

## Biochemical Genetics Program

### Our Team Members

**Maryam Banikazemi, M.D.**, is a biochemical geneticist and Assistant Professor of Clinical Pediatrics in the Division of Clinical Genetics, Department of Pediatrics. Her clinical work focuses on inborn errors of metabolism and the timely diagnosis and treatment of patients with these disorders. Dr. Banikazemi is nationally and internationally recognized for her work in the field of lysosomal storage disorders. Her patient-oriented clinical research and active involvement with investigations of new developing therapies have contributed to several advances in this field.

**Alfred E. Slonim, M.D.**, is a pediatric endocrinologist and Professor of Clinical Pediatrics in the Division of Clinical Genetics, Department of Pediatrics, and a national and international expert in rare metabolic diseases of muscle and liver. Dr. Slonim has extensive experience in the investigation and understanding of the metabolic disturbances that occur in inborn errors of metabolism and how the body attempts to compensate for these specific enzyme deficiencies. Information from these studies enables our physicians to devise therapies that compensate for the end product deficiency or minimize the accumulation of toxic metabolites that develop as a result of enzyme deficiencies.

Dr. Slonim's primary clinical research interest is in the investigation and treatment of disturbances in intermediary metabolism that involve genetic diseases of muscle, liver, and the gastrointestinal tract. Therapies that have been developed for these diseases consist mainly of nutritional, exercise, and hormonal means of compensating for the metabolic disturbances.

In addition to Drs. Banikazemi and Slonim, the Biochemical Genetics Program team includes a metabolic nurse, registered dietician, social worker, and genetic counselor. Clinical specialists include gastroenterologists, cardiologists, neurologists, nephrologists, orthopedic surgeons, ophthalmologists, pulmonologists, and transplant surgeons.

## Information and Referral

Referrals to the Biochemical Genetics Program at Morgan Stanley Children's Hospital can be made by physicians, and other health and allied healthcare providers. Patients, parents and guardians may also contact the program directly. For more information, or to make an outpatient referral, please call **(212) 305-6731**.

To arrange for an inpatient transfer, please call **1-800-NYP-STAT**.

### Biochemical Genetics Program

Morgan Stanley Children's Hospital  
of NewYork-Presbyterian  
6th Floor North, Room 601A  
3959 Broadway  
New York, NY 10032

Morgan Stanley Children's Hospital is easily accessible by car, bus, and subway. The main entrance, with full wheelchair and stroller accessibility, is located on the northwest corner of Broadway and West 165th Street.

For more information about Morgan Stanley Children's Hospital, please visit [www.childrensnyp.org](http://www.childrensnyp.org).

# Biochemical Genetics Program

*Offering Diagnostic and  
Treatment Services for  
Metabolic Disorders*



## The Biochemical Genetics Program

at Morgan Stanley Children's Hospital of NewYork-Presbyterian and Columbia University offers diagnostic and management services for children and adults with confirmed or suspected inborn errors of metabolism. Multidisciplinary teams of physicians and healthcare professionals specializing in these inherited disorders provide comprehensive care, including surveillance and management of symptoms and clinical manifestations.

Early and accurate diagnosis and detection is an essential first step for the appropriate care of individuals born with these complex, often multisystem disorders, giving them the opportunity to build healthy lives, improve their quality of life, and minimize disease burden.

Common reasons for referral include:

- positive newborn screen
- failure to thrive
- developmental delay
- hypoglycemia
- liver failure
- cardiomyopathy in children
- myopathy

## Our Services

Comprehensive services offered by the Biochemical Genetics Program include evaluation, diagnosis, treatment, and reproductive planning for all metabolic disorders. Treatment plans are tailored to each condition. Components of treatment may include:

- dietary management with a metabolic nutritionist
- cofactor supplementation
- enzyme replacement therapy
- organ transplantation including bone marrow, liver, kidney, and heart transplants

The Biochemical Genetics Program also offers patients the opportunity for enrollment in cutting-edge clinical trials and research studies for many of these rare disorders.

We are equipped to manage metabolic emergencies and accept transfers for the acute management of metabolic decompensation; liver, renal, or cardiac failure; and transplant evaluation.

*Dr. Alfred E. Slonim is a nationally and internationally recognized authority in metabolic muscle diseases, glycogen storage diseases, and inflammatory bowel diseases.*



*Dr. Maryam Banikazemi is one of the world's foremost experts on Fabry disease and other lysosomal storage disorders, a group of more than 40 rare genetic diseases that disrupt cell function.*

## Metabolic Disorders

Inborn errors of metabolism comprise a large class of genetic diseases involving disorders, including, but not limited to:

**Amino acid metabolism**—phenylketonuria and tyrosinemia

**Carbohydrate metabolism**—galactosemia and hereditary fructose intolerance

**Fatty acid oxidation**—medium-chain acyl-CoA, dehydrogenase deficiency (MCAD), and carnitine palmitoyl transferase-II (CPT-II)

**Glycogen storage diseases**—Pompe disease, McArdle disease, Von Gierke disease, and Cori disease

**Lysosomal storage disorders**—Gaucher disease, Fabry disease and Mucopolysaccharidosis

**Mitochondrial disorders**—MELAS (mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke) syndrome and MERRF (myoclonic epilepsy associated with ragged red fibers) syndrome

**Neurotransmitter disorders**

**Organic acidurias**—propionic acidemia and methylmalonic acidemia

**Peroxisomal biogenesis disorders**

**Urea cycle defects**—ornithine transcarbamylase