenoleukodystrophy
ABCD4	cblJ Type Methylmalonic aciduria and homocystinuria
ACAD8	Isobutyryl-CoA dehydrogenase deficiency
ACADM	MCAD DEFICIENCY
ACADS	SCAD Deficiency
ACADSB	2-methylbutyryl-CoA Dehydrogenase Deficiency
ACADVL	VLCAD deficiency
ACAT1	Alpha-methylacetoacetic aciduria (3-ketothialase deficiency)
ACSF3	Combined malonic and methylmalonic aciduria
ADA	Partial adenosine deaminase deficiency Severe combined immunodeficiency (SCID) due to adenosine deaminase deficiency (ADAD)
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
ARG1	Argininemia (Arginase Deficiency)
ASL	Arginosuccinic Aciduria
ASPA	Canavan disease
ASS1	Citrullinemia
AUH	3-methylglutaconic aciduria type I (MCGA1)
BCKDHA	Maple syrup urine disease, type Ia
BCKDHB	Maple syrup urine disease, type Ia
BLM	Bloom syndrome
BTD	Biotinidase deficiency
CBS	Homocystinuria, B6-responsive and nonresponsive types Hyperhomocysteinemic thrombosis
CD320	Methylmalonic aciduria due to transcobalamin receptor defect
CFTR	Cogenital bilateral absence of the vas deferens (CVAD) Cystic fibrosis Neonatal hypertrypsinemia
CPT1A	Hepatic carnitine palmitoyl transferase deficiency Type I
CPT2	CPT2 deficiency associated myopathy Hepatic carnitine palmitoyl transferase deficiency Type II Lethal neonatal CPT2 deficiency Susceptibility to acute-infection induced encephalopathy
CYP21A2	Congenital Adrenal Hyperplasia due to 21-hydroxylase deficiency Non-classic hyperandrogenism due to 21-hydroxylase deficiency
DBT	Maple syrup urine disease type II
DLD	Dihydrolipoamide dehydrogenase deficiency
DNAJC19	3-methylglutaconic aciduria, type V
DUOX2	Thryoid dyshormonogenesis 6
EFTA	Glutaric acidemia IIA
ETFB	Glutaric acidemia IIB

GENE	ASSOCIATED CONDITIONS
EFTDH	Glutaric acidemia IIC
FAH	Tyrosinemia, type I
FANCC	Fanconi anemia
G6PC	Glycogen storage disease Ia
G6PD	Favism Hemolytic anemia due to G6PD deficiency
GAA	Glycogen storage disease II
GALC	Krabbe disease
GALE	Galactose epimerase deficiency
GALK1	Galactokinase deficiency with cataracts
GALT	Galactosemia
GBA	Gaucher disease Type I Gaucher disease Type II Gaucher disease Type III Gaucher disease Type IIIC Perinatal lethal Gaucher disease
GCDH	Glutaric aciduria Type I
GCH1	BH4-deficient Hyperphenylalaninemia B DOPA-responsive dystonia (with or without hyperphenylalaninemia)
GJB2	Autosomal dominant deafness Type 3A Autosomal recessive deafness Type 1A Bart-Pumphrey Syndrome Hystrix-like ichthyosis with deafness Keratitis ichthyosis deafness syndrome Palmoplantar keratoderma with deafness Vohwinkel syndrome
GJB3	Autosomal dominant deafness Type IIB Autosomal recessive deafness Digenic deafness GJB2/GJB3 Erythrokeratodermia variabilis et progressiva
GJB6	Autosomal dominant deafness Type IIIB Autosomal recessive deafness Type IB Clouston type ectodermal dysplasia Type II Digenic GJB2/GJB6 deafness
GLA	Fabry Disease
GNMT	Glycine N-methyltransferase deficiency
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency Familial hyperinsulinemic hypoglycemia type 4
HADHA	LCHAD deficiency Trifunctional protein deficiency
HADHB	Trifunctional protein deficiency
HBA1	Alpha thalassemia Hb Barts Hb H (3 gene deletion) Hb H/Constant Spring disease
HBA2	Alpha thalassemia Hb Barts Hb H (3 gene deletion) Hb H/Constant Spring disease

GENE	ASSOCIATED CONDITIONS
HBB	Beta thalassemia major Hb C disease (Hb CC) Hb D disease (Hb DD) Hb E/Beta+ thalassemia Hb C/Beta0 thalassemia Hb C/Beta+ thalassemia Hb D/Beta0 thalassemia Hb D/Beta+ thalassemia Hb E/Beta0 thalassemia Hb EE Hb Variant/ Beta0 thalassemia Hb Variant/Beta+ thalassemia Hb Variants Hb S/Beta0 thalassemia Hb S/Beta + thalassemia Hereditary persistence of fetal hemoglobin Sickle cell anemia (S/S) Sickle cell disease variants Sickle hemoglobin C disease Sickle hemoglobin D disease Sickle hemoglobin E disease
HCFC1	X-linked mental retardation with methylmalonic acidemia and homocysteinemia
HEXA	GM2-gangliosidosis Hex A pseudodeficiency Tay-Sachs disease
HLCS	Holocarboxylase synthetase deficiency
HMGCL	HMG-CoA Lyase Deficiency
HPD	Hawkinsinuria Tyrosinemia, type III
HSD17B10	17-beta-hydroxysteroid dehydrogenase X deficiency Mental retardation X-linked syndromic 10 (MRXS10)
IDUA	Mucopolysaccharidosis Ih Mucopolysaccharidosis Ih/s Mucopolysaccharidosis Is
IKBKAP	Familial dysautonomia
IL2RG	X-linked severe combined immunodeficiency (SCID)
IVD	Isovaleric acidemia
LMBRD1	Methylmalonic acidemia with homocystinuria (Gobbledegook)
MAT1A	Autosomal dominant persistent hypermethioninemia due to methionine adenosyltransferase I/III deficiency
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency (MCC1D)
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency (MCC2D)
MCEE	Methylmalonyl-CoA epimerase deficiency
MCOLN1	Mucolipidosis IV
MLYCD	Malonyl-CoA decarboxylase deficiency
MMAA	Vitamin B-12 responsive methylmalonic aciduria MMAA
MMAB	cblB complement type Vitamin B-12 responsive methylmalonic aciduria (due to defect in synthesis of adenosylcobalamin)
MMADHC	cblD complement type homocystinuria (Variant 1) cblD complement type homocystinuria (Variant 2) cblD complement type Methylmalonic aciduria and homocystinuria (Variant 1)
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type

GENE	ASSOCIATED CONDITIONS
MTHFR	Homocystinuria due to MTHFR deficiency
MTR	cbl G complementation type homocystinuria-megaloblastic anemia
MTRR	cbl E complementation type homocystinuria-megaloblastic anemia
MUT	Methylmalonic Aciduria due to Methymalonyl-CoA Mutase deficiency
NPC1	Niemann-Pick disease, type C1 Niemann-Pick disease, type D
NPC2	Niemann-pick disease, type C2
OPA3	C3-methylglutaconic aciduria, type III Optic atrophy 3 with cataract
OTC	Ornithine transcarbamylase deficiency
PAH	Non-PKU hyperphenylalanemia Phenylketonuria
PAX8	Congenital hypothyroidism due to thyroid dysgenesis or hypoplasia
PCBD1	BH4-deficient Hyperphenylalaninemia D
PCCA	Propionicacidemia
PCCB	Propionicacidemia
PTS	BH4-deficient Hyperphenylalaninemia A
QDPR	BH4-deficient Hyperphenylalaninemia C
RECQL3	Bloom Syndrome
SLC5A5	Thyroid dyshormonogenesis 1
SLC22A5	Systemic primary carnitine deficiency
SLC25A13	Ault-onset citrullinemia Type II Neonatal onset citrullinemia Type II
SLC25A20	Carnitine-acylcarnitine translocase (CACT) deficiency
SLC26A4	Autosomal recessive deafness type IV Pendred syndrome
SMN1	Spinal muscular atrophy
SMPD1	Niemann-Pick disease, type A Niemann-Pick disease, type B
TAT	Tyrosinemia, type II
TAZ	Barth Syndrome Familial dilated cardiomyopathy
TCN2	Transcobalamin II deficiency
TG	Thyroid dyshormonogenesis 3 Suscptibility to autoimmune thyroid disease Type III
THRA	Congenital nongoitrous hypothyroidism 6
THRB	Thyroid hormone resistance
TPO	Thyroid dyshormonogenesis 2A
TSHB	Congenital nongoitrous hypothyroidism 4
TSHR	Congenital nongoitrous hypothyroidism 1 Familial gestational hyperthyroidism Nonautoimmune hyperthyroidism
TXNB	Congenital Adrenal Hyperplasia due to 21-hydroxylase deficiency