UNITED MITOCHONDRIAL DISEASE FOUNDATION®

HOPE. ENERGY. LIFE.

What are mitochondria?

Mitochondria are often called the 'cell's powerhouse.' They are specialized compartments within almost every cell. They are responsible for producing 90% of the energy needed by our body to sustain life. Mitochondria combine oxygen from the air we breathe with calories from food to produce energy.

What is mitochondrial disease?

Mitochondrial diseases result when there is a defect that reduces the ability of the mitochondria to produce energy. As the mitochondria fails to produce enough energy, the cell will not function properly and if this continues, cell death will eventually follow. Organ systems will begin to fail and the life of the individual is compromised, changed or ended.

Imagine a major city with half its power plants shut down. At least, such conditions would produce a "brown out" with large sections of the city working far below optimum efficiency. Now imagine your body working with one-half of its energy-producing facilities shut down. The brain may be impaired, vision may be dim, muscles may twitch or may be too weak to allow your body to walk or write, your heart may be weakened, and you may not be able to eat and digest your food. This is precisely the situation people with mitochondrial disease find themselves.

Mitochondrial disease can affect any organ of the body and at any age. Symptoms are extremely diverse and often progressive. They include: strokes and seizures, muscle weakness, gastrointestinal disorders, swallowing difficulties, cardiac disease, liver disease, diabetes, blindness and deafness and susceptibility to infections.

What causes mitochondrial disease?

For most patients, there is a genetic mutation in either the mitochondrial DNA or the nuclear DNA. The mutation may have been inherited from the mother or from both parents, or it may represent a spontaneous mutation. For most patients with mitochondrial disease, the genetic mutation has not yet been identified.

There are environmental factors, even certain medicines that may interfere with the mitochondria and result in symptoms.

How common are mitochondrial diseases?

Every 30 minutes a child is born who will develop a mitochondrial disease by age 10. At least one in 200 individuals in the general public have a mitochondrial DNA mutation that may lead to disease. Mitochondrial disease is a relatively newly diagnosed disease – first recognized in an adult in the 1960s and in the 1980s for pediatric onset cases. It is greatly under diagnosed and the true prevalence is difficult to determine.

Mitochondrial Disease Fact Sheet

Research has consistently shown that mitochondrial dysfunction is at the core of many very common illnesses and chronic conditions of adulthood. These include: Alzheimer's Dementia, Parkinson's disease, diabetes, hypertension, heart disease, osteoporosis, cancer and even the aging process itself. Furthermore, autoimmune disease such as multiple sclerosis, lupus, and rheumatoid arthritis appear to have a mitochondria basis to illness.

Why is research so critical?

There are no known treatments or cures for mitochondrial disease.

Mitochondria may play a far greater role in human health than scientists and doctors have realized. Any health concern that is an energy problem could be related to the mitochondria.

Further research into the mitochondrion and primary mitochondrial diseases (those due to genetic defect) would benefit millions of people. It would offer hope to the thousands suffering from this debilitating and often fatal disease and provide a broad range of new therapeutic approaches for attempting to treat these other very common and incapacitating illnesses and conditions.

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. The UMDF has awarded an unparalleled $10 million in grants to the most promising researchers, leading to important new discoveries.

For instance, scientists 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.