



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

Mitochondrial dysfunction is at the core of a surprising range of very common illnesses and conditions, and a promising new avenue for their treatment. As the mitochondria are responsible for producing energy, any illness that has an energy problem could be related to the mitochondria. Diseases in which mitochondrial dysfunction have been implicated include:

- Alzheimer's Dementia, 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 US suffer from these chronic degenerative disorders. While it cannot yet be said that mitochondrial defects cause 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.

## Highlights in Research

Until recently, the broad range of diseases that may be caused by mitochondrial dysfunction was not well understood or appreciated. A 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. This changed with the discovery that mutations of the mitochondrial DNA could cause certain diseases. For the first time, scientists showed that a single nucleotide change in mitochondrial DNA of a mouse led to the development of muscle weakness and progressive heart disease.

Research supporting the link between mitochondrial dysfunction and some of these other common illnesses includes:

- Mitochondrial coenzyme Q10 levels are reduced in patients with Parkinson's disease and mitochondrial function in these patients is impaired.
- Results of the first placebo-controlled clinical trial of the compound coenzyme Q10 suggest that it can slow disease progression in patients with early-stage Parkinson's disease.
- These findings are consistent with another recent study involving patients with early onset Huntington's disease. These patients showed slightly less functional decline in groups receiving coQ10.
- Investigators believe coQ10 works by improving the function of the mitochondria.

- A drug once approved as an antihistamine in Russia improved thinking processes and the ability to function in Alzheimer's disease patients. The drug works by stabilizing mitochondria.
- Cancers are also associated with defects in the mitochondria. Within the cell, signaling must occur between the mitochondria and the nucleus. When the signaling malfunctions, the defect can cause cancer.
- Researchers discovered that mutations in the mitochondrial DNA may play a role in tumor metastasis and suggests a possible new avenue for the development of a treatment to suppress metastasis.
- Researchers have found a very consistent decline in mitochondrial function that is found in diabetes and pre-diabetes.
- There is increasing interest in the possibility that mitochondrial dysfunction might play an important role in the etiology of autism. A subset of autistic children have already been shown to manifest biochemical alterations that are commonly associated with mitochondrial disorders, and a few have been linked to specific alterations in the mitochondrial genes.

It is clear that research into mitochondrial disease offers hope to the millions who are afflicted with these other common conditions and diseases.