

MELAS (Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like episodes) syndrome is a rare disorder that begins in childhood, usually between two and fifteen years of age, and mostly affects the nervous system and muscles. The most common early symptoms are seizures, recurrent headaches, loss of appetite and recurrent vomiting. Stroke-like episodes with temporary muscle weakness on one side of the body (hemiparesis) may also occur and this can lead to altered consciousness, vision and hearing loss, loss of motor skills and intellectual disability. MELAS is caused by mutations in mitochondrial DNA and in one patient, this syndrome has been associated with mutations in a nuclear gene, POLG1. MELAS syndrome is a rare disorder that affects males and females in equal numbers. Although rare, MELAS syndrome is probably the most common type of mitochondrial myopathy caused by mutations in mtDNA. Some researchers believe that mitochondrial myopathies may go unrecognized and underdiagnosed in the general population, making it difficult to determine the true frequency of disorders like MELAS syndrome.