

New Assays Added to the Menu!

REDUCED AND OXIDIZED GLUTATHIONE

Consider ordering for: patients in whom mitochondrial dysfunction or other conditions where oxidative stress is suspected, monitoring for known mitochondrial disease, and monitoring the efficacy of n-acetylcysteine therapy.

The analysis: The Glutathione Panel quantifies reduced (GSH) and oxidized glutathione (GSSG). GSH is used as a reducing agent for reactive oxygen species in the mitochondria and converts it into GSSG. The ratio between the two is determined by the amount of oxidative stress in the mitochondria. Low total glutathione, high GSSG or a low GSH/GSSG ratio, can reflect high oxidative stress, which can be seen in primary mitochondrial disease or conditions with secondary mitochondrial stress. These values can acutely worsen in patients with mitochondrial disease undergoing metabolic stress. GSH/GSSG has been used to measure disease risk in a variety of other conditions, including diabetes, renal failure, neurodegenerative diseases, multiple sclerosis and cystic fibrosis. Glutathione may also be depleted by some pharmacologic agents, including ethanol and acetaminophen, particularly in supratherapeutic amounts.

OXIDATIVE PHOSPHORYLATION FIBROBLAST

Consider ordering for: patients requiring subcategorization of the presence of mitochondrial dysfunction, such as in genetic disease (primary mitochondrial disease) or secondary to other diseases.

The analysis: The Fibroblast OxPhos (oxidative phosphorylation) Analysis analyzes oxygen consumption as a proxy measure of mitochondrial function in skin cells. Cells are fed substrates and inhibitors to identify oxygen consumption rates specific for mitochondrial complexes I-IV. Oxygen consumption is also measured both coupled and uncoupled from the production of ATP. Fibroblast OxPhos Analysis will be offered as a panel with cell line culture and grow.

KETONE BODIES PANEL

Consider ordering for: patients in whom mitochondrial dysfunction is suspected or other conditions in which acetoacetate is a key component, such as diabetes mellitus type 1 and catabolic states. This test can be used to monitor adherence to therapeutic diets (ketogenic diets), assessment of low-level ketosis for treatment adherence, efficiency of ketone utilization, and monitoring for complications such as ketoacidosis.

The analysis: The Ketone Bodies Panel is a new rapid LC-MS/MS-based multiplex panel that quantifies 5 ketone bodies (acetoacetate (AcAc), beta-hydroxybutyrate (BHB), alpha-hydroxybutyrate (AHB), gamma-hydroxybutyrate (GHB), and beta-hydroxyisobutyrate). The ratio between BHB and AHB is determined by the ratio of NAD⁺ and NADH in the mitochondria, and therefore can be used as a proxy measure of the mitochondrial redox state and a sensitive, specific marker for mitochondrial dysfunction. **It is the only currently available version of this test.** The ratios assessed are used as a proxy measure

Metabolic & Advanced Diagnostics Lab

Clinical or Diagnostic Consultations: Please contact Rebecca Ganetzky, MD – a clinical attending and Director of Mitochondrial Biochemical Diagnostics: ganetzkyr@chop.edu. Additional Lab Directors are available, as requested

Mitochondrial Medicine Program Patient Referrals: call 267-426-4961 or email mmfp@chop.edu.

Test Menu



Requisition Form



Glutathione Kit Request



Specimen Requirements



Please send samples to:

Attn: Metabolic and Advanced Diagnostics | STAT Lab
Specimen

Children's Hospital of Philadelphia
Main Hospital, 5th Floor Room 5NW55
3401 Civic Center Blvd
Philadelphia, PA 19104

For pricing and/or more information about
the test menu and how to order, please
contact:

Laboratory Outreach Team
clinicalaboutreach@chop.edu