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**Medical Genetics**

*The 2016 Storybook Ball will showcase the incredible strides MGHfC physicians and researchers have made in the diagnosis and treatment of genetic disorders and the ability to test for genetic outcomes.*

The Genetics and Metabolism Program at MGHfC is at the forefront of revolutionary advances in genetics that will help us better understand the basis of many health conditions and deliver more personalized care. State-of-the-art genetics laboratories, world-wide collaborations, and research trials ensure patients have access to the latest testing and treatment options.

**Clinical Programs**

*Below is a brief snapshot of the clinical programs and research initiatives at MGHfC.*

**General Genetics Clinic**

The Genetics Program at MGHfC specializes in the evaluation of patients with illnesses that have a suspected genetic cause. The program performs many services including pre-conceptual and prenatal counseling for:

* couples carrying a baby at risk or suspected to have a genetic condition
* newborns with birth defects
* children with failure to thrive
* children with neurological problems such as seizures
* individuals with developmental delays or loss of skills
* adults with a family history of an adult-onset genetic condition

The program also performs consultations for patients of all ages when there is a suspected genetic contribution to their symptoms. These patients are often referred to the clinic by specialists in many disciplines including neurology, cardiology, endocrinology, ophthalmology, nephrology, orthopaedics and oncology. A clinical team comprised of physicians, genetic counselors, post-graduate physicians in training, social workers and a dietician provide families with diagnosis, treatment, genetic counseling and follow-up care in a supportive environment.

**Metabolic Clinics**

The Metabolic Program cares for individuals with a large variety of inborn errors in metabolism including mitochondrial disorders and lysosomal storage disease. Many of these disorders are detected during newborn screening while others may not develop until adulthood. Referrals are often made for individuals with complex medical conditions to determine if there is a contributing metabolic disorder. In certain cases, specialized diets or enzyme replacement infusions are needed to maintain health. Complex coordinated care with a medical team including counselors, therapists and a nutritionist is available to optimize their medical care.

**Specialty Clinics**

MGHfC is home to a large number of specialty clinics focusing on rare and complex genetic disorders. A few examples of our specialty clinics include:

* Down Syndrome Program
* Pitt Hopkins Clinic
* 22q11.2 Deletion Syndrome Clinic
* Turner Syndrome Clinic
* Williams Syndrome Clinic
* Lurie Center for Autism
* Hearing loss clinic

**Genetic Psychiatry**

Mass General has a robust genetic psychiatry program that includes the [Psychiatric & Neurodevelopment Genetics Unit (PNGU)](http://www.massgeneral.org/psychiatry/research/pngu_home.aspx). The PNGU is focused on psychiatric and neurodevelopmental disorders including Gilles de la Tourette syndrome (GTS), Obsessive Compulsive Disorder (OCD), Attention Deficit Hyperactivity Disorder (ADHD), autism and related disorders, reading disabilities (such as dyslexia), and bipolar affective disorder.

**Research Initiatives**

The Medical Genetics and Metabolism Program has a number of areas of active research:

* **Use of whole genome and exome sequencing to diagnose rare and complex genetic disorders**

New technology allows us to sequence every gene in the body. Judicious use of these powerful new genetic tests is helping to solve many diagnostic dilemmas.

* **Genetic, Phenotypic and Biologic Sample Repository for Rare Genetic Diseases**

This repository, which includes detailed genetic and phenotypic data along with biological samples, is facilitating collaborations with researchers at Mass General, the Broad Institute and around the world to discover new genetic diseases and better understand and treat known diseases.

* **Down syndrome** is an especially active area of research:
	+ 1.) Several clinical trials are underway with new medications that hold promise to improve cognition and behaviors in those with Down syndrome

 2.) We are developing reliable predictors of Obstructive sleep apnea to avoid sleep studies with monitoring devices that are often poorly tolerated in those with Down syndrome

3.) Creation of a mobile app to improve eating options for people with Down syndrome

* **Williams syndrome** – Individuals with this syndrome classically have unique psychological as well as physical and metabolic issues. Active research is underway to determine factors contributing to anxiety, characterization of fat distribution and glucose metabolism, skin and vessel elasticity. An active Patient and Clinical Research Registry created and maintained by MGHfC keeps track of Williams syndrome patients from all over the world.
* **Congenital diaphragmatic hernia** –This is an often fatal disorder where the diaphragm and lungs fail to properly develop. Using innovative strategies to study samples from patients around the world, researchers in our program are discovering causes of this rare disorder.
* **Characterization of novel regulators of cancer, bone metabolism and immunity –** Researchers have discovered a set of proteins that act as potent gatekeepers to prevent cancer development and also are critical for controlling bone mineralization as well as excessive inflammation. The study of how these proteins work is suggesting new potential treatments for these disorders.

**Undiagnosed Disease Network**

A network of seven medical centers across the country, including Mass General, has been established by the National Institutes of Health to discover the cause of rare and new diseases in individuals whose diagnosis has defied extensive clinical investigations. In collaboration with Brigham and Women’s Hospital and Boston Children’s Hospital, Mass General is tapping unprecedented clinical expertise and cutting-edge technologies to investigate the pathophysiology of these new and rare diseases in children and adults. This information will then be shared across institutions to better identify options for treatment and patient care.

To learn more about these and other programs and research initiatives or for information about supporting these efforts,

please contact Tracie DeGuglielmo (617-643-6779 or TDeGuglielmo@partners.org).

[www.massgeneral.org/children](http://www.massgeneral.org/children)