

What are Mitochondrial Myopathies?

Mitochondrial Myopathies are a group of diseases that affect the mitochondria, the tiny energy factories found inside almost all cells. These diseases interfere with the function of muscles. Because mitochondria occur in all cells in the body, mitochondrial myopathies can also interfere with the function of other organs in the body. The disease group is comprised of Kearns-Sayre syndrome (KSS); Leigh's syndrome; mitochondrial DNA depletion syndrome (MDS); mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes (MELAS); myoclonic epilepsy with ragged red fibers (MERRF); mitochondrial neurogastrointestinal encephalopathy syndrome (MNGIE); neuropathy, ataxia and retinitis pigmentosa (NARP); Pearson syndrome; and chronic progressive external ophthalmoplegia (CPEO).

Symptoms and progression differ with each type.

What are Symptoms of Mitochondrial Myopathies?

In general, there is nervous system impairment, eye problems, hearing problems, cardiac abnormalities, skeletal muscle abnormalities, and disorders of the gastrointestinal tract. Some symptoms can be so mild that they're hardly noticeable, while others are life-threatening. Muscle weakness, muscle pain, fatigue, lack of endurance, poor balance and difficulty in swallowing may be experienced.

What is the age of onset?

The age varies according to disease.

What causes Mitochondrial Myopathies?

A defect exists in either a mitochondrial gene or a gene in the cell nucleus that affects the functioning of the mitochondria.

Is this disease inherited?

In general, if the defect is in a mitochondrial gene, inheritance is from the mother only. If the defect is in a nuclear gene, it may be inherited from either the mother or father. Some of the mitochondrial myopathies are sporadic, meaning that the abnormal gene only occurs in the affected person. It was not inherited from a parent and will not be passed on to children.

How are Mitochondria Myopathies diagnosed?

A physician evaluates the extent of the symptoms by taking the personal medical history, and then proceeds with the physical and neurological exams. Diagnostic tests may include tests of strength and endurance, such as an exercise test, which can involve

activities like repeatedly making a fist, or climbing up and down a small flight of stairs. The neurological exam can include tests of reflexes, vision, speech and basic cognitive (thinking) skills.

Depending on the results of the above tests, a muscle biopsy may be done. This is usually the most definitive way to diagnose a mitochondrial myopathy. Abnormalities can be seen in the muscle mitochondria under the microscope. As well, genetic testing can be performed on muscle DNA.

If a mitochondrial myopathy is suspected or confirmed, there are many other non-invasive tests that may be done to determine the extent of organ involvement as this varies from patient to patient. These tests may include CT scans, MRI scans, EEG's (to monitor brain wave activity) and ECG's and echocardiograms (to monitor the heart's activity). Blood tests are usually done to assess blood counts, liver and kidney function.

None of the frequent symptoms of mitochondrial disease such as muscle weakness, exercise intolerance, hearing impairment, ataxia, seizures, learning disabilities, cataracts, heart defects, diabetes and stunted growth are unique to mitochondrial myopathies. However, a combination of three or more of these symptoms in one person strongly points to mitochondrial myopathy, especially when the symptoms involve more than one organ system. always the case. People with CCD should be considered potentially susceptible to MH unless proved otherwise by a special type of muscle biopsy, which screens for MH.

What Treatments are available for Mitochondrial Myopathies?

Unfortunately, there is no curative therapy yet for any of the mitochondrial myopathies. However, supportive treatment is available which can modify

some of the symptoms and this can be quite helpful for many patients. Again, the symptoms show a great deal of individual variability in both type and severity.

Some examples of treatment:

If there is hearing impairment, it can be managed using hearing aids.

Many of the other potential symptoms can be managed with medications (i.e. headaches, heart problems, seizures).

If drooping of the eyelids is causing impaired vision, it can be corrected by surgery or by using specialized glasses.

If there is a loss of muscle strength in the arms or legs, braces, a cane or a wheelchair may be required. It is usually recommended that you be assessed by a physical or occupational therapist to determine which supportive aids would be best in your situation.

If there is weakness in the face and neck with slurred speech and/or difficulty with swallowing, speech therapy or changing the diet to easier-to-swallow foods can be useful.

If respiratory care is needed to support breathing, mild respiratory problems might require occasional respiratory support, such as pressurized air, while someone with severe problems might require permanent support from a ventilator.

Supplements : Creatine, L-carnitine and coenzyme Q10 are supplements often combined into a "cocktail" for treating mitochondrial diseases. Although there is little scientific evidence that the treatment works, some patients report minor improvement in symptoms.

Overall, treatments for any of these diagnoses must also involve people with these disorders taking care of themselves by monitoring their health and scheduling regular medical exams.

Special Considerations

If surgery is being considered, it is important to inform doctors and anaesthetists that you have a neuromuscular disorder, as there is an increased risk of anesthetic complications in patients with neuromuscular diseases. This risk can be minimized if the anesthetist knows of your diagnosis. A medical alert bracelet specifying your diagnosis and the potential risk of adverse reactions to anesthesia is recommended.

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