

## **Emory Genetics: Bridging the Gap from Research to Clinical Care**

Since 1972, Emory Genetics has provided a vast array of clinical, research, and laboratory services. The current Department of Human Genetics was founded in 2001 as part of the Emory University School of Medicine, from the restructuring of the previously distinct medical and academic genetic departments.

The DOHG has a vibrant clinical program, organized under the Division of Medical Genetics. The Division holds numerous general and specialized genetic clinics for children and adults in the Atlanta area with several outreach clinics throughout Georgia. The Division is also the referral center for Georgia's newborn screening program. Clinical management of newborns identified with metabolic diseases is provided by the Nutrition Section within the Department.

### **Nutritional Program for Metabolic Patients**

Each year, geneticists, nutritionists, and genetic counselors from Emory are responsible for the follow-up of nearly 5000 abnormal newborn screening tests and the treatment of over 250 children with metabolic disorders. Emory Genetics provides follow-up testing and management guidelines for the Newborn Screening Program of Georgia. The nutritional program is administered by Rani H. Singh, Ph.D. R.D., and is one of the nationally leading programs offering complete services to manage metabolic disorders. Our program also includes an annual week-long metabolic camp for young women with phenylketonuria (PKU) and maple syrup urine disease (MSUD); [www.metcamp.org](http://www.metcamp.org).

### **Research**

Advances in genomic science will revolutionize patient care in the coming years. The Department of Human Genetics is committed to translating major research discoveries quickly into patient care. We are currently seeking individuals to participate in research directed toward the genetics of Maple Syrup Urine Disease as well as many other genetic conditions.

### **Emory Genetics Laboratory**

Emory Genetic Laboratory provides cytogenetic, biochemical and molecular diagnostic testing, with special emphasis in molecular cytogenetics, rare disease testing, and comprehensive testing for newborn screening follow-up. Please visit our website for a complete test list: [www.genetics.emory.edu](http://www.genetics.emory.edu).

### **Specialty Tests in Organic Acidemias include:**

- **Methylmalonic Aciduria (MMA) mutase deficiency:**
  - **Sequencing of the MUT gene**  
EGL is the only clinical laboratory in the US to offer molecular genetic testing for MMA. Sequence analysis of the MUT gene is offered to patients to confirm a biochemical diagnosis of MMA. If mutations are identified in the patient, carrier testing and prenatal diagnosis is available to family members.
  - EGL is currently developing a biochemical test to measure the activity of the mutase enzyme, which would allow for fully comprehensive testing for MMA patients, as well as facilitating prenatal diagnosis.
  
- **Maple Syrup Urine Disease (MSUD):**

EGL is the only clinical laboratory in the US to offer comprehensive biochemical and molecular genetic testing for MSUD

- **Sequencing of Branched-Chain  $\alpha$ -Keto Acid Dehydrogenase (BCKD) Complex Genes:** Sequence analysis of the three genes that form the BCKD gene complex is offered to patients with a biochemical diagnosis of MSUD. If mutations are identified in the patient, carrier testing and prenatal diagnosis is available to family members.
  - **BCKD enzymatic assay for MSUD:** Biochemical testing to measure the activity of the BCKD enzyme is used to clinically diagnose MSUD and to facilitate prenatal diagnosis.
- EGL also offers comprehensive biochemical and molecular testing for:
    - Galactosemia
    - Phenylketonuria (PKU)

Bookmark our website, [www.genetics.emory.edu](http://www.genetics.emory.edu), to check for updates on testing, clinical services, and research studies.