
Cerebellar Ataxia, Myoclonus, Cervical Lipomas, and MERRF Syndrome. Case Report

Mitochondrial DNA (mtDNA) accounts for only 1% of the total cellular nucleic acid content, encoding for 13 polypeptides that are essential for aerobic metabolism. Defects of the mitochondrial genome are an important cause of human disease, including encephalopathies. One such example is myoclonic epilepsy with ragged-red fibers (MERRF), in most cases due to an A to G transition at position 8,344 in the tRNA Lys gene of mtDNA. Adding to the classical description, several variants of MERRF have been described including signs such as cognitive impairment, sensory hearing loss, optical atrophy and peripheral neuropathy. Here, we report a patient with an unusual presentation of genetically proven MERRF including cerebellar ataxia, myoclonus, and cervical lipomas, compatible with Ekbom’s syndrome.

A 52-year-old man had a 15-year history of slowly progressive dysarthria, dizziness, and gait disturbances. Two years after initial symptoms onset, short-term memory deficits developed, progressing to disorientation for time and place. In the last 4 years there was progressive worsening of gait ataxia and dysarthria as well as cognition. Family history was positive for a maternal cousin with a diagnosis of a mitochondrial disorder (MERRF). General examination revealed symmetrical, posterior, cervical lipomas. Neurological examination revealed cognitive dysfunction (MMSE: 18/30), moderate scanning dysarthria with slow but understandable speech. Eye movements were full with bilateral horizontal nystagmus on lateral gaze. Remaining cranial nerves were normal. Moderate, intermittent segmental action myoclonus was present in the upper extremities. Tone was slightly reduced in the upper and lower limbs and rebound was present in the upper limbs. Synergy, trajectory, and placement of the limbs were abnormal with dysmetria and dysdiadochokinesia that were moderate in the upper and mild in the lower extremities. Strength was 4/5 throughout and stretch reflexes were hypoactive but symmetric. Plantar responses were flexor. Vibration and position senses were normal. Gait and stance were ataxic and only two to three steps were possible with tandem gait. A head CT scan showed cerebellar atrophy and extensive symmetrical lipomas in the posterior cranio-cervical area. MRI demonstrated cerebellar atrophy (see Fig. 1). CSF anal-

FIG. 1. Brain MRI (T1-weighted sagital image) of the patient shows cerebellar atrophy.
Our case, with its distinct phenotypical presentation of mitochondrial encephalopathy, MERRF, symmetrical lipomas, cerebellar ataxia, upper extremities action myoclonus, and dementia, is compatible with the description of Ekbom’s syndrome. Our case also highlights aspects of possible overlapping clinical features between this syndrome and other disorders related to mtDNA mutations, particularly MERRF.

Hélio A.G. Teive, MD, PhD*
Renato P. Munhoz, MD
Juliano A. Muzzio, MD
Rosana H. Scola, MD, PhD
Cláudia K. Kay, MD
Salmo Raskin, MD, PhD
Lineu C. Werneck, MD, PhD

Movement Disorders Unit
Neurology Service
Hospital de Clínicas
Federal University of Paraná
Curitiba, Pr, Brazil

*E-mail: hagteive@mps.com.br

Helene Bruhn, MSc
Centre for Inherited Metabolic Diseases
Karolinska University Hospital
Huddinge, Stockholm, Sweden

REFERENCES