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 patients a single test that can assist in the diagnosis of a broad range of disorders and recognize new IEMs never before described. 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**
Global Metabolomic Assisted Pathway Screen (GLOBAL MAPS)

Testing Options

<table>
<thead>
<tr>
<th>Test Code</th>
<th>4900</th>
<th>4901</th>
<th>4902</th>
</tr>
</thead>
<tbody>
<tr>
<td>Specimen Type</td>
<td>Plasma</td>
<td>Urine</td>
<td>Cerebrospinal Fluid</td>
</tr>
<tr>
<td>TAT (Days)</td>
<td>21</td>
<td>21</td>
<td>21</td>
</tr>
</tbody>
</table>

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.
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 #1 NIH 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.
Global MAPS Disorders List
## 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

### 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.

<table>
<thead>
<tr>
<th>Organic Acidemias</th>
<th>Amino Acid Disorders</th>
<th>Other</th>
</tr>
</thead>
<tbody>
<tr>
<td>2-hydroxyglutaric acidemia (likely L-form)</td>
<td>Citrin deficiency</td>
<td>Adenylosuccinate lyase deficiency</td>
</tr>
<tr>
<td>3-hydroxyisobutyryl-CoA hydrolase deficiency (HIBCH)</td>
<td>Classical homocystinuria (cystathionine β-synthetase deficiency)</td>
<td>Aromatic L-amino acid decarboxylase deficiency</td>
</tr>
<tr>
<td>3-hydroxy-3-methylglutaryl-CoA lyase deficiency</td>
<td>Glycine encephalopathies</td>
<td>β-Ureidopropionase deficiency</td>
</tr>
<tr>
<td>3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency</td>
<td>Hyperphenylalaninemia</td>
<td>Citrate transporter deficiency</td>
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<tr>
<td>Cobalamin biosynthesis disorders</td>
<td>Lysinuric protein intolerance</td>
<td>Creatine biosynthesis defects (GAMT and AGAT deficiencies)</td>
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<td>Ethylmalonic encephalopathy</td>
<td>Maple syrup urine disease</td>
<td>GABA transaminase deficiency</td>
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<tr>
<td>Glutaric acidemia type I</td>
<td>Phenylketonuria</td>
<td>Galactosemia</td>
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<tr>
<td>Holocarboxylase synthetase deficiency</td>
<td>Tyrosinemia type I</td>
<td>Glycerol kinase deficiency</td>
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<td>Isovaleric acidemia</td>
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<td>Methylmalonic acidemia</td>
<td></td>
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<td>Propionic Acidemia</td>
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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-Oxoadic CoA Transferase (SCOT) Deficiency
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)**

**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**

**Creatine transporter deficiency (detectable in urine Global MAPS only)**

**NFU1 deficiency (detectable in CSF Global MAPS only)**