



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.

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.

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.

What is the prognosis for these individuals?

Currently, there are no effective 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.

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.

How many individuals are affected?

Every 30 minutes, a child is born who will develop a mitochondrial disease by age 10. While exact numbers of children and adults suffering from mitochondrial diseases are hard to determine because so many people who suffer are frequently misdiagnosed.

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.

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, PA.

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. Since 1996, UMDF has awarded nearly \$12 million in grants to the most promising mitochondrial disease research proposals—leading to a range of invaluable new breakthroughs.

What is the ‘Roadmap to a Cure’?

The Roadmap to a Cure focuses on three pillars for better patient outcomes. The “pillars”, detailed below, are DIAGNOSIS, THERAPEUTIC DEVELOPMENT and PATIENT CARE. 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 bio samples. 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. 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.

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 connect@umdf.org .