



## **Baylor Institute of Metabolic Disease Mission**

The Institute of Metabolic Disease aims to provide expert clinical, biochemical, and molecular diagnostic services that complement the goals of ongoing research and novel clinical services designed to improve the lives of individuals with genetic diseases worldwide.

## **Kimberly H. Courtwright and Joseph W. Summers Institute of Metabolic Disease**

The Kimberly H. Courtwright and Joseph W. Summers Institute of Metabolic Disease provides comprehensive diagnostic and treatment services to children and adults suffering from inherited metabolic diseases. Since its creation, the institute has developed clinical, research and educational programs and is recognized as a world-renowned research institute in metabolic disease.

A metabolic disease is a disorder caused by a problem with the accumulation of chemicals produced naturally in the body, many of which interfere with the ability to produce energy from protein, fats and carbohydrates in the diet. These diseases are usually serious, some even life-threatening, and others may slow down physical development or cause mental retardation.

The Baylor Institute of Metabolic Disease is part of Baylor Research Institute, an affiliate of Baylor Health Care System. For information about the institute and tests offered, visit [www.baylorhealth.edu/imd](http://www.baylorhealth.edu/imd).

### **CLINICAL HIGHLIGHTS**

The Baylor Institute of Metabolic Disease offers clinical testing to assist medical professionals in the diagnosis and treatment of inherited metabolic diseases. The institute is composed of three main laboratories:

- **The Mass Spectrometry laboratory** utilizes tandem mass spectrometry and gas chromatography/mass spectrometry to diagnose a number of amino acid, organic acid and fatty acid oxidation disorders. Recently, new tests have been added for the rapid follow-up of abnormalities detected by expanded newborn screening. The institute's Supplemental Newborn Screening program offers screening for approximately 40 inherited disorders, testing for more disorders than most state newborn screening programs. Additionally, the institute is developing new screening tests to increase the number of metabolic disorders that can be detected.
- **The Neuropharmacology laboratory** studies several inborn errors of metabolism that affect the central nervous system. Also known as Pediatric Neurotransmitter Diseases (PNDs), they often result in severe mental retardation, developmental delay, seizures, motor abnormalities and cognitive deficits. Early detection and treatment of PNDs is critical to prevent irreversible damage to the central nervous system. The Baylor Institute of Metabolic Disease has recently introduced new tests that can aid in the diagnosis of PNDs by performing specialized tests on cerebrospinal fluid using High Performance Liquid Chromatography.
- **The Molecular Diagnostic laboratory** offers DNA testing for several fatty oxidation disorders, such as Medium Chain Acyl-CoA Dehydrogenase Deficiency, also known as MCAD. DNA sequencing and enzyme assays for certain fatty oxidation disorders also is available.

#### **FOR MORE INFORMATION, CONTACT:**

Baylor Health Care System  
Marketing/Public Relations  
2001 Bryan Street, Ste. 750  
Dallas Texas 75201  
Phone: 214-820-3055  
Fax: 214-820-4697

[www.BaylorHealth.com](http://www.BaylorHealth.com)

**1 - 800 - 4 BAYLOR**

## RESEARCH HIGHLIGHTS

- **Inherited Disorders of Fat Metabolism**

Several conditions frequently encountered in pediatric medicine are associated with metabolic disease. Researchers at the institute seek ways to improve diagnosis of these conditions and devise new more effective treatments, including special diets, vitamin supplements and medication. Among the Baylor Institute of Metabolic Disease's successes is a method to determine which enzymes are defective in the metabolism or breakdown of fatty acids and amino acids. This system has improved physicians' ability to diagnose certain metabolic diseases before and after birth, and has led to new methods of treatment. It also has allowed researchers to identify previously unknown inherited defects.

- **Disorders of Folate, B-vitamins and Homocysteine Metabolism**

Inherited and acquired disorders of folate, vitamin B12, vitamin B6 and homocysteine metabolism can lead to vascular disease and various neurological and psychiatric complications. Researchers at the institute have been actively studying the effects of altered folate, B-vitamins and homocysteine metabolism in relation to cardiovascular, cerebrovascular disease and disorders of the central nervous system, including depression, dementia and Parkinson's disease.

- **Molecular Genetics Research**

Current research is focused on developing novel diagnostic and treatment strategies for lysosomal and myocardial glycogen storage disease, such as Pompe's disease. Researchers are also working on developing mutation screening techniques for familial hypertrophic cardiomyopathy.