

MITOCHONDRIAL DISEASE INFORMATION FOR TEACHERS

Definition

Mitochondrial diseases are a group of progressive metabolic, often neurological, disorders that result from defects in the mitochondria, which are in almost every type of cell in the body. Mitochondria are responsible for creating more than 90% of the energy needed to sustain life and support growth. Mitochondrial failure results in energy deprivation within the cells. Cell injury and even cell death follow. If this process is repeated throughout the body, whole systems begin to fail, and the affected person's life is severely compromised. The disease primarily affects children, but adult onset is becoming increasingly common.

Diseases of the mitochondria appear to cause the most damage to organs requiring high energy levels including: brain, heart, liver, skeletal muscles, kidney, eyes, and the endocrine and respiratory systems.

Mitochondrial disorders are notorious for not following a set pattern. It is not uncommon for patients to first be misdiagnosed with another disorder. Symptoms can begin at any age. Depending on which cells are affected, symptoms may include: loss of motor control, muscle weakness and pain, gastro-intestinal disorders and swallowing difficulties, poor growth, cardiac disease, liver disease, diabetes, respiratory complications, seizures, visual/hearing problems, lactic acidosis, developmental delays, autism, and susceptibility to infection.

Mitochondrial disorders are not always physiologically obvious. Intelligence can range from gifted to profoundly retarded. Children with fewer affected mitochondria may be mislabeled as lazy, uncooperative, or underachievers. At the beginning of the school day, for example, they may appear energetic and focused. But as the day progresses, their small reserve of energy evaporates and they are unable to complete the same tasks as their classmates. These children may also be labeled as mildly retarded because of their slowness, when in reality they may be merely fatigued.

Etiology

More than 1 in 4,000 children born each year in the US will develop a mitochondrial disease by age 10. Because they are under-recognized, this figure may be grossly under-estimated. Most cases are inherited, the majority of which are probably through autosomal recessive inheritance (both parents contributing a defective gene). Another mode of transmission is maternal inheritance, which may be as common. There are other rarer modes of inheritance and even some sporadic cases in which the mutation is a new one. Males and females are equally affected.

Development and Symptoms

There is no one identifying feature of mitochondrial disease. Patients can have combinations of problems whose onset may occur from before birth to late adult life. Typically, the earlier the onset of symptoms, the more severe the disease. Symptoms can be progressive and may include, but are not limited to, combinations of the following: seizures, developmental delay, regression, movement disorders,

migraines, strokes, cardiac problems, hearing deficit, eye muscle dysfunction and visual loss, diabetes, GI problems, temperature irregularities, fatigue, failure to gain weight, mental retardation, autism, behavior problems, and dementia. Siblings with the same defect can have differing levels and types of symptoms.

Three “rules of thumb” generally characterize mitochondrial disorders:

- 1) A “common disease” (such as diabetes) has atypical features
- 2) Three or more organ systems are involved
- 3) Recurrent setbacks or flare-ups of the disease occur with acute infections

Treatment and Mortality

At present, there are no cures for mitochondrial disorders and no truly effective treatments. The goals of current treatments are: to alleviate symptoms, and to slow down the progression of the disease.

General therapies often include supplements such as CoQ10 and vitamin “cocktails” which may enhance enzyme activity within the mitochondria, and act as antioxidants. Effectiveness of such treatments is largely anecdotal, noticeably helping some and not others. As a rule, those with mild disorders tend to respond better than those with severe disorders. While effectiveness may not be definable, these treatments may delay the progression of the disorder.

Prescription medications may also be necessary to control such symptoms as seizures, heart irregularities, and secondary diabetes. Physical therapy, speech therapy, and other types of therapies may also be utilized.

Some typical considerations regarding treatment of mitochondrial patients include the following:

- 1) Standard therapies for specific symptoms may be necessary.
- 2) Dietary – some mitochondrial patients may be on strict diets; others may not. Avoidance of fasting is critical; small, frequent meals may be necessary. Patients with GI and swallowing difficulties may require gastric tube feedings.
- 3) Vitamins and supplements are usually prescribed.
- 4) Treatments must be tailored to the individual, often resulting from trial and error by the physician.
- 5) Avoidance of physiologic (external) stress factors. Those with autonomic symptoms (inability to control involuntary functions such as temperature control and heart rate) may require a more controlled temperature environment. Over- or under-heating may trigger a medical crisis.
- 6) Avoidance of exposure to illness – even minor acute illnesses, such as colds, may cause a medical emergency, resulting in hospitalization.

Mitochondrial disorders are generally progressive and may be fatal. However, due to the individuality of symptoms and the differences in specific mitochondrial disorders, it is sometimes difficult even for physicians to predict a prognosis. Some patients live into adulthood. However, earlier and more severe symptoms generally result in a shorter life span.

Specific Mitochondrial Disorders

There are numerous disorders which fall under the classification of mitochondrial diseases, and more are being discovered all the time. Some of the more common names include the following:

Mitochondrial myopathy
Mitochondrial encephalomyopathy
Leigh's disease/syndrome
Mitochondrial encephalomyopathy, lactic acidosis and strokelike episodes (MELAS)
Kearns-Sayre syndrome (KSS)
Myoclonus epilepsy with ragged red fibers (MERRF)
Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE)
Neuropathy, ataxia and retinitis pigmentosa (NARP)
Chronic progressive external ophthalmoplegia (PEO or CPEO)
Complex I, II, III, IV or V (or combination of complexes)
Carnitine Deficiency
Lactic Acidosis
Leber's hereditary optic neuropathy (LHON)
Fatty oxidation disorders (FODs)

For more information:

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