BAYLOR GENETICS

GLOBAL MAPS® DISORDERS LIST 2023

PLASMA (CONFIRMED DISORDERS DETECTED BY GLOBAL MAPS®)

Urea Cycle Disorders

- Argininemia
- Argininosuccinic aciduria
- Citrullinemia
- Ornithine transcarbamylase deficiency
- Orotic aciduria

Fatty Acid Oxidation Disorders

- Short chain acyl-CoA dehydrogenase (SCAD) deficiency
- Medium chain acyl-CoA dehydrogenase (MCAD) deficiency
- Very long chain acyl-CoA dehydrogenase (VLCAD) deficiency

Organic Acidemias

- 2-hydroxyglutaric acidemia (likely L-form)
- 3-hydroxyisobutyrl-CoA hydrolase deficiency (HIBCH)
- 3-hydroxy-3-methylglutaryl (HMG)-CoA lyase deficiency
- 3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency
- Cobalamin biosynthesis disorders
- Ethylmalonic encephalopathy
- Glutaric acidemia type I
- Holocarboxylase synthetase deficiency
- Isovaleric acidemia
- Methylmalonic acidemia
- Propionic Acidemia

Amino Acid Disorders

- Citrin deficiency
- Classical homocystinuria (cystathionine β–synthetase deficiency)
- Glycine encephalopathies
- Hyperphenylalaninemia
- Lysinuric protein intolerance
- Maple syrup urine disease
- Phenylketonuria
- Serine biosynthesis disorders (phosphoserine aminotransferase deficiency, phosphoglycerate dehydrogenase deficiency)
- Tyrosinemia type I

Other

- Adenylosuccinate lyase deficiency
- AICA-ribosiduria (ATIC deficiency)
- Aromatic L-amino acid decarboxylase deficiency
- β-Ureidopropionase deficiency
- Citrate transporter deficiency
- Creatine biosynthesis defects (GAMT and AGAT deficiencies)
- DEGS1 Deficiency
- GABA transaminase deficiency
- Galactosemia
- Glycerol kinase deficiency
- Glycine N-methyltransferase deficiency
- Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome
- Kynurenine 3-monooxygenase (KMO) deficiency
- MTHFR deficiency
- Mitochondrial neurogastrointestinal encephalopathy (MNGIE)
- NAXE gene mutation-related encephalopathy
- NAD(P)HX dehydratase (NAXD) deficiency
- Peroxisome biogenesis disorders/Zellweger spectrum disorders
- Primary carnitine deficiency
- Pyridoxine-dependent epilepsy
- Riboflavin transporter deficiency (SLC25A2)
- Ribose-5-phosphate isomerase deficiency
- Smith-Lemli-Opitz syndrome
- Spondyloepimetaphyseal dysplasia, Genevieve type
- Succinic Semialdehyde Dehydrogenase (SSADH) deficiency
- Thiamine transporter deficiency
- Trimethyllysine hydroxylase epsilon deficiency
- Transaldolase deficiency
- Transketolase deficiency
- Urocanase deficiency (benign condition)
- Xanthurenic aciduria (KYNU deficiency)

PLASMA (DISORDERS THAT SHOULD BE DETECTED BUT HAVE NOT BEEN CONFIRMED)

For this group, we routinely detect one or more plasma analytes that are well established biomarkers for disease.

- 2-Methylbutyryl-CoA Dehydrogenase Deficiency
- 3-Hydroxyacyl-CoA Dehydrogenase (SCHAD) Deficiency
- AMACR Deficiency
- Beta-Ketothiolase Deficiency
- Canavan Disease
- Carbamoyl Phosphate Synthetase I Deficiency
- Carnitine-Acylcarnitine Translocase Deficiency
- Carnitine Palmitoyltransferase I (CPT1) Deficiency
- Carnitine Palmitoyltransferase II (CPT2) Deficiency
- Combined Malonic And Methylmalonic Aciduria
- Dihydropyrimidinase Deficiency
- Dimethylglycine Dehydrogenase Deficiency
- Fructose-1,6-Bisphosphatase Deficiency
- Glutaric Acidemia II
- Gyrate Atrophy Of Choroid And Retina
- Hereditary Fructose Intolerance
- Holocarboxylase Synthetase Deficiency
- Hypermethioninemia due to S-Adenosylhomocysteine Hydrolase Deficiency
- Hypermethioninemia Due To Adenosine Kinase Deficiency
- Hyperoxaluria Type I
- Hyperoxaluria Type II
- Hyperprolinemia, Type I
- Hyperprolinemia, Type II
- Lathosterolosis
- Lesch-Nyhan Syndrome
- Malonyl-CoA Decarboxylase Deficiency
- Molybdenum Cofactor Deficiency
- N-Acetylglutamate Synthase (NAGS) Deficiency
- Phosphoribosylpyrophosphate Synthetase (PRPPS) Superactivity
- Phosphoserine Phosphatase Deficiency
- Purine Nucleoside Phosphorylase Deficiency
- Succinyl-CoA:3-Oxoacid CoA Transferase (SCOT) Deficiency

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GLOBAL MAPS® DISORDERS LIST 2023

PLASMA (DISORDERS THAT MAY BE DETECTED BUT HAVE NOT BEEN VALIDATED)

We identify one or more compounds that are predicted to be relevant to the disorder on the basis of their position within the affected metabolic pathway.

- α-Aminoadipic Aciduria
- 2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency
- 3-Methylglutaconic Aciduria, Type I
- 3-Methylglutaconic Aciduria, Type III
- 3-Methylglutaconic Aciduria, Type V
- 3-Methylglutaconic Aciduria Type VI (with deafness, encephalopathy, and Leigh-like syndrome)
- D-2-Hydroxyglutaric Aciduria Type 1
- D-2-Hydroxyglutaric Aciduria Type 2
- Adenine Phosphoribosyltransferase (APRT) Deficiency
- Asparagine Synthetase Deficiency
- Carnosinemia
- Congenital Bile Acid Synthesis Defect 1 (CBAS1)
- Congenital Bile Acid Synthesis Defect 3 (CBAS3)
- Dihydropyrimidine Dehydrogenase Deficiency
- Essential Fructosuria (considered benign)
- Familial Hypercholanemia (Bile Acid Biosynthesis Disorder)
- Glutamine Deficiency, Congenital
- Glycine N-Methyltransferase Deficiency
- Hydroxykynureninuria
- Hyperphenylalaninemia, BH4-Deficient, A (PTS Deficiency)
- Hyperphenylalaninemia, BH4-Deficient, B (GTP Cyclohydrolase Deficiency)
- Hyperphenylalaninemia, BH4-Deficient, C (DHPR Deficiency)
- Hyperphenylalaninemia, BH4-Deficient, D (PCBD Deficiency)
- Hyperuricemic Nephropathy, Familial Juvenile
- Intrinsic Factor Deficiency; IFD
- Kelley-Seegmiller Syndrome
- Methionine Adenosyltransferase Deficiency
- Prolidase Deficiency
- Succinic Semialdehyde Dehydrogenase Deficiency
- Transcobalamin II Deficiency
- Tyrosinemia, Type II
- Tyrosinemia, Type III
- Xanthinuria, Type I

Limitations to testing: Disorders we presume we cannot detect on this small molecule test

- Congenital disorders of protein glycosylation
- Glycogen storage diseases
- Oligosaccharidoses
- Mucopolysaccharidoses
- Other lysosomal storage diseases

URINE (THE FOLLOWING DISORDERS DETECTABLE ONLY BY URINE)

- Creatine transporter deficiency (detectable in urine Global MAPS® only)