MELAS: Mitochondrial Myopathy Encephalopathy Lactic Acidosis and Stroke-Like Episodes

Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-Like Episodes Syndrome, also known as MELAS, is a rare mitochondrial condition where the body is not able to make enough mitochondrial proteins needed to make energy. 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 creating other materials the body needs. Mitochondrial conditions can 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.

Alternative Names

- MELAS
- MELAS syndrome

Cause and Genetics

Affected genes: MT-ND1, MT-ND5, MT-TH, MT-TL1, MT-TV

MELAS is an inherited genetic condition, meaning it is passed down in a family. MELAS is caused by changes in the mitochondrial genome (mtDNA) or set of DNA contained in the mitochondria of a cell. Mitochondria are inherited from the mother through her eggs. People typically do not inherit any mitochondria from sperm, making it very unlikely for mitochondrial conditions to be passed down from a father. Both males and females can have MELAS.

Frequency

MELAS is a rare condition, but doctors do not know exactly how common it is. Clinicians are working towards a better understanding of classification and diagnosis.

Signs and Symptoms

MELAS can cause a range of signs and symptoms. The name MELAS stands for: Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-like episodes.

- Myopathy: muscle condition causing weakness, fatigue, and pain
- Encephalopathy: changes in the brain that can cause mental fog (example: confusion, memory loss, trouble focusing), personality changes, and physical symptoms (example: balance and coordination changes, tremors, uncontrolled movements (ataxia), seizures, aphasia (difficulty speaking or processing speech.)
- Lactic acidosis: a buildup of lactic acid in the body. Increased acidity in the blood can lead to vomiting, abdominal pain, extreme tiredness or fatigue, muscle weakness, and difficulty breathing. (See our factsheet on lactic acidosis for more information)
- Stroke-like episodes: can cause temporary weakness (usually on one side of the body, called hemiparesis) and other movement issues, vision loss or other vision issues, confusion, seizures, severe headaches with or without vomiting, slurred speech and altered consciousness. For some people, these episodes may present as a headache with vision loss. These episodes typically begin before age 40, but can happen at any time in life. Repeated stroke-like episodes can lead to brain damage, vision loss, problems with movement, and loss of intellectual abilities (dementia).

People with MELAS may experience involuntary muscle spasms called myoclonus, impaired muscle coordination (ataxia), hearing loss, heart, gastrointestinal and kidney problems, diabetes, and hormonal imbalances.

Signs and symptoms of MELAS can present at any age. Historically, most people with MELAS were diagnosed after signs and symptoms appeared in childhood following a period of normal development. Now, people are also being diagnosed as adults. The exact features, onset trajectory, 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

MELAS is diagnosed using a combination of detailed medical history, physical exam, lab tests, and imaging. When a doctor is determining if someone has MELAS, they may order several different kinds of tests to look for signs of MELAS including:

- Muscle biopsy showing ragged red fibers (RRF)
- SDH staining showing overabundance of mitochondria in smooth muscle and blood vessels
- Measuring biochemical markers in blood or the cerebrospinal fluid (fluid surrounding the brain and spinal cord)
- Brain studies and imaging looking for changes in the structure of the brain or seizure activity (MRI, MRS, EEG)
- Heart studies looking for rhythm changes (EKG)
- Performing a genetic test to look for changes in the genes known to cause MELAS

MELAS is not included on newborn screening panels. If there is a known family history of MELAS, 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 no FDA-approved therapy for MELAS. Treatment and management of MELAS is symptomatic and supportive.

This may include:

- Traditional medications for seizures (antiepileptic drugs)
- Physical therapy and aerobic exercise to support muscle function
- Traditional treatment of heart conditions
- Hearing aids or cochlear implants for hearing loss
- Avoidance of mitochondrial toxins like certain drugs, tobacco, and alcohol
- Mitochondrial supplements like riboflavin and Coenzyme Q10
- Traditional treatment of diabetes
- Traditional treatment for migraines

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Type of Specialists and Clinicians:

- Audiology
- Cardiology
- Endocrinology
- Neurology
- Ophthalmology

Clinical Trials

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

Resources

- Mitochondrial encephalomyopathy lactic acidosis and stroke-like episodes – About the Disease – The National Institutes of Health
- MELAS Syndrome – 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.