SHAPING THE FUTURE OF MITOCHONDRIAL MEDICINE

Mitochondrial Medicine at Children's Hospital of Philadelphia (CHOP) is the world's premiere center for individualized clinical care, advanced diagnostics and innovative therapies for children and adults with complex and rare diseases that stem from mitochondrial energy deficiency. Pairing clinical care with extensive research efforts, we're paving the way to improving the health and well-being of people living with mitochondrial disease.

CHALLENGES OF MITOCHONDRIAL DISEASE

On average, patients can have up to 16 unique symptoms, making mitochondrial disease difficult to diagnose. It can impair the functions of different organs in different people and cause a variety of conditions and diseases that share impaired energy production. Because of its complex and difficult-to-measure nature, it's often misdiagnosed.

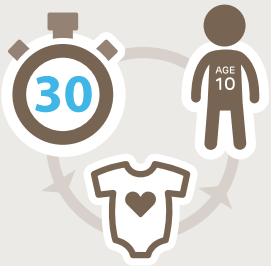
PERSONALIZED DISEASE MANAGEMENT

Our dedicated team is focused on determining the best course of treatment for each patient while working in collaboration with each patient's primary care physician, neurologist and other specialists. Although there is no cure for mitochondrial disease, patients can be well managed by a personalized approach that considers changes in diet and nutrition, exercise, and vitamin or amino acid supplements. We also help people navigate challenges they may face supporting a family member with mitochondrial disease.

TESTING FOR MITOCHONDRIAL DISEASE

While we already know more than 350 genes in which pathogenic mutations may cause mitochondrial disease, we are continually working toward new discoveries to understand the precise genetic cause in each individual and to create precision testing and treatments. Today, there are several ways to diagnose mitochondrial disease:

- Genomic diagnostic testing in blood or salivia from patients and family members, including mitochondrial DNA genome analysis
- Genetic or biochemical tests in affected tissues, such as muscle or liver
- Blood or urine tests to identify biochemical markers of disease cause, severity or treatable problems
- Tissue biopsies done through surgery or in the clinical setting
- Radiology-based imaging tests



Every 30 minutes, a child is born who will develop a mitochondrial disease by age 10.

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Mitochondrial Medicine

continued >

OUR TEAM

The Mitochondrial Medicine team is made up of a core group of specialized physicians with expertise in mitochondrial disease and multiple subspecialists, which include:

- Anesthesiologists
- Clinical and Biochemical Geneticists
- Cardiologists
- Clinical Care Coordinators
- Critical Care Specialists
- Diagnostic Pediatricians
- Dietitians
- Dysautonomia Specialists
- Endocrinologists
- Exercise Physiologists
- Financial Counselors
- Gastroenterologists
- Genetic Counselors
- Immunologists
- Nephrologists
- Neurologists
- Neuroradiologists
- Neurosurgeons
- Ophthalmologists
- Patient Navigators
- Physical Therapists
- Pulmonologists
- Social Workers
- Surgeons

OUR COMMITMENT

Mitochondrial Medicine has been designated a Frontier Program by Children's Hospital. Frontier Programs are unique, cutting-edge programs that will forge important new discoveries, deliver novel therapies, and help even more children and adults thrive. CHOP allocates additional resources to Frontier Programs to aid their growth and reach.

DISCOVERY IS ON THE HORIZON

Researchers and clinicians at CHOP are also working every day to advance the understanding of mitochondrial disease. We have a dedicated research team focused on investigating mitochondrial and epigenomic dysfunction and developing new therapies for a wide range of clinical problems.

With more than 350 different gene disorders causing different mitochondrial diseases and many more to be discovered, there's still more to be done. Our researchers are working to identify additional disease causes, develop more precise tests for diagnosis and personalize treatments.

Because mitochondria are involved in many complex processes, hundreds of different conditions can be caused by genetic mutations or disruptions in how mitochondria function.

PARTNER WITH US
To refer a patient or request a second opinion:
267-426-6298
CHOPUSA@email.chop.edu

LEARN MORE
chop.edu/mito

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Mitochondrial Medicine

MITOCHONDRIAL MEDICINE TEAM

LEADERS AND PIONEERS IN MITOCHONDRIAL MEDICINE

Marni Falk, MD Executive Director of Mitochondrial Medicine and attending physician: A board-certified clinical geneticist who has directed the mitochondrial medicine program at CHOP since 2006, Falk oversees an extensive translational research laboratory and integrated clinical research program to improve care, deepen mitochondrial disease knowledge and interdisciplinary collaborations, and realize precision medicine by identifying effective treatments for each mitochondrial disease patient.

Rebecca Ganetzky, MD, attending physician and Biochemical Laboratory Test Director: Ganetzky, a board-certified clinical and biochemical geneticist, joined our faculty in 2017. Her clinical and research efforts focus in complex biochemical forms of mitochondrial disease, including complex V deficiency and pyruvate dehydrogenase complex deficiency, among others. She also is Director of the CHOP Biochemical Genetics Fellowship Program.

Amy Goldstein, MD, Clinical Director and attending physician: A board-certified pediatric neurologist with extensive experience in mitochondrial disease clinical care and clinical trials, Goldstein provides our inpatient consult service, coordinates the Mitochondrial Medicine multidisciplinary clinical care conferences and is physician lead on several CHOP Mitochondrial Medicine clinical trials.

Shana McCormack, MD, attending endocrinologist and clinical research lead: McCormack provides clinical management of endocrine manifestations of mitochondrial disease, and leads an active clinical research program to develop improved radiologic, exercise and nutritional diagnostic assessments and therapeutic interventions for mitochondrial disease.

Eiko Nakamaru-Ogiso, PhD, Research Laboratory Director: A basic biochemist and biophysicist with extensive expertise in mitochondrial biology and enzymatic analyses, Nakamaru-Ogiso oversees a highly collaborative team of research scientists, post-doctoral fellows and students working to develop improved models, diagnostic tests and therapies for primary mitochondrial disease.

Douglas C. Wallace, PhD: A pioneer and internationally prominent scientist in the field of human mitochondrial genetics, Wallace came to Philadelphia in 2010 to launch and lead the Center of Mitochondrial and Epigenomic Medicine (CMEM) within the CHOP Research Institute.

Zarazuela Zolkipli Cunningham, MBChB, MRCP, attending physician and clinical researcher: Zolkipli-Cunningham cares for patients of all ages with mitochondrial myopathies and oversees performance of clinical needle muscle biopsies under local anesthesia. She leads an integrated clinical and research team of clinicians, exercise physiologists and physical therapists to better define and manage mitochondrial myopathy, including exercise intolerance, and directs clinical investigations to develop new devices and interventions for mitochondrial myopathy.

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