Inherited Metabolic Diseases Program (IMDP)

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About Us

The Inherited Metabolic Diseases Program (IMDP) at AUBMC is the first and only comprehensive program for inherited metabolic disorders in Lebanon and other countries in the region. We provide clinical evaluation, metabolic diagnostic investigations and treatment for babies, children, adolescents and adults with inherited metabolic diseases (also known as inborn errors of metabolism). During the past 16 years, the program has diagnosed and treated more than 1700 patients suffering from IMD.

Our team of leading clinical specialists such as Dr. Pascale Karam and Mrs. Abir Barhoumi, dietitian, put their in-depth knowledge and long years of experience into actual practice in the field of inborn errors of metabolism. The department offers state-of-the-art comprehensive joint care for IMDP patients.

As part of our mission, the IMDP can recommend performing diagnostic metabolic tests for those patients that require them at AUBMC in the Department of Pathology and Medicine Laboratory.

IMDP provides a multidisciplinary approach to care by collaborating with other highly specialized programs within different departments at AUBMC (such as neurology, psychology, cardiology, ophthalmology) to provide all IMD patients with a multidisciplinary approach.

Our Mission

To provide the highest standards of care including clinical evaluation, metabolic diagnostic investigations and treatment for babies, children, adolescents and adults with inherited metabolic diseases (also known as inborn errors of metabolism).

Services and Procedures

We have all the required expertise to diagnose and treat a wide variety of inherited metabolic diseases such as:

- Inherited metabolic disorders detected by newborn screening.
- Amino acid disorders, such as: phenylketonuria, tyrosinemia, urea cycle defects, maple syrup urine disease, homocystinuria, etc.
- Organic acid disorders, such as: methylmalonic acidemia, propionic acidemia, isovaleric acidemia, etc.
- Mitochondrial diseases.
- Lipid acid disorders/Fatty acid oxidation defects.
- Lysosomal storage disorders.
- Wilson's Disease.

Frequently asked Questions:

What are Inherited Metabolic Diseases?

Metabolism refers to a series of chemical reactions that occur inside our cells to produce energy for the body and transform one chemical into another. Inherited Metabolic Diseases (also known as inborn errors of metabolism) are genetic diseases caused by the disruption of a metabolic pathway in the body thus leading to serious health problems.

What might a metabolic disorder lead to?

- Accumulation of toxic substances in different important organs like the brain, heart, liver, bones, etc. and/or lack of energy production.
- Defect in the body's energy utilization.

What are the symptoms of Inherited Metabolic Diseases?

Inherited Metabolic Diseases can cause a variety of symptoms.

- Neurological: psychomotor delay, myopathy, neuropathy, epilepsy, intellectual disability, autism, etc.
- Digestive: persistent nausea and vomiting, protein or sugar aversion, chronic diarrhea, etc.
- Cardiac: cardiac muscle dysfunction, hypertrophy or dilatation.
- Vision disturbance: like retinal deposits, bilateral cataract, etc.
- Hearing deficit.
- Bone diseases: like rickets, scoliosis, and bone deformities.
- Liver diseases: like acute or chronic dysfunction, cirrhosis.
- Failure to thrive: poor weight gain, short stature.

At what age can IMD start?

Symptoms of IMD can start at any age! It can affect babies, children, adolescents and even adults.

How can we diagnose an IMD?

The IMD specialist can diagnose an IMD based on the below criteria:

- **In newborns:** when a baby has a positive neonatal screening, which is a blood test that can be offered at birth to all babies.
- **At any age:** when a patient comes forward with one or more suggestive symptoms of IMD.

After a thorough clinical evaluation, the IMD specialist will ask for specific metabolic investigations. These tests can include mainly highly specialized blood and urine metabolic tests which can be performed at the AUBMC Clinical Chemistry Laboratory.

How is IMD treated?

Some IMD can be treated by having the patient follow a diet restricting the toxic substance that cannot be metabolized by the body and/or providing vitamins and medications that can help the metabolism function properly.
Management of IMD is provided by a skilled IMD team including the clinical specialist and sometimes the IMD Dietitian.

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Location and Appointment Scheduling

The Inherited Metabolic Diseases Program is located in Wassef and Souad Sawwaf Building, 3rd floor. To schedule an appointment, please call 01 - 350 000 ext. 7523.
Email: imdp@aub.edu.lb

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