Inpatient rehabilitation of a 28-month-old male with mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes (MELAS) syndrome: a case report

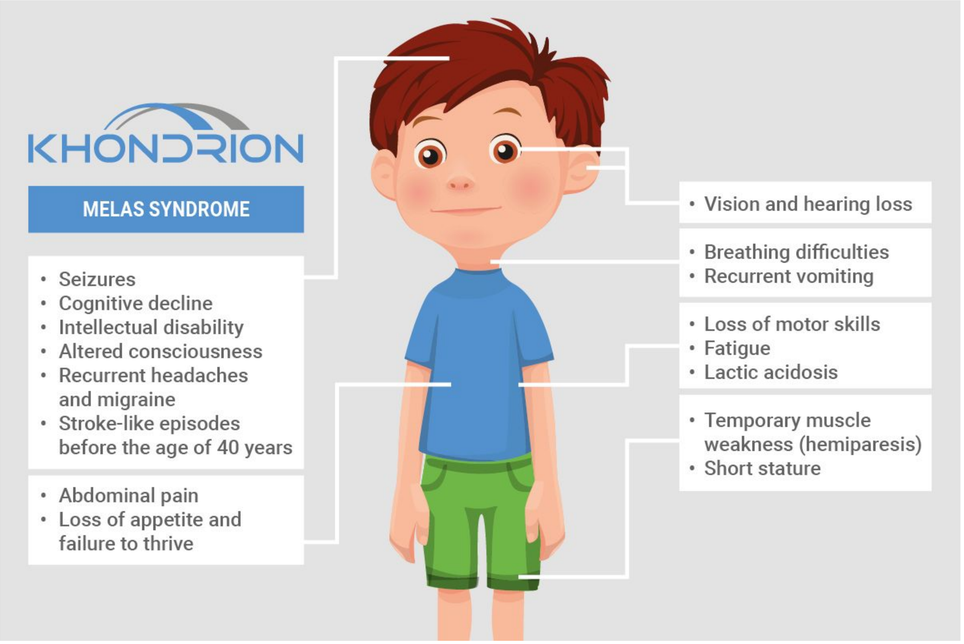
George Schilling, DO, Dept. Physical Medicine & Rehabilitation
Leonardo Villarosa, MD, Dept. Physical Medicine & Rehabilitation

George J. Schilling
Physical Medicine and Rehabilitation
East Carolina University
Greenville, North Carolina 27834
georgeschilling@gmail.com

CASE DESCRIPTION

The patient presented to the ED with left upper and lower extremity weakness and stiffening preceded by a diarrheal illness. Brain MRI demonstrated T2 hyperintensity in the right caudate, thalamus, and anterior hippocampus concerning for post-viral changes. After extensive workup, the patient was diagnosed to have mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes (MELAS) syndrome in the setting of encephalopathy, high serum lactate, and elevated alanine. L-arginine was started and the patient was admitted to the pediatric inpatient rehabilitation hospital. The patient initially required maximal assist to stand and total assistance to walk 5'. On discharge, the patient only needed contact guard assistance to stand and minimum assistanceto walk up to 150 and ascend four 6-inch steps.

INFOGRAPHIC



Above: Infographic describing common manifestations of MELAS syndrome. The patient presented with many of the above signs and symptoms including stroke-like episodes, loss of motor skills, hemiparesis, seizures, fatigue, and loss of appetite.

Source: Khondrion B.V.

DISCUSSION

MELAS syndrome is a rare, maternally inherited genetic syndrome that can result in stroke-like episodes including hemiparesis and is associated with seizures and vomiting. Most MELAS cases are associated with a variant in the MTTL1 gene. In addition to impaired energy production, nitric oxide (NO) deficiency occurs in MELAS and leads to impaired blood perfusion in the microvasculature that can contribute to several complications including stroke-like episodes, myopathy, and lactic acidosis. L-arginine is believed to benefit MELAS by potentiating nitric oxide pathways and inducing vasodilation. The stroke-like lesions differ from traditional embolic or thrombotic ischemic events as lesions are not limited to vascular territories and signal changes may migrate, fluctuate, or resolve more quickly than a typical ischemic stroke. Because of the neurologic damage, patients often present with severe functional deficits and can benefit from intensive interdisciplinary therapies.

CONCLUSION

MELAS syndrome is a rare mitochondrial genetic disorder which can manifest as a stroke.MELAS patients can benefit from acute inpatient rehabilitation.

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