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Editorial Note on MELAS Syndrome

Sanike Swapna*

Department of Biotechnology, Osmania University, Hyderabad, Telangana, India

Editorial

MELAS syndrome (Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like Episodes) is a rare disorder that begins in childhood, usually between the ages of two and fifteen, and primarily affects the nervous system and muscles. Seizures, recurrent headaches, loss of appetite, and recurrent vomiting are the most common early symptoms. Stroke-like episodes with temporary muscle weakness on one side of the body (hemiparesis) are also possible, and can result in altered consciousness, vision and hearing loss, motor skill loss, and intellectual disability. MELAS is caused by mitochondrial DNA mutations, and in one patient, this syndrome has been linked to POLG1 nuclear gene mutations.

MELAS syndrome symptoms typically appear between the ages of two and fifteen years, but delayed onset cases between the ages of fifteen and forty years have also been reported, as have late onset cases after forty years. In approximately 75% of cases, the disorder manifests itself before the age of 20. MELAS syndrome symptoms and physical findings differ greatly between affected individuals within the same family and between families. MELAS syndrome is distinguished by the recurrence of stroke-like episodes. It is currently thought that the stroke-like episodes are caused by a lack of a compound called nitric oxide in the brain's small blood vessels.

MELAS syndrome is characterised by an accumulation of lactic acid in the blood (lactic acidosis), which can cause vomiting, abdominal pain, fatigue, muscle weakness, and difficulty breathing. Lactic acid buildup has also been observed in spinal fluid and the brain. In some cases, affected individuals will experience a gradual decline in intellectual function (dementia) and/or a reduced ability to communicate through speech, writing, and/or signs (aphasia). MELAS syndrome patients may also experience episodes of confusion and hallucination, which are frequently caused by a preceding fever (febrile illness) and/or headache. Involuntary muscle spasms (myoclonus), impaired muscle coordination (ataxia), cardiomyopathy, diabetes mellitus, depression, bipolar disorder, gastrointestinal problems, and kidney problems are among the less common symptoms.

MELAS is caused by mitochondrial DNA mutations (mtDNA). Mutations in the mtDNA genes are inherited from the mother. MtDNA found in sperm cells is usually lost during fertilisation, so all human mtDNA comes from the mother. A mother who has the mutation will pass it on to all of her children, but only her daughters will pass it on to their children. Mitochondria, which can be found in hundreds or thousands in the body's cells, especially in muscle and nerve tissue, contain the blueprints for regulating energy production.

Normal and mutated mtDNA can coexist in the same cell, a condition known as heteroplasmy. The number of defective mitochondria may outnumber the number of normal mitochondria. Symptoms may not appear in any given generation until the mutation affects a significant proportion of mtDNA. The uneven distribution of normal and mutant mtDNA in different tissues can affect different organs in members of the same family. This can cause a variety of symptoms in affected family members.

MELAS syndrome is a rare disorder that affects both men and women in equal numbers. MELAS syndrome, though uncommon, is most likely the most common type of mitochondrial myopathy caused by mtDNA mutations. Some researchers believe that mitochondrial myopathies are underrecognized and underdiagnosed in the general population, making it difficult to determine the true prevalence of disorders like MELAS syndrome [1-5].

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***Address for Correspondence:** Sanike Swapna, Department of Biotechnology, Osmania University, Hyderabad, Telangana, India, E-mail: sanika.swapna5@gmail.com*

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