Condition: MELAS (mitochondrial encephalopathy, lactic acidosis, strokelike episodes)

Inheritance:

Mitochondrial (maternal transmission).

Genetic etiology:

Mutation in *MT-ND5* or *MT-TL1*, encoding NADH-ubiquinone oxidoreductase chain 5 or mitochondrial tRNA leucine 1, respectively. The most common mutation is A3243G in *MT-TL1*.

Frequency:

Estimated at approximately 1/6,000 in study done in northern Finland.

Clinical features:

MELAS is a highly variable disorder that affects energy metabolism in multiple tissues. Onset is most commonly in childhood, with seizures, headaches, weakness, stroke-like episodes of transient neurological dysfunction and lactic acidosis, sensorineural hearing loss. There is a gradual loss of cognitive function. Other neurological features can include ataxia and myoclonus.

Management:

Supportive care; administration of coenzyme Q10 and L-carnitine.

Genetic counseling:

Follows pattern of maternal transmission, but with widely variable expression due to heteroplasmy. Molecular genetic testing is available.

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