



*HOPE. ENERGY. LIFE.*

***MEDIA KIT***



## UMDF Mission

Our mission is to promote research and education for the diagnosis, treatment and cure of mitochondrial disorders and to provide support to affected individuals and families. We are committed to increasing funding for mitochondrial disease research now – to alleviate the suffering of thousands who have this debilitating and often fatal disease and to develop better understanding of and treatments for the many common illnesses and chronic conditions associated with it.

## Research

Understanding mitochondrial disease has the potential to open a new world of knowledge, transforming medicine and cutting across all medical disciplines to uncover better treatments for a range of serious diseases. As the largest non-governmental funder of mitochondrial disease research, UMDF has already achieved tremendous success in marshalling resources for new research and improving awareness and understanding of mitochondrial disease. In the last decade, UMDF and its Scientific Advisory Board of experts have awarded an unparalleled \$6 million in grants to the most promising mitochondrial disease research proposals—leading to important new discoveries. To learn more about UMDF research efforts visit [www.UMDF.org](http://www.UMDF.org).

## Family Support

UMDF creates caring, supportive communities across the nation for adults, children and families suffering from mitochondrial disease through our local chapters and affiliated groups and ambassadors. This allows UMDF members to network with other families and individuals to talk about mitochondrial disorders. UMDF keeps members updated with the latest treatment advances and information through a quarterly newsletter.

## Information and Communication

UMDF is committed to answering patients', policymakers' and the medical community's questions about mitochondrial disease – including important details about diagnosis, wide-ranging symptoms and existing treatment.

Each year, UMDF hosts the largest international research symposium dedicated to mitochondrial disease where the world's leading researchers meet with doctors, patients and their families to exchange valuable knowledge and ideas. UMDF also strives to build awareness of mitochondrial disease among physicians and healthcare providers by bringing the world's experts to them through its "Grand Rounds Program." Grand Rounds bring world renowned mitochondrial disease specialists directly to affected patients and families in their own communities.

UMDF advocates on the national level and empowers members to advocate on the local level regarding issues of medical funding and health-related issues that impact those who are affected and their families. In addition, UMDF provides information about local fundraisers and educational programs and maintains [UMDF.org](http://UMDF.org) to provide the latest news and information about issues relating to mitochondrial disease research.



## Overview

We need to increase funding for mitochondrial disease research now in order to alleviate the suffering of thousands who have this debilitating and often fatal disease and to develop better understanding of and treatments for the many common illnesses and chronic conditions associated with it.

## What is a mitochondrial disease?

Mitochondria are present in every single cell in our bodies – except for red blood cells – and are responsible for producing more than 90 percent of the energy needed by the body to sustain life and support growth. Mitochondrial diseases result from the failure of these tiny “powerhouses.” When the mitochondria fail, less and less energy is generated within our cells. Cell injury and even cell death follow. As this process repeats itself throughout the body, whole systems begin to fail and the life of the person in whom this is happening is severely compromised. Mitochondrial diseases can affect any organ of the body at any age. They are extremely complex and the range of symptoms include:

- Progressive muscle weakness
- Cardiac disease
- Liver disease
- Diabetes
- Gastrointestinal disorders
- Blindness and deafness
- Strokes and seizures
- Susceptibility to infections

## Incidence and prevalence

Every 30 minutes, a child is born with a mitochondrial disease. According to official records, each year between 1,000 and 4,000 children in the U.S. are born with a mitochondrial disease and suffer from severe and devastating symptoms for which there is no cure and no effective treatment. However, because the disease is complicated and difficult to diagnose, the actual number of children born with the disease is thought to be much higher. Most patients suffer symptoms for months or often years before they are accurately diagnosed with a mitochondrial disease. The mortality rate among children varies from 10 to 50 percent per year. Many children do not survive beyond their teen years.

## Research demonstrates that mitochondrial diseases are not rare.

The research, conducted by a Dr. Patrick Chinnery at Newcastle University in the United Kingdom, shows that **1 in every 200** people have a DNA mutation that could potentially cause a mitochondrial disease in them or their offspring.

### Links to other diseases

Defects in mitochondrial function are at the core of many common diseases and conditions, including autoimmune diseases. Therefore, research into mitochondrial disease could open a new avenue to treatments for these other diseases and conditions.

Mitochondrial dysfunction is implicated in:

- Alzheimer's disease
- Parkinson's disease
- Diabetes
- Hypertension
- Heart disease
- Osteoporosis
- Cancer
- Multiple sclerosis
- Lupus
- Rheumatoid arthritis
- Aging

### Success to date

The United Mitochondrial Disease Foundation and private researchers have already achieved tremendous success in marshalling resources to fund new research and improve awareness and understanding of mitochondrial disease. In the last decade, UMDF has awarded an unparalleled \$8 million in grants to the most promising researchers, leading to important new discoveries.

For instance, scientists recently discovered that inherited Parkinson's disease is caused by a gene that disables the cell's mitochondria, a finding which is paving the way for more effective treatments for that disease. Research also uncovered a direct cause and effect relationship between mitochondrial dysfunction and common age-related diseases like heart disease. The National Institutes of Health also recently recognized the importance of mitochondrial disease research by considering the disease for an NIH Roadmap for Medical Research.

### Why more funding is required

We need to increase funding to alleviate the suffering of thousands who have this debilitating and often fatal disease, and to develop better understanding of and treatments for the many common illnesses and conditions associated with it. Understanding mitochondrial diseases has the potential to transform medicine and improve the diagnosis of and treatments for a range of diseases which affect millions of Americans.



### Mitochondrial defects are a central factor in human health and disease

Defects in mitochondrial function are at the core of many common diseases and conditions. Therefore, research into mitochondrial disease could open a new avenue to treatments for these other illnesses and chronic conditions.

“Mitochondrial genome” refers to the entire collection of mitochondrial DNA (mtDNA) and nuclear DNA (nDNA) genes that are involved in the structure, function, assembly, regulation and turnover of the mitochondrion.

Variation in either a nDNA or a mtDNA gene that influences mitochondrial function will define an individual’s initial energy capacity. Over time, additional mtDNA mutations accumulate which further erode function. Ultimately, the combination of the inherited energy defect and the age-related acquisition of additional mtDNA mutations push the individual’s energy generating capacity below the minimum threshold resulting in symptoms called aging or age-related diseases.

- Until recently, the relationship between mitochondrial dysfunction and a wide range of disease states was known to exist, but whether mitochondrial dysfunction was responsible for the particular disease was still in question. For the first time, Dr. Douglas C. Wallace, University of California, Irvine, proved that a mitochondrial energy gene mutation can in fact cause age-related heart and muscle disease in the mouse.

Mitochondrial dysfunction is a significant component in the etiology of a surprising range of very common illnesses and conditions, and research into mitochondrial dysfunction offers a promising new avenue for their treatment.

- The illnesses include Alzheimer’s disease, Parkinson’s disease, Huntington Disease, Amyotrophic Lateral Sclerosis (ALS), mental retardation, deafness and blindness, diabetes, obesity, cardiovascular disease and stroke. Over 50 million people in the U.S. suffer from these chronic degenerative disorders. While it cannot yet be said that mitochondrial dysfunction causes these problems, it is clear that mitochondria are involved because their function is measurably disturbed.
- Even autoimmune diseases such as multiple sclerosis, Sjogrens syndrome, lupus and rheumatoid arthritis appear to have a mitochondrial basis to illness.
- Mitochondrial dysfunction has been associated with a wide range of solid tumors, proposed to be central to the aging process, and found to be a common factor in the toxicity of a variety of physical and chemical agents.

Therefore, research into basic mitochondrial physiology and mitochondrial diseases offers hope to the thousands who suffer from this debilitating disease and to the millions who are afflicted with these other common conditions and diseases.



### **What is mitochondrial disease?**

Mitochondria exist in nearly every cell of the human body, producing 90 percent of the energy the body needs to function. In a person with mitochondrial disease, the mitochondria are failing and cannot adequately convert food and oxygen into life-sustaining energy. For many, mitochondrial disease is an inherited genetic condition, while for others the body's mitochondria can be affected by environmental factors.

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### **How does mitochondrial disease affect the body?**

The parts of the body that need the most energy, such as the heart, brain, muscles and lungs, are the most affected by mitochondrial disease. The affected individual may have strokes, seizures, gastro-intestinal problems, (reflux, severe vomiting, constipation, diarrhea), swallowing difficulties, failure to thrive, blindness, deafness, heart and kidney problems, muscle failure, heat/cold intolerance, diabetes, lactic acidosis, immune system problems and liver disease.

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### **What symptoms could an undiagnosed individual exhibit?**

The child or adult may have seizures, severe vomiting, failure to thrive, heat/cold intolerance, muscle weakness, delayed achievement of milestones, severe diarrhea/constipation, feeding problems, unable to fight typical childhood infections or repeated infections and fevers without a known origin. A "red flag" for mitochondrial disease is when a child or adult has more than 3 organ systems with problems or when a "typical" disease exhibits atypical qualities.

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### **What is the prognosis for these individuals?**

Currently, there are no treatments for children and adults with mitochondrial disease. Although some of the affected children and adults are living fairly normal lives with the disease, many people are severely affected, and many children do not survive their teenage years. More research dollars are needed to find more effective treatments and ultimately a cure for mitochondrial diseases.

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### **When is someone with mitochondrial disease at the highest risk?**

The child or adult is at highest risk for neurological and organ damage during and for the two weeks following an illness. Therefore even a simple flu or cold virus can have devastating effects on the patient, even death. Any illness must be treated immediately with medical interventions, like IV fluids and IV antibiotics.

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### How many individuals are affected?

Each year between 1,000 and 4,000 children in the U.S. are born with a mitochondrial disease. While exact numbers of children and adults suffering from mitochondrial diseases are hard to determine because so many people who suffer are frequently misdiagnosed, researchers believe the disease is approaching the frequency of childhood cancers. Many are misdiagnosed with atypical cerebral palsy, various seizure disorders, childhood diseases and diseases of aging. Still others aren't diagnosed until after death.

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### Can adults have mitochondrial disease?

Yes, many adults are diagnosed with adult-onset mitochondrial disease. Some of these individuals have been ill their whole lives but were undiagnosed. Others have carried the genetic mutation that causes mitochondrial disease since birth but did not show any symptoms until triggered by an illness. Adult mitochondrial disease patients are affected in a similar manner to that of children.

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### What is the United Mitochondrial Disease Foundation (UMDF)?

The UMDF was created in 1996 by parents of affected children. The UMDF exists to promote research and education for the diagnosis, treatment and cure of mitochondrial disorders and to provide support to affected individuals and families. The national headquarters of the UMDF is in Pittsburgh, and there are 14 Chapters and 22 Mito Groups<sup>SM</sup> across the United States.

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### How does UMDF support research to find a cure?

As the largest non-governmental funder of mitochondrial disease research, UMDF has already achieved tremendous success in marshalling resources for new research and improving awareness and understanding of mitochondrial disease. In the last decade, UMDF and its Scientific Advisory Board of experts have awarded an unparalleled \$8 million in grants to the most promising mitochondrial disease research proposals—leading to a range of invaluable new breakthroughs. The UMDF's scientific and medical advisory board is composed of mitochondrial disease experts from around the world, and these experts select which research grants receive funding. The UMDF is considered the premier foundation for mitochondrial disease by the world medical community.

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### How can I reach UMDF for more information?

To speak to someone at UMDF's national office, please call 1-888-317-UMDF (8633) or email [info@umdf.org](mailto:info@umdf.org). To find your local Chapter, Mito Group<sup>SM</sup> or Ambassador visit [www.UMDF.org](http://www.UMDF.org) and go to our map of events and activities.