MISSION

Our mission is to provide cutting edge medical care to individuals affected by neurometabolic and mitochondrial disorders, to perform both clinical and basic science research to provide treatment options for patients with mitochondrial disease, and to educate medical professionals and the community about mitochondrial disease.

The UT Mitochondrial Center of Excellence is a multidisciplinary team of physicians, along with nurses, social workers, researchers, a patient liaison and special programs coordinator, and an administrative and accounting team. We provide comprehensive services to aid in the diagnosis and management of neurometabolic and mitochondrial disorders in both adult and pediatric patients. We consult with patients, their primary care physician or pediatricians, and subspecialists to develop an individualized long-term treatment and management plan. We also perform research in the field of neurometabolic and mitochondrial disorders and investigate novel treatment approaches in these devastating diseases.

The UT Mitochondrial Center of Excellence operates under the direction of Dr. Mary Kay Koenig, Associate Professor of Pediatrics and Neurology at the University of Texas Health Science Center at Houston, McGovern Medical School.
We are proud to be the host of seven different clinical studies here in our mitochondrial center.

Erydel S.p.A is sponsoring two studies:

1. **EDS in Ataxia Telangiectasia (ATTesT).**
   “This is an international, multi-center, one-year, randomized, prospective, double-blind, placebo-controlled, phase III study, designed to assess the effect of two non-overlapping dose ranges of EDS EP, administered by IV infusion once per month, on neurological symptoms of patients with Ataxia Telangiectasia.”

2. **Open-label, Long-term, Extension Treatment Using Intra-Erythrocyte Dexamethasone Sodium Phosphate in Patients With Ataxia Telangiectasia Who Participated in the IEDAT-02-2015 Study (OLE-IEDAT).**
   “This is an international (North America, Europe, Africa, Asia and Australia), multi-center, prospective, open-label treatment study, designed to continue to provide the study medication to all patients who completed 12 months of treatment (including those treated with placebo) in the IEDAT-02-2015 trial, completed the study assessments, do not present safety contraindication to continuation of treatment, and provided informed consent. The study aims to collect information on the long-term safety and efficacy of the trial treatment.”

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**NEW TEAM MEMBERS**

Brunessia Lewis  
Social Worker

Saba Iqbal  
Research Coordinator

Lindsey Miller  
Research Nurse

Kenny Cuevas  
Administrative Support

Stephanie Maldonado  
Research Coordinator

Netsuhe Mengesha  
Research Nurse

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**THE BEGINNING**

Over 10 years ago, Dr. Mary Kay Koenig was faced with a challenge. She and Dr. Ian Butler met and diagnosed a patient with MELAS (mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes). Dr. Koenig’s conclusion was to treat the patient, since the patient’s condition had been diagnosed. When Dr. Koenig spoke to Dr. Butler about treatment, she discovered that no treatment existed and this patient would not live. Dr. Koenig decided this was unacceptable and set out to find a way to treat MELAS. In 2006, she attended her first ever meeting with the United Mitochondrial Disease Foundation (UMDF) as her first steps to find a treatment for MELAS.”
Stealth BioTherapeutics Inc. is sponsoring two studies:

1. A Trial to Evaluate the Safety and Efficacy of Elamipretide in Subjects With Primary Mitochondrial Myopathy Followed by an Open-Label Extension (MMPOWER-3)

   “This is a multicenter phase 3 randomized, double-blind, parallel-group, placebo-controlled trial to evaluate the safety and efficacy of daily subcutaneous injections of elamipretide in subjects with primary mitochondrial myopathy. This will be followed by an open-label treatment extension.”

2. An Observational Study of Patients With Primary Mitochondrial Disease (SPIMM-300) (RePOWER)

Retrophin Inc.:

Efficacy and Safety Study of Fosmetpantotenate (RE-024) in PKAN Patients (PKAN)

“This study will investigate whether fosmetpantotenate (RE-024), a phosphopantothenate replacement therapy, is safe and effective in treating patients with Pantothenate Kinase-Associated Neurodegeneration (PKAN).”

Ultragenyx Pharmaceutical Inc.:

Compassionate Use of Triheptanoin for LIPT1-related disease.

BioElectron (formerly Edison Pharmaceuticals)

EPI-743 for Mitochondrial Respiratory Chain Diseases

“This study evaluates the safety and efficacy of EPI-743 in patients with severe mitochondrial respiratory chain diseases who are considered to be within 90 days of end-of-life care.”

Metabolomics Profile in ABAT Deficiency Pre- and Post-treatment. Koenig MK, Bonnen PE. PMID 29480352.

The Expanding Neurological Phenotype of DNM1L-related Disorders. Wangler MF, Assia Batzer N, Robak LA, Koenig MK et al. PMID 29529134.

Idiopathic Central Nervous System Inflammatory Disease in the Setting of HLA-B27 Uveitis. Crowell EL, Pfeiffer ML, Kamdar AA, Koenig MK et al. PMID 29652210.

Efficacy and Safety of Topical Rapamycin in Patients with Facial Angiofibromas Secondary to Tuberous Sclerosis Complex: The TREATMENT Randomized Clinical Trial. Koenig MK, Bell CS, Hebert AA, Roberson J et al. PMID 29800048.


IRF2BPL is associated with neurological phenotypes. Marcogliesse PC, Shashi V, Spillmann RC, Stong N et al. PMID 30057031.

A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. Mucha BE, Banka S, Ajeawung NF, Moldpere S et al. PMID 30245510.

**SUPPORTING ORGANIZATIONS**

**PALS**
People Against Leigh Syndrome (PALS) was formed in 2013 by the friends of William Martin, a Houstonian diagnosed with a progressive and often fatal form of a mitochondrial disease called Leigh syndrome. PALS seeks to build awareness of this little disease and support efforts that would lead to discover a treatment or a cure. Currently, PALS is working in partnership with the Children's Memorial Hermann Hospital and the UT Mitochondrial Center of Excellence.

http://peopleagainstleighs.org/

The PALS Leigh Syndrome Registry is an international database of patients with Leigh syndrome. This project was developed by the UT Mitochondrial Center of Excellence, with the assistance and support of People Against Leigh Syndrome (PALS), the United Mitochondrial Disease Foundation (UMDF), and the Mitochondrial Medicine Society (MMS). The registry launched in the summer of 2015 and has received overwhelming support from the mitochondrial community. There are over 20 mitochondrial disease specialists and support organizations assisting with information dissemination and international recruitment. We have patients enrolled throughout the US, Japan, Canada, Europe, and Australia and our registry continues to expand. In uniting the medical and patient communities of Leigh syndrome, we gain vital information needed to develop a cure. Each participant who registers allows us to gather meaningful data which connects patients to clinical trials, distributes information to physicians, and brings awareness to Leigh syndrome. As more individuals are diagnosed with Leigh syndrome, it is vital for clinicians and patients to promote global research. The registry can be accessed from the People Against Leigh Syndrome website: www.peopleagainstleighs.org

**UMDF Run/Walk**
The United Mitochondrial Disease Foundation will be hosting their annual run/walk. The event will be April 13, 2019 at Sugar Land Memorial Park. For more information feel free to check http://www.energyforlifewalk.org/

**Viva La Vida**
Save the Date! The Fourth Annual People Against Leigh Syndrome (PALS) Fundraiser - ¡Viva la Vida! - will be held on Wednesday, May 15, 2019 at Saint Arnold Brewing Company. Chaired by Elizabeth and James Elder, the event is an opportunity for friends and supporters to enjoy time together, learn more about Leigh syndrome, and discover the latest research being conducted in the fight against this progressive and fatal form of mitochondrial disease.

Funds raised at the event support the UT Mitochondrial Center of Excellence at Children's Memorial Hermann Hospital, as well as the only international patient registry database for Leigh syndrome patients.

Email peopleagainstleighsyndrome@gmail.com for information about sponsorship opportunities and tickets.

**MMS Care Network**
Founded in 1998 by doctors Richard Haas and Robert Navaiax, the MMS represents an international group of physicians, researchers and clinicians working towards advancing education, research and global collaboration in clinical mitochondrial medicine.

http://www.mitosoc.org/

In 2018 the MMS developed a process of recognizing mitochondrial care networks clinics. We are proud to announce that the Mitochondrial Center of Excellence is recognized as one of these clinics.

**Mito Action**
MitoAction is a nonprofit organization founded by patients, parents, and Boston hospital healthcare leaders who had a vision of improving quality of life for children and adults with mitochondrial disease. The organization began in 2005 as an idea and has evolved from a small New England support group to a dynamic, active service organization helping thousands of patients and families. Despite the growth of the organization, the mission remains the same: to make a measurable impact in the lives of those who are affected by mitochondrial disease.

http://www.mitoaction.org/

**UMDF**
To promote research and education for the diagnosis, treatment, and cure of mitochondrial disorders and to provide support to affected individuals and families.

https://www.umdf.org/