

FREQUENTLY ASKED QUESTIONS

ABOUT MITOCHONDRIAL DISEASE AND THE UNITED MITOCHONDRIAL DISEASE FOUNDATION

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 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 other 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, poor muscle tone, 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?

As more research dollars are raised to find more effective treatments and ultimately a cure, some of the affected children and adults are living fairly normal lives with mitochondrial disease. At the opposite end of the spectrum, many are severely affected, and some children do not survive their teenage years.

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 15 minutes, a child is diagnosed with a mitochondrial disease, or will develop one by the age of 10. While exact numbers of children and adults suffering from mitochondrial disease are hard to determine because so many people who suffer from mitochondrial disease are frequently misdiagnosed, we now know 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.

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 went undiagnosed. Others have carried the genetic mutation that causes mitochondrial disease since birth but did not show any symptoms until a severe illness brought them on. Adult mitochondrial patients are affected in a similar manner to the kids who are affected.

MITOCHONDRIAL DISEASE IS THE BODY'S INABILITY TO TURN FOOD INTO THE ENERGY TO SUSTAIN LIFE.

WHAT IS THE UNITED MITOCHONDRIAL DISEASE FOUNDATION (UMDF)?

The UMDF was created in 1996 by parents of affected children. The Foundation 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 UMDF is in Pittsburgh, and there are 11 chapters and 15 Mito GroupsSM across the United States.

HOW DOES UMDF SUPPORT RESEARCH TO FIND A CURE?

In 2007, UMDF funded more than \$1 million in research. UMDF is one of the leading contributors of grants to mitochondrial disease researchers and is considered the premier foundation for mitochondrial diseases by the world medical community. UMDF's scientific and medical advisory board is composed of mitochondrial disease experts from around the world, and these experts approve which research grants receive funding. UMDF has made a commitment to raise over \$15 million for research by 2009.

HOW CAN I REACH UMDF FOR MORE INFORMATION?

Visit the UMDF website at www.umdf.org, or contact us at 1-888-317-UMDF or info@umdf.org.



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