MITO PROFILE

MELAS INFORMATION

PROVIDED BY THE UNITED MITOCHONDRIAL DISEASE FOUNDATION

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MELAS - Mitochondrial Myopathy (muscle weakness), **Encephalopathy** (brain and central nervous system disease), **Lactic Acidosis** (buildup of a cell waste product), and **Stroke-like** Episodes (partial paralysis, partial vision loss, or other neurological abnormalities)

MELAS is a progressive neurodegenerative disorder with typical onset between the ages of 2 and 15, although it may occur in infancy or as late as adulthood. Initial symptoms may include stroke-like episodes, seizures, migraine headaches, and recurrent vomiting.

Usually, the patient appears normal during infancy, although short stature is common. Less common are early infancy symptoms that may include developmental delay, learning disabilities or attention-deficit disorder. Exercise intolerance, limb weakness, hearing loss, and diabetes may also precede the occurrence of the stroke-like episodes.

Stroke-like episodes, often accompanied by seizures, are the hallmark symptom of MELAS and cause partial paralysis, loss of vision, and focal neurological defects. The gradual cumulative effects of these episodes often result in variable combinations of loss of motor skills (speech, movement, and eating), impaired sensation (vision loss and loss of body sensations), and mental impairment (dementia). MELAS patients may also suffer additional symptoms including: muscle weakness, peripheral nerve dysfunction, diabetes, hearing loss, cardiac and kidney problems, and digestive abnormalities. Lactic acid usually accumulates at high levels in the blood, cerebrospinal fluid, or both.

MELAS is maternally inherited due to a defect in the DNA within mitochondria. There are at least 17 different mutations that can cause MELAS. By far the most prevalent is the A3243G mutation, which is responsible for about 80% of the cases. The incidence is unknown, although the epidemiological studies of the MELAS-3243 mtDNA mutation have estimated the prevalence to be 1-16/100,000 in the adult population.

There is no cure or specific treatment for MELAS. Although clinical trials have not proven their efficacy, general treatments may include such metabolic therapies as: CoQ10, dichloroacetate (DCA), creatine, menadione, phylloquinone, and other vitamins and supplements. Drugs such as seizure medications and insulin

may be required for additional symptom management. Some patients with muscle dysfunction may benefit from moderate supervised exercise.

The prognosis for MELAS is poor. Typically, the age of death is between 10 to 35 years, although some patients may live longer. Death may come as a result of general body wasting due to progressive dementia and muscle weakness, or complications from other affected organs such as heart or kidneys.

RESOURCES FOR MELAS INFORMATION:

MELAS by Salvatore DiMauro, MD, through *GENEReviews* www.genetests.org

http://www.genetests.org/servlet/access?db=geneclinics&site=gt&id=8888891&key=i0MCvmaQttROa&gry=&fcn=y&fw=XJ80&filename=/profiles/melas/index.html

MELAS Syndrome by Fernando Scaglia, MD *eMedicine* article
http://www.emedicine.com/ped/topic1406.htm

MELAS Syndrome by William Shiel, Jr. MD, FACP, FACR *MedicineNet.com*
http://www.medicinenet.com/melas_syndrome/article.htm

MELAS Syndrome by Mary Kugler MSN, RN, BC
http://rarediseases.about.com/cs/melassyndrome/a/021204.htm

Strokes and Transient Events in Mitochondrial Cytopathies by Bruce Cohen, MD
Article in *Think Mitochondria*, compendium, available through UMDF

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