

Mitochondrial Evaluation at UPMC Children's Hospital of Pittsburgh

Thank you for contacting the certified Mitochondrial Medicine Center (MMC) at UPMC Children's Hospital of Pittsburgh and for your interest in an evaluation. We look forward to meeting you.

Prior to your visit, we will review your medical record. If you have been evaluated by a specialty physician outside of the UPMC health system or completed genetic testing or other studies related to the reason for referral, these records must be sent to our office. All new patients require a physician referral, and all medical records must be received to our office prior to the scheduling an appointment.

Please provide the following information to schedule a new patient appointment:

- Patient's full name and date of birth
- Patient's address and phone number
- Parent and/or guardians full name (if patient is a minor)
- Name of referring physician and physician's office phone number
- Primary care physician (PCP) and office phone number
- Physician referral made to the Mitochondrial Medicine Center at UPMC Children's

Medical records should include the following, if applicable:

- Physician referral and office note describing the referral
- All genetic testing completed to date
- Office notes from specialists, including:
 - Neurological evaluation(s)
 - Clinical Genetics evaluation(s)
 - Ophthalmologic evaluation(s), including electroretinogram (ERG)
 - Audiological evaluation(s)
 - Cardiology evaluation(s), including echocardiogram and electrocardiogram (EKG)
 - Gastrointestinal evaluation(s), including biopsy reports
 - Other specialist evaluations
- Records from prior hospitalizations and/or surgeries
- Reports of imaging studies within the last 2 years, including:
 - Brain MRI and/or MRS (spectroscopy)
 - Echocardiograms (written report only)
 - Ultrasounds
- Reports of prior biochemical studies within the last 2 years on blood or urine, including:
 - Plasma amino acid analysis
 - Urine organic acid analysis
 - Lactate and/or pyruvate level
 - Carnitine analysis: acylcarnitine profile, total carnitine, free carnitine
 - Creatine kinase
 - Comprehensive metabolic panel (CMP), complete blood count with differential (CBC)
 - Other routine blood and/or urine studies
- Reports of prior tissue studies, including any additional studies completed on these tissues:
 - Muscle biopsy, skin biopsy, and/or liver biopsy

Office Contact Information:

- Office Phone: 412-692-5070; to schedule an appointment select option 1
- Office Fax: 412-692-6472
- Email: mitomed@chp.edu

If sending medical records by mail, please put to the attention of:

- Mitochondrial Medicine Center, Division of Genetic and Genomic Medicine
UPMC Children's Hospital of Pittsburgh
4401 Penn Ave, Faculty Pavilion Suite 1000
Pittsburgh, PA 15224

Please be prepared to spend up to 2-3 hours at your initial appointment. During your visit, our team will review your medical and family history, perform a detailed physical examination, and provide counseling and recommendations for biochemical and/or genetic laboratory testing. Insurance prior authorization may be required for some recommended tests. A follow-up visit may be necessary to discuss testing results and/or additional recommendations. Thank you!