



# Global MAPS®

## Global Metabolomic Assisted Pathway Screen

Metabolomic profiling is a large scale, semi-quantitative screening test that looks at perturbations in both individual analytes and pathways related to biochemical abnormalities. Using state-of-the-art UPLC-MS/MS technologies, Global MAPS® provides small molecule metabolic profiling to identify >700 metabolites in human plasma, urine, or CSF. Metabolites range in size from 50-1500 Da and include, but are not limited to, amino acids, organic acids, fatty acids, neurotransmitters, nucleotides, and bile acids, all identified in a single test.

Testing has the ability to identify 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.

A significant feature of this testing is the potential to combine the analysis with Proband Whole Exome Sequencing data to assess the consequences of DNA variants in known or suspected metabolic pathways. The potential for discovery with this approach is considerable, including the identification of new inborn errors of metabolism and new regulatory factors for metabolic genes, as well as previously unknown metabolic associations/perturbations with known disorders. Global MAPS offers a broad range of analyses in a single metabolic screen, requiring less sample volume from the patient and reducing the potential for sample mix-up due to multiple sample collections and standardization of results across the population.



### GLOBAL MAPS® INDICATIONS FOR TESTING

- Developmental delay
- Seizures
- Autism spectrum disorder
- Undifferentiated phenotype possibly related to perturbation in a biochemical pathway
- Equivocal molecular test results in a gene known to be involved in small molecule metabolism

Use of this test in combination with exome analysis may be most beneficial in more difficult cases or in cases where a small molecule inborn error of metabolism is suspected or when a variant of unknown significance in a metabolic pathway is identified on genomic analysis.

Go to [www.bmgl.com](http://www.bmgl.com) to view specific specimen requirements and shipping conditions.