PEDIATRIC

BAYLOR GENETICS

GLOBAL MAPS™
Global Metabolomic Assisted Pathway Screen

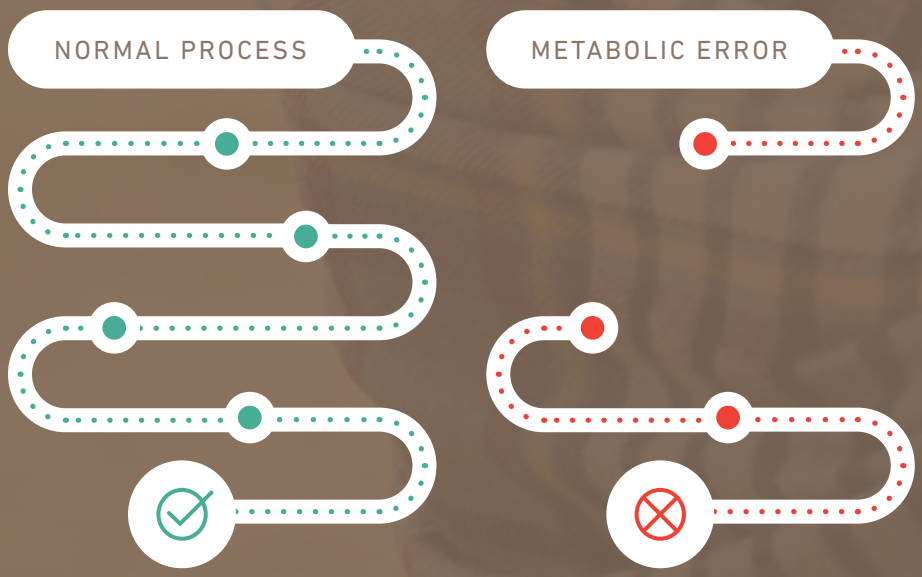
Assessing or diagnosing a metabolic disorder commonly requires several tests. Global Metabolomic Assisted Pathway Screen, commonly known as Global MAPS, is a unifying test for analyzing hundreds of metabolites to identify changes or irregularities in biochemical pathways. Let Global MAPS guide you to an answer.

Diagnose a broad range of metabolic disorders with a single test, Global MAPS

Global MAPS is a large scale, semi-quantitative metabolomic profiling screen that analyzes disruptions in both individual analytes and pathways related to biochemical abnormalities.

Using state-of-the-art technologies, Global Metabolomic Assisted Pathway Screen (Global MAPS) provides small molecule metabolic profiling to identify >700 metabolites in human plasma, urine, or cerebrospinal fluid. Global MAPS identifies inborn errors of metabolism (IEMs) that would ordinarily require many different tests. This test defines biochemical pathway errors not currently detected by routine clinical or genetic testing.

IEMs are inherited metabolic disorders that prevent the body from converting one chemical compound to another or from transporting a compound in or out of a cell.



These processes are necessary for essentially all bodily functions. Most IEMs are caused by defects in the enzymes that help process nutrients, which result in an accumulation of toxic substances or a deficiency of substances needed for normal body function. Making a swift, accurate diagnosis of an IEM and prescribing the appropriate diet or medication is critical in preventing brain damage, organ damage, and even death.

Global MAPS Indications For Testing

- AUTISM SPECTRUM DISORDER
- DEVELOPMENTAL DELAY
- VARIANTS OF UNCERTAIN CLINICAL SIGNIFICANCE IN A GENE KNOWN TO BE INVOLVED IN SMALL MOLECULE METABOLISM
- FAILURE TO THRIVE
- HYPOGLYCEMIA
- HYPOTONIA
- NON-SYNDROMIC INTELLECTUAL DISABILITY
- RECURRENT VOMITING
- SEIZURES
- SPEECH/LANGUAGE DELAY
- UNDIFFERENTIATED PHENOTYPE POSSIBLY RELATED TO PERTURBATION IN A BIOCHEMICAL PATHWAY

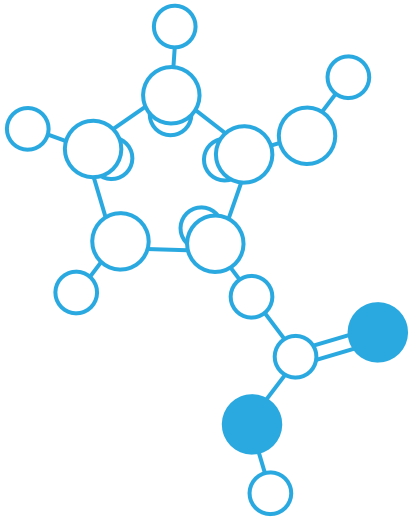
Global MAPS provides pati
assist in the diagnosis of a
recognize new IEMs never

The possibility of discovery by using Global MAPS is considerable, including the identification of new inborn errors of metabolism and regulatory factors for metabolic genes, as well as previously unknown metabolic associations/ disruptions with known disorders. Global MAPS offers a broad range of analyses in a single metabolic screen, requiring less sample volume from the patient making it more convenient and cost-effective.

Global MAPS is a unique broad screening test that can detect disorders involving metabolism of amino acids, organic acids, fatty acid oxidation, vitamin cofactors, pyrimidine biosynthesis, creatine biosynthesis, and urea cycle metabolism, among other known disorders.

Metabolites range in size and include, but are not limited to:

- AMINO ACIDS
- FATTY ACIDS
- ORGANIC ACIDS
- NEUROTRANSMITTERS
- NUCLEOTIDES
- BILE ACIDS



ents with a single test that can broad range of disorders and before described.

Testing Options

GLOBAL METABOLOMIC ASSISTED PATHWAY SCREEN (GLOBAL MAPS)

Test Code	4900	4901	4902
Specimen Type			
TAT (Days)	21	21	21

Specimen Requirements & Shipping Conditions

	Send 1-2 cc of plasma. Draw blood in an EDTA (purple top) tube(s) and separate plasma as soon as possible, freezing immediately. Store the specimen frozen at -20°C. Specimen may be stored frozen up to 7 days.	Ship frozen sample in insulated container, with 3 -5 lbs. dry ice, by overnight courier.
	Send 1-2 cc of cerebrospinal fluid. Store the specimen frozen at -20°C. Specimen may be stored frozen for up to 7 days.	Ship frozen sample in insulated container, with 3 -5 lbs. dry ice, by overnight courier.
	Send 3-5 cc of a random urine. Do not add preservatives. Store the specimen frozen at -20°C.	Ship frozen sample in insulated container, with 3 -5 lbs. dry ice, by overnight courier.

Plasma | Urine | Cerebrospinal Fluid | EDTA (Purple Top)

40 YEARS OF INNOVATION

4 MILLION+ CLINICAL TESTS PERFORMED

1 MILLION+ FAMILIES HELPED

3 THOUSAND+ TESTS OFFERED

1 MISSION IMPROVE HEALTHCARE THROUGH GENETICS

Baylor Genetics pioneered the history of genetic testing. Now, we're leading the way in precision medicine.

Baylor Genetics is a joint venture of Miraca Holdings, Inc. and Baylor College of Medicine, including the #1NIH funded Department of Molecular and Human Genetics. A pioneer of precision medicine for nearly 40 years, Baylor Genetics now offers a full spectrum of clinically relevant genetic testing, access to world-renowned experts, and the confidence to provide patients with the best care.

1.800.411.4363
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METABOLON®

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10.24.18

Global MAPS Disorders List

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GLOBAL MAPS™ DISORDERS LIST 2018

PLASMA (CONFIRMED DISORDERS DETECTED BY GLOBAL MAPS)

Urea Cycle Disorders

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

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

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

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
- Aromatic L-amino acid decarboxylase deficiency
- β-Ureidopropionase deficiency
- Citrate transporter deficiency
- Creatine biosynthesis defects (GAMT and AGAT deficiencies)
- GABA transaminase deficiency
- Galactosemia
- Glycerol kinase deficiency
- Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome
- MTHFR deficiency
- Mitochondrial neurogastrointestinal encephalopathy (MNGIE)
- Peroxisome biogenesis disorders/Zellweger spectrum disorders
- Primary carnitine deficiency
- Pyridoxine-dependent epilepsy
- Ribose-5-phosphate isomerase deficiency
- Smith-Lemli-Opitz syndrome
- Spondyloepimetaphyseal dysplasia, Genevieve type
- Thiamine transporter deficiency
- Trimethyllysine hydroxylase epsilon deficiency
- Transaldolase deficiency
- Transketolase deficiency
- Urocanase deficiency (benign condition)

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

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 2018

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)

CEREBROSPINAL FLUID (THE FOLLOWING DISORDERS DETECTABLE ONLY BY CEREBROSPINAL FLUID)

NFU1 deficiency (detectable in CSF Global MAPS only)