



Introduction

Mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes (MELAS) is a maternally inherited disorder characterized by mutations of mitochondrial DNA, most commonly the .m3243A>G mutation in the MT-TL1 gene. This mutation can lead to decreased mitochondrial protein synthesis, which can result in insufficient energy production and multi organ dysfunction. Symptoms can present at any time after a normal childhood development and can include muscle weakness, recurrent headaches, and seizures. Most individuals experience stroke-like episodes that result in hemiparesis, hemianopia, and/or cortical blindness. It is a rare disorder that requires a multidisciplinary approach for proper treatment.

Case Report

A 20 year old male with a history of migraines, provoked seizures, and exercise induced asthma presented to the emergency department with two weeks of headache, dizziness, and vision changes, preceded by visual aura. His exam was notable for a homonymous hemianopsia. Labs were notable for elevated serum and cerebrospinal fluid lactate. A magnetic resonance imaging (MRI) of the brain was notable for diffuse diffusion restriction throughout the left occipital lobe with an apparent diffusion coefficient (ADC) correlate, consistent with acute left posterior cerebral artery infarct. However, the region affected did not fully respect one vascular territory. Additionally, this MRI was compared to an outside MRI from three years prior, which showed an area of diffusion restriction in the left occipital lobe, which was normal on the current exam suggesting a reversible ischemia. Upon further discussion with his family, the patient had seen a pediatric neurologist as a teenager and underwent genetic testing, that was reportedly inconclusive. However, a review of outside records showed a 57% mitochondrial heteroplasmy of the .m3243A>G gene, which was compatible with a diagnosis of MELAS. The patient underwent a transthoracic echocardiogram, which showed severe right ventricular dysfunction. This was followed with a cardiac MRI which showed biventricular enlargement and systolic dysfunction, suggesting cardiac involvement of his mitochondrial disorder. He also developed recurrent clinical and subclinical seizures which were managed medically with lacosamide, perampanel, and levetiracetam. Eventually, his headaches and seizures improved and he was discharged with follow up with Neurology, Clinical Genomics, Neuro Ophthalmology, and Cardiology.

Headache and Hemianopia: A Case of Mitochondrial Encephalomyopahty with Lactic Acidosis and Stroke Like Episodes

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Image 1

Diffusion weighted imaging of the patient, which shows diffuse diffusion restriction throughout the left occipital lope extending slightly into the parietal lobe. Of note, the patient had an MRI from 2016 which showed a similar area of diffusion restriction in the right occipital lobe, which is not present on the current exam.



MELAS is a rare, inherited mitochondrial disorder that can result in multi organ dysfunction, due to inadequate energy production from the mitochondria. The presenting feature of this syndrome is the occurrence of stroke like episodes, resulting in hemiparesis, hemianopia, or cortical blindness. Some MRI features of these stroke-like episodes include lesions that don't strictly follow vascular territories, varied ADC correlate, and MR signal changes that can improve or

Image 2

Shows the apparent diffusion coefficient (ADC) mapping, which shows a correlation to the diffusion restriction seen, consistent with a "stroke like" finding.



Discussion

completely resolve, unlike ischemic stroke. Other symptoms include recurrent headaches, exercise intolerance, seizures, and muscle weakness. This syndrome can result in multi organ dysfunction and requires a multidisciplinary approach from specialties including neurology, clinical genetics, neuro-ophthalmology, cardiology, and nephrology.