BIOCHEMICAL GENETICS (BCG) LAB

Co-Directors: Drs. Patrice Held & Gregory Rice
Staff: April Meiller, Phil Williams, Brandon Shelton

The Biochemical Genetics (BCG) laboratory performs testing for patients (primarily children) who have suspected or known inborn (hereditary) metabolic disorders (i.e. those due to genetic defect). BCG lab services compliment the Newborn Screening Program.

Clinical history and information pertaining to recent medications/therapy is required for appropriate interpretation of test results.

Specimen handling:
- *plasma specimens (from heparinized blood) and serum specimens should be separated from blood cells as soon as possible after collection (centrifuged within 20 minutes);*
- *avoid dilute urine specimens if possible; first morning urine is preferred;*
- *serum, plasma and urine specimens should be frozen as soon as possible after collection and shipped frozen (to avoid multiple freeze-thaw cycles).*
- *whole blood should NOT be frozen.*

BCG TESTS OFFERED

506 Amino Acids Quantitative – plasma (from heparinized blood)
plasma (1 mL), frozen

552 Amino Acids Quantitative – serum
serum (1 mL), frozen

553 Amino Acids Quantitative – urine
urine (1 mL), frozen

stability: ambient – unacceptable
refrigerated – 24 hours
frozen – 1 month

Background: Defects in amino acid metabolism may results in the accumulation of one or more amino acids in the blood (aminoacidemia) or excess excretion in urine (aminoaciduria) or both. Errors in amino acid metabolism may lead to such disorders as phenylketonuria (PKU), maple syrup urine disease (MSUD), homocystinuria, glycine encephalopathy and various urea cycle defects. (Note: Many defects in organic acid metabolism also have secondary abnormalities in amino acid levels.) Clinical information is particularly important for appropriate interpretation.

565 Amino Acids Dietary Screen Quantitative – filter paper
filter paper stability: ambient (do not expose to extreme heat or cold)

Dietary screen is performed for the monitoring of certain amino acids in patients with known defects in amino acid metabolism who are receiving dietary therapy. Results may vary from plasma levels, thus this test should not be used for diagnostic purposes.

520 Biotinidase
serum (1 mL), frozen on dry ice

avoid more than one freeze-thaw cycle

stability: ambient – unacceptable (1 hour)
refrigerated – unacceptable (1 hour)
frozen – 1 month

Biotinidase is an enzyme that frees biotin (vitamin B) from food for use by the body. Symptoms of untreated biotinidase deficiency may appear anytime between 1 week and 10 years of age. Biotinidase deficiency may lead to seizures, delayed development, vision problems, speech problems, respiratory problems, eczema, hearing loss, etc.

Proper specimen handling is critical. This is a test for enzyme activity. Exposure to heat, or multiple freeze-thaw cycles can damage the enzyme and lead to a false negative result.

531 Carnitine
plasma (from heparinized blood), (1 mL), frozen

Separate plasma from cells (centrifuge within 20 minutes of collection) and freeze immediately (avoid hemolysis)
avoid more than one freeze-thaw cycle (ship frozen)

stability: ambient – unacceptable
refrigerated – 12 hours
frozen – 1 month

Useful in diagnosing mitochondrial fatty acid beta oxidation disorders and several other organic acidemias.

Background: Carnitine is a protein essential to fat metabolism (it is required to carry long chain fatty acids across the mitochondrial membrane; fatty acid oxidation occurs in the mitochondria to produce energy). Carnitine is stored mostly in muscle, only about 2% is found in blood, but blood is the source specimen of choice for testing because it is less invasive than obtaining muscle tissue. Carnitine deficiency can lead to muscle weakness (including the heart) and even sudden death. Treatment with oral carnitine is quite effective in treating cardiomyopathy and muscle weakness in children with carnitine deficiency.

554 Organic Acids, Comprehensive Quantitative
urine (5-10 mL), frozen

First morning urine is preferred; avoid dilute urine if possible
Freeze as soon as possible after collection
Provide clinical history; required for appropriate interpretation

Stability: ambient – unacceptable
refrigerated – unacceptable
frozen – 1 month

Will identify nearly all known organic acidemias.

Organic acids are a broad class of compounds used in fundamental metabolic processes of the body (i.e. to generate cellular energy from dietary protein, fat and carbohydrate and provide the building blocks necessary for cell function). Accumulation of specific organic acids in urine often signals a metabolic inhibition or block. Examples of disorders identified by this test include methylmalonic acidemia, propionic acidemia, isovaleric acidemia, glutaric acidemia types I and II, Canavan disease and many others. Patients with certain fatty acid oxidation disorders and amino acid disorders also have organic acid abnormalities in urine.