

What is Plasma Homocystine Quantitation?

This test is useful for faster quantification of homocystine (and methionine) to confirm Homocystinuria and Methylenetetrahydrofolate reductase (MTHFR) deficiency. It is also helpful in detecting methionine synthase deficiency or disorders of methylcobalamin synthesis.

What is Carnitines-Organic Acids Panel?

This is a package test inclusive of Plasma Acylcarnitines Quantitation and Urine Organic Acid Analysis for wide-range metabolic profiling.

What are overseas tests?

These are tests done in laboratories abroad. The Biochemical Genetics Laboratory has collaborated with laboratories in the USA, Australia, and Taiwan so that samples are prepared locally and sent out to them for the requested IEM tests. Please inquire from your physician regarding the specified tests. Other metabolic tests not mentioned on the list could likewise be arranged by the laboratory. Please contact the laboratory for further details such as costing, sample requirements, and schedule.

What are the sample and transport requirements for the tests?

Samples can be collected at the clinical room of the institute, or can be done in other medical facility or patient’s home depending on the requirement.

What should be done if the result is positive?

Results should be discussed with your physician. For any assistance, please feel free to contact the laboratory.

Contact Biochemical Genetics Laboratory for more information:

Biochemical Genetics Laboratory

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For inquiries on services, please contact:



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BIOCHEMICAL GENETICS LABORATORY

Provides services for the diagnosis and management of inborn errors of metabolism



The Institute of Human Genetics is ISO 9001:2008 certified



BIOCHEMICAL GENETICS LABORATORY

The Biochemical Genetics Laboratory is a specialized laboratory that renders services for the diagnosis and clinical management of inborn errors of metabolism (IEM). The laboratory started in 2001. Locally-available tests are offered, as well as overseas tests for some highly specialized diagnostic tests.

The locally-available tests are as follows:

- Urine Metabolic Screening
- Urine Organic Acid Analysis
- Comprehensive Urine Metabolic Profile
- Plasma Quantitative Amino Acid Analysis
- Paired CSF/Plasma Quantitative Amino Acid Analysis
- Plasma Branched Chain Amino Acids Quantitation
- Plasma Homocystine Quantitation
- Metabolic Acids Panel
- Plasma Acylcarnitines Quantitation
- Carnitines-Organic Acids Panel

The overseas tests currently available are as follows:

- Biotinidase screen
- Tetrahydrobiopterin (BH4) Deficient Hyperphenylalaninemia
- Lysosomal Enzyme Assay
- Peroxisomal Lipid Panel
- Transferrin Isoforms
- Very Long Chain Fatty Acid Quantitation
- Lysosomal Storage Disease Enzyme Assays

FREQUENTLY ASKED QUESTIONS (FAQs)

What are inborn errors of metabolism (IEM)?

IEM is a group of disorders that arise from a block in a metabolic pathway, which could be due to either an enzyme deficiency or an abnormality of a transport process (Holton, 1987).

What are the common signs and symptoms of IEM?

The signs and symptoms that are commonly seen in inborn metabolic disorders are:

- Developmental delay
- Coarse facial features (prominent forehead, abundant dry coarse hair, flat nose, thick lips, large tongue)
- Seizures / epilepsy
- Movement disorder
- Enlarged liver, spleen, or heart
- Skeletal abnormalities (joint contractures, short stature)
- Intellectual disability
- Muscle weakness / poor muscle tone
- Hematologic abnormalities (thrombocytopenia, underwent splenectomy)
- Liver dysfunction
- Renal tubular disease
- Laboratory abnormalities (hyperammonemia, hypoglycemia, metabolic acidosis)

What is Urine Metabolic Screening?

This test is a semi-quantitative screen mainly for amino acid disorders such as Maple Syrup Urine Disease (MSUD), Phenylketonuria (PKU), Homocystinuria, Cystinuria, Non-ketotic hyperglycinemia (NKH), Tyrosinemia, sarcosinemia, hyperlysinemia and histidinemia. It also screens for organic acidopathies namely methylmalonic aciduria and malonic aciduria.

What is Urine Organic Acid Analysis?

Organic acid analysis by Gas Chromatography/Mass Spectrometry (GC/MS) is a well-established procedure for the diagnosis of classical and other organic acidurias, ornithine transcarbamoylase deficiency, and fatty acid oxidation defects. This technique detects multiple obstructed metabolite systems in a single analysis and multiple marker metabolites can be monitored simultaneously. It is a semi-quantitative analysis.

What is Comprehensive Urine Metabolic Profile?

This is Urine Metabolic Screening and Organic Acid Analysis, requested as one test, with discounted test fee and comprehensive diagnosis

What is Plasma Quantitative Amino Acid Analysis?

This test is for definitive diagnosis and monitoring of amino acid disorders such as Phenylketonuria (PKU), Urea Cycle Defects (Ornithine transcarbamylase deficiency, Carbamoyl phosphate synthase deficiency, Argininosuccinate synthase deficiency, argininosuccinate lyase deficiency), Tyrosinemia, Glycine encephalopathy (Non-ketotic hyperglycinemia), Maple Syrup Urine Disease (MSUD), etc.

What is Paired CSF/Plasma Quantitative Amino Acid Analysis?

It is an ideal diagnostic tool for Nonketotic hyperglycinemia, preferably correlated with urine organic acid analysis to rule out ketotic hyperglycinemia.

What is Plasma Branched Chain Amino Acids Quantitation?

This is offered for faster quantification of branched chain amino acids leucine, isoleucine, allo-isoleucine, and valine for confirmation and monitoring of MSUD.