The Institute of Metabolic Disease (IMD), part of Baylor Research Institute, develops diagnostics and therapeutics that harness the complementary fields of metabolomics and metabolic disease, which are the focus of the Center of Metabolomics and the Center for Metabolic Translational Research, respectively. The two centers work in concert to address areas of unmet medical needs that span both rare and common diseases. Under the leadership of Dr. Raphael Schiffmann, IMD has developed the know-how and expertise to make significant strides, as evidenced by the discovery and development of novel diagnostics and therapeutics in a large number of disease indications.

**Center of Metabolomics**

The Center of Metabolomics, led by Dr. Teodoro Bottiglieri and Dr. Lawrence Sweetman, is at the forefront of developing new metabolic biomarker panels in various disease states including cancer, diabetes, heart disease, renal insufficiency, depression and dementia. The center is expanding its disease scope using a combination of targeted and discovery (untargeted) metabolomics, thereby increasing the chance of revealing clinically useful information and potentially identifying novel biomarkers.
Through its CLIA-certified, CAP-accredited laboratory, the center provides analysis of dried blood spots, urine and plasma samples for organic acids (100+ compounds), carnitine/acyl carnitines (32 compounds) and amino acids (13 compounds) using mass spectrometry (MS). The laboratory can diagnose more than 40 specific metabolic disorders and performs about 7,380 tests each year using advanced GC-MS and LC-MS analysis, primarily for the testing of newborns. The laboratory also provides diagnostic testing for difficult-to-diagnose pediatric neurotransmitter diseases, which affect dopamine, serotonin and GABA metabolism. Using LC-MS/MS and HPLC, the laboratory analyzes about 400 patient samples annually.

In addition to providing vital diagnostic testing for newborn and pediatric patients, the laboratory provides analytical biomarker services for clinical trials to enable patient stratification and monitoring.

**Center of Metabolic Translational Research**

The Center of Metabolic Translational Research, led by Drs. Schiffmann and Bottiglieri, is harnessing the knowledge and infrastructure of IMD to address serious unmet medical needs.

With a focus on lysosomal storage disorders, the Center of Metabolic Translational Research has discovered and patented novel biomarkers and treatments for Fabry disease. Serendipitously, Dr. Schiffmann and colleagues discovered that elevated urinary globotriasylceramide (Gb₃) a hallmark of Fabry disease, is a biomarker for the risk of near-term mortality in heart disease patients who do not have Fabry disease. This is the first time that heart disease has been linked to lipid abnormalities in organs outside of the heart with common forms of heart disease, and “could be of great significance for the future study of heart disease,” Dr. Schiffmann said. Importantly, this discovery illustrates IMD’s paradigm of using metabolomics to uncover the connection between rare and common diseases.

Another focus for the center is exploring the role of folate metabolism and methylation in major vascular diseases, including coronary heart disease and renal disease, as well as neurodegenerative diseases such as Alzheimer’s and Parkinson’s disease.

**Biobank and Cell Lines of Genetic Diseases**

Over the past decade of clinical research, IMD has amassed invaluable resources, including a large biobank of plasma, serum, urine, cerebrospinal fluid and tissue collected from more than 4,000 patients with a variety of genetic disorders, as well as healthy controls. In addition, IMD has well over 1,000 cell lines representing a broad range of heritable diseases such as lysosomal storage diseases (including Fabry, Gaucher, mucolipidosis IV), leukodystrophies and many more. This is arguably one of the largest genetic disease cell line collections of its kind.

**Opportunities**

Patent applications have been filed for the biomarkers and therapies discovered by IMD researchers and are available for licensing. Opportunities also exist for partnering and collaboration to further advance these technologies.

In addition, the services of the CLIA laboratory are available for clinical trials and other translational research endeavors.

**Selected Publications:**


