

Neurological finding of Mitochondrial Syndromes (MERRF and MELAS)

How to Cite This Article: Taghdiri MM. Neurological finding of Mitochondrial Syndromes (MERRF and MELAS). *Iran J Child Neurol Autumn 2013; 7:4(suppl. 1):15.*

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MELAS (Mitochondrial Encephalopathy with Lactic Acidosis and Stroke Episodes), first described by Pavlakis et al. MELAS generally presents in children or young adults after normal early development. Symptoms include recurrent vomiting, migraine-like headache, and recurrent stroke-like episodes causing cortical blindness, hemiparesis or hemianopia. Seizures are often preceded by strokes or episodes of migraine with aura and partial motor in type. Intellectual regression and behavioral problems are prominent. The myopathy is usually asymptomatic. Additional features include stature, sensational hearing loss.

Lactic levels in plasma and CSF are usually high, and CPK inconsistently increased and CSF protein is normal. The EEG is usually abnormal. Neuroimaging shows focal lucencies and Ragged-Red fibers are present in muscles. The most common mtDNA mutation is A3243G in the RNA gene.

Myoclonic Epilepsy with Ragged Red Fibers (MERRF) is a maternally inherited encephalopathy, characterized by myoclonous, cerebellar ataxia and mitochondrial myopathy. Seizure and hearing loss are frequently associated. In most cases, symptoms begin between the age 5 and 13 years with a cerebellar ataxia, tremor and myoclonic jerks.

The EEG records abnormal background activity.

Keywords: MELAS; MERRF; Mitochondrial syndromes

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