PMM: Primary Mitochondrial Myopathy

Primary mitochondrial myopathy, also known as PMM, is a group of genetic conditions with similar features that cause a variety of signs and symptoms in many parts of the body, particularly those that use a lot of energy, like muscles and the brain. Mitochondria are parts of a cell that help turn the energy we get from food into energy that the body can use. They are also important in the communication between body parts and in creating other materials the body needs.

It is important to note that other mitochondrial conditions like MELAS or MERRF can also present with myopathy (muscle weakness). People with these conditions usually have additional clinical features.

Alternative Names

- PMM

Cause and Genetics

PMM is a group of genetic conditions caused by changes, sometimes called mutations or variants, in many genes. Both males and females can have PMM. PMM can either be passed down in a family (inherited) or can be the result of a new, sometimes called de novo, genetic change in a baby.

PMM can be due to changes in either the nuclear DNA (or the set of DNA present in every cell of the body and inherited from both parents) or the mitochondrial DNA (or the set of DNA contained in the mitochondria of a cell). The way the condition is passed down in families will depend on the exact gene involved for the person with PMM.

Frequency

Primary mitochondrial myopathy is thought to affect around 30,000 people in the United States. PMM may be misdiagnosed or underdiagnosed.

Signs and Symptoms

PMM, by definition, mainly affects the skeletal muscles. These include muscles that work with bones, tendons, and ligaments such as the shoulder muscle or abdominal muscles. The body also has other types of muscles including cardiac muscles (lining the walls of the heart) and smooth muscles (which line the insides of other organs like the stomach). While most PMM only affects the skeletal muscles, some affected individuals may have additional health concerns in other parts of the body.

Signs and symptoms of primary mitochondrial myopathy may include:

- Exercise intolerance (pain and/or fatigue after exercise)
- Fatigue or a feeling of extreme tiredness
- Muscle weakness and cramping
- Nausea
- Headache
- Breathlessness

PMM is generally progressive, meaning it gets worse over time. PMM can start at any age, though "more severe" presentations usually occur at a younger age. Most occur before the age of 20. The exact features, onset, and severity can vary widely among people with this condition, even among members of the same family. Always check with your provider if new symptoms appear or you are concerned.

Diagnosis

Primary mitochondrial myopathy can be diagnosed by:

- Measuring biochemical markers in blood and urine
- Testing a sample from the muscle (biopsy)
- Careful physical examination and exercise studies
- Performing a genetic test

PMM is not included on newborn screening panels. If there is a known family history of a PMM, prenatal testing can be performed on amniotic fluid or chorionic villi. Results interpretation of this testing for mitochondrial conditions is complicated and genetic counseling is recommended.

Treatment and Management

Before beginning any treatment or therapy, please consult with your physician.

As of 2022, there is currently no FDA-approved therapy for any PMM.

Treatment and management of PMM are symptomatic and supportive. This may include:

- Physical and occupational therapy
- Avoidance of mitochondrial toxins like certain drugs, tobacco, and alcohol
- Mitochondrial supplements like riboflavin, coenzyme Q, and carnitine

Clinical Trials

For specific details on other clinical trials, visit the MitoAction Clinical Trials page or www.clinicaltrials.gov.

Resources

- Mitochondrial Myopathy Fact Sheet – National Institute of Neurological Disorders and Stroke
- Primary Mitochondrial Myopathies – National Organization for Rare Disorders

Connecting with others who are impacted by a rare disease allows for important information to be shared about day-to day life, prevents isolation, and gives hope. Please contact MitoAction for peer support opportunities at 888-MITO-411 or email mito411@mitoaction.org. Other resources we recommend are:

- New Patient Kit for Mitochondrial Conditions
- Planning and Preparation
- Monthly Expert Series
- Energy in Action Podcast

MitoAction does not provide medical advice, diagnosis, treatment, or legal advice. It is essential that all those living with or caring for someone with a Mitochondrial or FAOD disease have an emergency protocol letter. These letters, which are written and signed by a doctor, share details about prescribed treatment during crises and in emergency room settings. Always check with your doctor if you or your child has concerns as everyone may present with symptoms differently. Before beginning any treatment or therapy, please consult with your physician.