

# United Mitochondrial Disease Foundation

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

## About UMDF Grants

The United Mitochondrial Disease Foundation (UMDF) is committed to finding cures for mitochondrial illness and recognizes that innovative and cutting-edge research is the path to success. Yet we also realize that new, aggressive research for rare disorders is often not underwritten by federal grants. We seek to fund research and training grants that support the mission of the Foundation.

### **Proposals fitting one of the following categories will be considered:**

- Post-doctoral and clinical training fellowships in mitochondrial genetic disease
- Seed money for new researchers in the field of mitochondrial genetic disorders
- New area of research for experienced investigators

**Research Priorities:** The UMDF has established the following research priorities for its research grant program:

#### **1. Basic Science and Research**

- Develop animal models of nuclear and mitochondrial DNA mutations.
- Develop methods to transform mitochondria with exogenous DNA.
- Support basic biology that would lead to a treatment of genetic mitochondrial disease.
- Understand better the transmission and segregation of mitochondria and mitochondrial DNA.
- Develop new approaches to mitochondrial biology using cellular and animal systems.
- Encourage new basic scientists to enter the field.

#### **2. Clinical Research**

##### **(a) General**

- Understand the natural history of specific mitochondrial diseases.
- Understand epidemiology (e.g. frequency and, where appropriate, ethnicity, resistant populations, disparity).
- Develop common clinical and laboratory definitions.
- Understand effects of stress on mitochondria and on patients with mitochondrial disease.
- Understand effects of exercise and diet on mitochondria and on patients with mitochondrial disease.
- Develop clinical-biochemical correlations.
- Create an international longitudinal database on the natural history of mitochondrial diseases.

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- Develop improved genetic counseling, as well as research required to develop the information base to provide accurate information about prognosis and recurrence risk.
- Encourage new clinical scientists to enter the field.

### (b) Treatment

- Develop and test strategies leading to extended life span, improved quality of life, and cure for mitochondrial disease.
- Support clinical trials for mitochondrial disease using appropriate, objective outcome criteria.
- Understand the response to, and side effects (including psychological and economic effects) of, therapeutic strategies, including exercise, diet, and pharmacological treatments of mitochondrial disease (including individually and combined elements of mitochondrial "cocktails").
- Develop strategies to shift mitochondrial heteroplasmy
- Develop and use objective outcome criteria, including biochemical, clinical, and radiological correlations.

### (c) Diagnosis

- Develop quality control strategies and common definitions leading to diagnostic criteria for mitochondrial disorders and standards for all diagnostic testing, including imaging.
- Develop rapid and accurate detection of nuclear and mtDNA mutations; accurate detection of heteroplasmy.
- Develop functional/physiological diagnosis and monitoring strategies (e.g. exercise, MRS, whole body testing), focusing on the least invasive methods of diagnosis and monitoring mitochondrial disorders.
- Explore the pros and cons of testing using fresh or frozen samples.
- Develop a micropolarographic method for diagnosing mitochondrial disease.

### Review Process:

Letters of Intent are reviewed and ranked by the UMDF Scientific and Medical Advisory Board. Investigators providing Letters of Intent of greatest interest and scientific value for projects in conformance with UMDF mission goals will be invited to present a formal proposal. Formal proposals are reviewed by the Grant Review Committee and recommendations made to the Board of Trustees regarding funding. Each award is disbursed over the one, two or three year life of the grant, according to the terms of the grant.

### Letters of Intent:

The UMDF must receive electronic Letters of Intent by the September 17<sup>th</sup> deadline. Incomplete Letters of Intent or those received past the deadline will not be considered. FAX transmittal is not acceptable.

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Letters of Intent will be ranked with respect to the “Points of Review” listed below. Our desire is to subsequently invite grant applications only for those projects which the UMDF would possibly fund.

Points of Review (Letters of Intent):

- Viability of the project.
- Likelihood of subsequent NIH funding for this project.
- Impact on patient care or understanding of mitochondrial medicine.
- Likelihood applicant will make an impact in the field and how this project may help.
- Conformance with UMDF research goals.
- Post-doc Fellowship: Mentor’s knowledge, resources and drive to assist the fellow toward the project goals.
- Clinical Research: Likelihood of enlargement of the scale of the study.
- Clinical Research: Impact on patient care or understanding of mitochondrial medicine.
- Clinical Research and Basic Science: Applicant’s experience and achievements.

### Formal Proposals:

Investigators whose Letters of Intent pass the initial review phase will be invited to submit a formal proposal. Solicited proposals are due in mid-February of the next calendar year.

Scoring Criteria: Formal grant proposals will be scored based on the following criteria:

1. **Significance/Impact:** The extent to which the project, if successfully carried out, will make an original and important contribution to the detection, diagnosis, treatment, management, and family care of mitochondrial disease patients.
2. **Approach:** The extent to which the conceptual framework, design, methods, and analyses are properly developed, well integrated, and appropriate to the aims of the project.
3. **Feasibility:** The likelihood that the investigators can accomplish the proposed work. Consideration will be given to the investigator’s documented experience and expertise, past progress, preliminary data, requested and available resources, institutional commitment, and (if appropriate) documented access to special reagents or technologies and adequacy of plans for the recruitment and retention of subjects.