

# Fulfilling the UMDF Mission

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

## UMDF RESEARCH



- **Over \$11 Million in Research**

Since 1996, UMDF funded projects hoping to make new gains in the diagnosis and treatment of mitochondrial disease.

### 2017 Highlights

- **Mitochondrial Medicine 2017: Washington, DC**

Over 600 attending – the largest group to date. Attendees were patients and families, clinicians and researchers from the US and around the world.

- **External Meetings Expand Outreach**

The National Society for Genetic Counseling and Child Neurology Society Annual meetings attracted more than 3000 clinicians. The awareness, networking, and educational outreach will feed the UMDF Roadmap Initiatives in the years to come!

## UMDF EDUCATION



## UMDF DIAGNOSIS/TREATMENT



- **Collaborating To Optimize Clinical Care**

Developed the framework with partner organizations to develop a Mitochondrial Care Network that launches in 2018.

- **Mitochondrial Disease Community Registry Grows**

Patient populated registry increases by 40% to over 2,000 registrants in 2017.

- **Patient/Family Support**

In 2017, UMDF conducted more than 50 patient/family meetings across the country, bringing medical experts directly to patients.

- **UMDF Connect**

UMDF provided help and support to more than 4,100 patients and families online and over the phone, connecting them to resources, support, clinicians, and most importantly – answers.

## UMDF SUPPORT

**UMDF  
connect**  
Resources for help & support.



## UMDF ADVOCACY



- **UMDF Advocacy Impacts Research Dollars**

Fighting for inclusion in the Congressionally Directed Medical Research Program resulted in the allocation of more than \$11 million in funding for mitochondrial disease research projects.

- **Day on the Hill Collaboration**

The event was billboarded as a 'community' activity resulting in the scheduling and completion of over 400 House and Senate visits.

# 2018 and Beyond

## ROADMAP TO A CURE

The Roadmap to a Cure focuses on three pillars for better patient outcomes.

### DIAGNOSIS

Genetic testing presents an excellent opportunity to achieve a specific diagnosis, but less than half of all cases are successfully diagnosed by genetic testing. Other testing methods include muscle biopsies, fibroblasts or buccal swabs. Brain imaging, exercise physiology and various lab measurements of mitochondrial function show some promise. In general, the pathway to diagnosis is not standardized.

**UMDF wants to create a better diagnostic scenario for mitochondrial disease patients, there is a clear need to broadly identify and characterize patients based on health information, genetic testing and biosamples.**



### THERAPEUTIC DEVELOPMENT

There are no licensed therapies for mitochondrial disease in the United States. Numerous investigator-initiated trials have been conducted, but in general, there is a notable absence of well-controlled studies within the field. Industry sponsored clinical trials are rapidly increasing in number.

**Our role is to coordinate stakeholders in academia, government and the drug development industry to address important topics such as validated outcome measures, patient-report outcomes and regulatory guidance. These steps are necessary in gaining treatments and cures for mitochondrial disease more efficiently and quickly.**



### PATIENT CARE

Mitochondrial disease patients receive care from a relatively small number of knowledgeable specialists. The many different types of mitochondrial disease and the many symptoms associated with each challenge even the most knowledgeable of doctors. The result is clinical care that is often inconsistent. Additionally, insurance reimbursement for rare disease care is an increasingly challenging situation.

**UMDF's goal is to take advantage of a national focus on "personalized medicine" affording an opportunity for the mitochondrial disease community to help develop the programs and tools that will advance optimized patient care in the 21st century. We are committed to collaboration that leads to standards of care and Centers of Excellence models**

