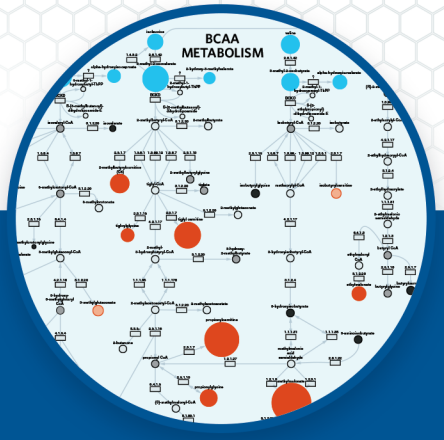
MetaIMD™



Meta IMD™

THE POWER OF PRECISION MEDICINE FOR INHERITED METABOLIC DISORDERS

INHERITED METABOLIC DISORDERS (IMDs) CAN CAUSE DEVELOPMENTAL DELAYS, OTHER SERIOUS MEDICAL PROBLEMS, AND EVEN DEATH IF THEY ARE NOT CONTROLLED. EARLY DIAGNOSIS IS ESSENTIAL, BUT OFTEN DIFFICULT BECAUSE OF THE DIVERSITY OF METABOLIC PATHWAYS INVOLVED AND THE COMPLEXITIES THAT ACCOMPANY AN UNDIFFERENTIATED PHENOTYPE.

Meta IMD is a comprehensive biochemical pathway analysis that can potentially save significant cost and time to diagnose. Used as an adjunctive, first-line clinical test, Meta IMD helps the clinician optimize selection of confirmatory targeted assays. The test requires only 250 µL of human plasma. With its ability to detect more than 800 metabolites, Meta IMD may also spot disorders for which there is presently no biochemical testing available.

META IMD

- Adjunctive, first-line clinical test
- Convenient
  - One small sample
- Enables quicker diagnosis
  - Results in about 21 days

Image header: Pathway image demonstrating how the Meta IMD test visualizes the disorders.

Meta IMD should be considered for individuals with:

- ✓ Developmental delay
- ✓ Non-syndromic intellectual disability
- ✓ Seizures
- ✓ Autism spectrum disorder
- ✓ Failure to thrive
- ✓ Hypoglycemia
- ✓ Recurrent vomiting
- ✓ Speech and/or language delay
- ✓ Hypotonia
- ✓ Undifferentiated phenotype (possibly related to a biochemical pathway perturbation)
- ✓ Equivocal molecular test results in a gene known to be involved in small molecule metabolism

Meta IMD tests for:

- ✓ Amino acid metabolism disorders
- ✓ Organic acidemias
- ✓ Fatty acid oxidation disorders
- ✓ Disorders of fatty acid transport
- ✓ Purine disorders
- ✓ Pyrimidine disorders
- ✓ Certain mitochondrial disorders
- ✓ MNGIE (mitochondrial neurogastrointestinal encephalopathy)
- ✓ Creatine disorders
- ✓ Bile acid disorders
- ✓ Urea cycle disorders
- ✓ Neurotransmitter disorders (previously only diagnosed in CSF)

How Does Meta IMD Work ?

Meta IMD is a Laboratory-Developed Test (LDT) run in a CLIA-certified/CAP-accredited laboratory. The test simultaneously evaluates hundreds of compounds in a single plasma sample using Metabolon's proprietary analytics and informatics to identify significant single metabolite or metabolic pathway abnormalities relative to a reference cohort.

About Inherited Metabolic Disorders

IMDs, also known as inborn errors of metabolism (IEMs), are rare genetic disorders that often disrupt bodily processes such as the conversion of nutrients into energy and structural molecules, the breakdown and clearance of waste, or the synthesis and function of regulatory signals. The disorders are usually caused by defects in specific proteins (enzymes) that help facilitate these processes.

Generally, IMDs represent a group of about 500 rare genetic disorders with an overall estimated incidence of one out of 2,500 people.

Presentation of an IMD is usually in the neonatal period or infancy, but can occur at any time, even in adulthood. Some metabolic disorders can be identified by routine newborn screening tests. Others are identified only after a child or adult shows symptoms of a disorder.

Requirements & Limitations

Meta IMD must be ordered by a physician and accompanied by detailed clinical information. Special diets, supplements and medications should be disclosed. For example, total parenteral nutrition (TPN) is known to affect test results.

A few of the key limitations are provided below.

The test does not measure:

- Proteins or large peptides
- Complex oligosaccharides
- Large lipids
- Elements such as potassium or sodium

The test is not intended to identify lysosomal disorders, congenital disorders of glycosylation, mucopolysaccharidoses, mucolipidoses, or similar large-molecule disorders. However, Meta IMD can detect relative abnormal levels of metabolites that are the building blocks of small molecules associated with these disorders.

Meta IMD is not meant to replace diagnostic testing for specific conditions, but is used to assist the clinician in choosing the appropriate diagnostic and confirmatory tests.

Supported by analytical validation of
METABOLITES

Clinically validated
INHERITED METABOLIC DISORDERS

Technical Information

Metabolomic profiling is a large-scale, semi-quantitative technology offering highly efficient small-molecule analysis of metabolites ranging in size from 50 to 1,500 Da.

Methodology: Meta IMD identifies known metabolites in human plasma. The test analyzes hundreds of compounds simultaneously using UPLC-MS/MS technologies.

Significant abnormalities in metabolic pathways and/or in selected single analytes may be identified. Follow-up testing for confirmation should be considered at the discretion of the physician.

Test type: Analyte analyses which have been grouped to validate changes associated with disease.

Reference Values or Ranges: Metabolites and biomarkers that fall out of the expected range will be reported relative to a reference population consisting of 866 pediatric patient samples. Individual concentrations are not reported.

About Metabolon

Metabolon was founded in 2000 and is the world's leading health technology company, advancing metabolomics for precision medicine and every area of life sciences research.

FOR MORE INFORMATION, PLEASE CONTACT US AT META-IMD@METABOLON.COM OR 1.844.363.META (1.844.363.6382).

METABOLON®
Where knowing comes to life™

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