Case 15941

Eurorad

Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke (melas) syndrome: a 2 year follow-up

Published on 22.07.2018

DOI: 10.1594/EURORAD/CASE.15941
ISSN: 1563-4086
Section: Neuroradiology
Area of Interest: Neuroradiology brain
Procedure: Teleradiology
Procedure: Complications
Procedure: Technical aspects
Imaging Technique: MR
Imaging Technique: MR-Spectroscopy
Imaging Technique: MR-Diffusion/Perfusion
Special Focus: Metabolic disorders Case Type: Clinical Cases
Authors: Andrés Cárdenas, MD; Silvia Cárdenas, MD.
Patient: 4 years, female

Clinical History:

A 4-year-old female with recent and progressive bilateral sensorineural hearing loss, extrapyramidal symptoms and hyperlactacidaemia, came to our institution and a MRI was performed. She returned two years later for a control with right anacusia, left hearing loss and strength and motility deterioration. A second MRI was performed.

Imaging Findings:

The first MRI, no significant findings are found in the morphological sequences; However, spectroscopy shows moderate increase of the lactic acid peak. On the second MRI, brain atrophy is observed. T2 and FLAIR images reveal abnormal hyperintensity with cortical and juxtacortical involvement over both cerebral hemispheres. They are located predominantly on both sides of the parasagittal convexity, perisylvian and perirolandic areas, as well as in the inferior and medial occipital cortical regions. These lesions have high signal on diffusion-weighted images (DWI), with variable values in the apparent diffusion coefficient (ADC). The spectroscopy again reveals a lactate peak although to a lesser degree.

Discussion:

Mitochondrial diseases (MD) are a heterogeneous group of disorders with symptoms of organ dysfunction across multiple body systems and occur when alteration of mitochondrial respiratory chain complex function caused by genetic mutation produces a detectable disease state [1]. Clinical features manifest typically in tissues with the highest energy requirements including skeletal muscles, brain, myocardium, and endocrine systems [2]. Several distinct MD syndromes have been described, comprising mitochondrial encephalomyopathy and lactic acidosis

stroke-like episodes [3, 4, 5]. Clinically it presents with signs and symptoms of encephalomyopathies, growth disturbance, seizures, and stroke-like events that might be reversible or irreversible with permanent neurologic deficits [6, 7]. Muscle biopsies reveal the presence of ragged red fiber.
MRI remains the main imaging technique to evaluate metabolic disorders [8]. In an acute episode the disease appears on CT as hypodense cortical areas usually located in one or both parieto-occipital lobes and not confined to single vascular territory [9]. On MR, T2 hyperintense lesions may appear with a predilection for the cerebral cortex rather than underlying white matter. These lesions migrate over time, and have some predilection to occipital and parietal lobes [10, 11]. Progressive atrophy of the basal ganglia with calcifications, temporal- parietal-occipital cortex with preservation of hippocampal, entorhinal structures, and multifocal basal ganglia, deep white matter hyperintensities are chronic manifestations [12].
DWI shows high signal during acute episode, with variable ADC [13, 14]. Recent studies suggest that the development of cytotoxic oedema as represented by decreased ADC is likely due to initial neuronal energy insufficiency. Subsequent development of extracellular oedema in surrounding region leads to increased ADC signal [15].
MR spectroscopy shows a characteristic large lactate peak at 1.3 ppm [16], usually reflects anaerobic metabolism, though it has been reported to occur even in the normal-appearing brain [17]. Lactate peak has also been associated with increased disease severity and reduced survival. Elevated lactate in affected regions of the brain tends to gradually resolve as these lesions evolve further into cerebral atrophy [16].

There is no specific treatment for individuals with MELAS. Therapeutic compounds may ameliorate symptoms in individual cases; however, the available therapeutic interventions are not able to affect the essential progression of this disease. Some of the most frequently prescribed agents include ubidecarenone, idebenone, edaravone, levoarginine [18], complex B vitamins, vitamin C, E and levocarnitine.

Think MELAS in patient with acute "stroke-like" cortical lesion that crosses usual vascular territories.

Written informed patient consent for publication has been obtained.

Differential Diagnosis List: Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke (melas) syndrome, Ischaemic stroke, Kawasaki disease, Leigh’s disease, Status epilepticus, Myoclonic epilepsy with ragged-red fibres

Final Diagnosis: Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke (melas) syndrome

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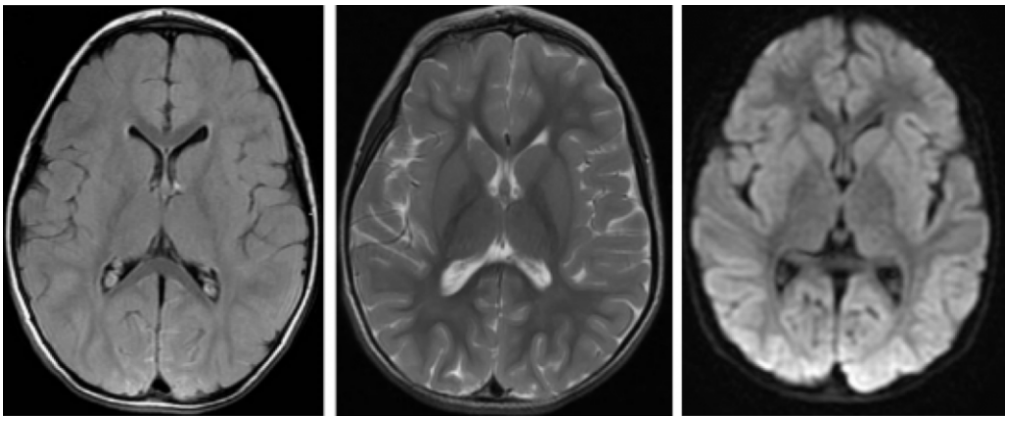
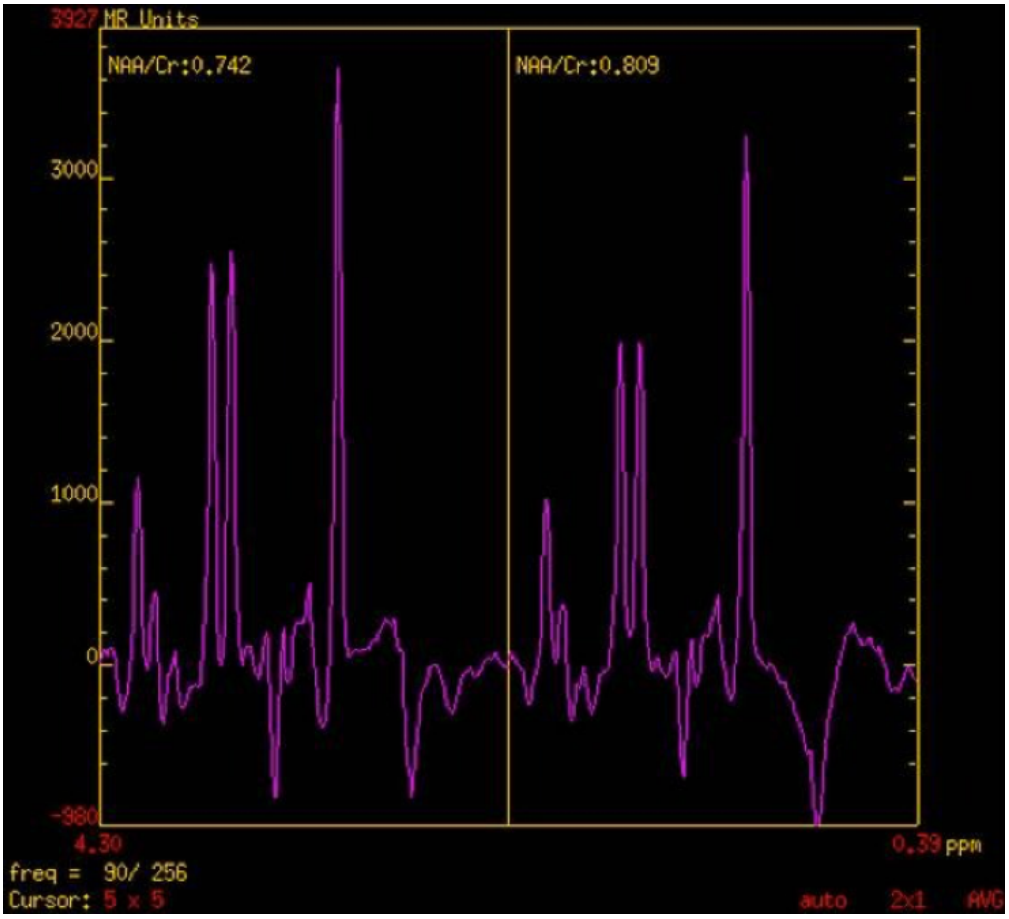
Figure 1

a



Description: AXIAL FLAIR, T2 and DIFFUSION weighted MR images don't show relevant abnormal signal or important structural changes. **Origin:** Digital file of Diagnostico Maipú,Buenos Aires,Argentina.

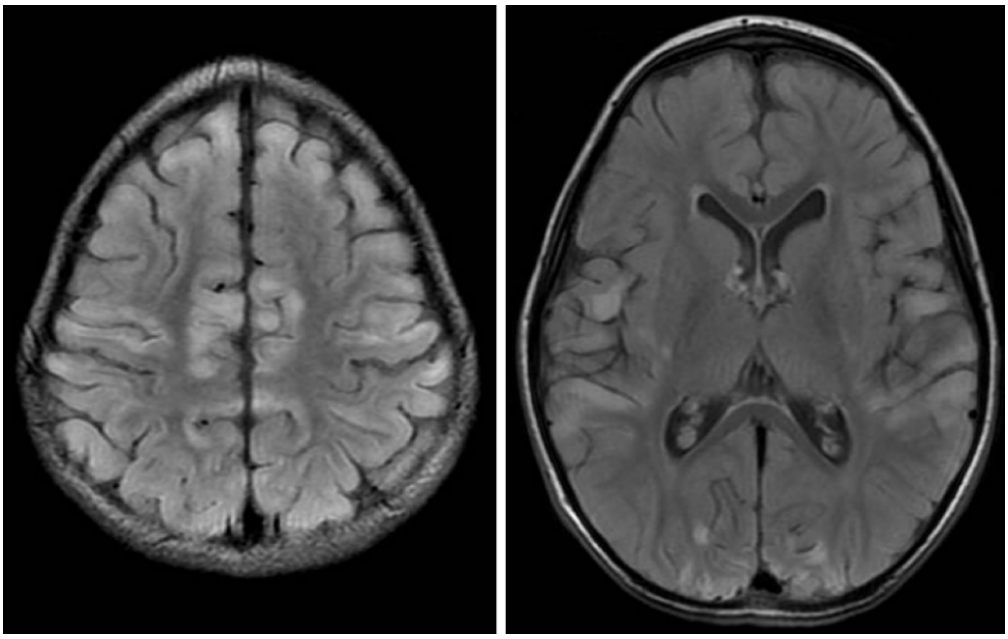
b



Description: Multi-voxel spectroscopy (TE 144 ms), at the level of the left basal ganglia reveals an inverted peak at 1.33 ppm, a finding that is characteristic of lactate accumulation. **Origin:** Digital file of Diagnostico Maipú,Buenos Aires,Argentina.

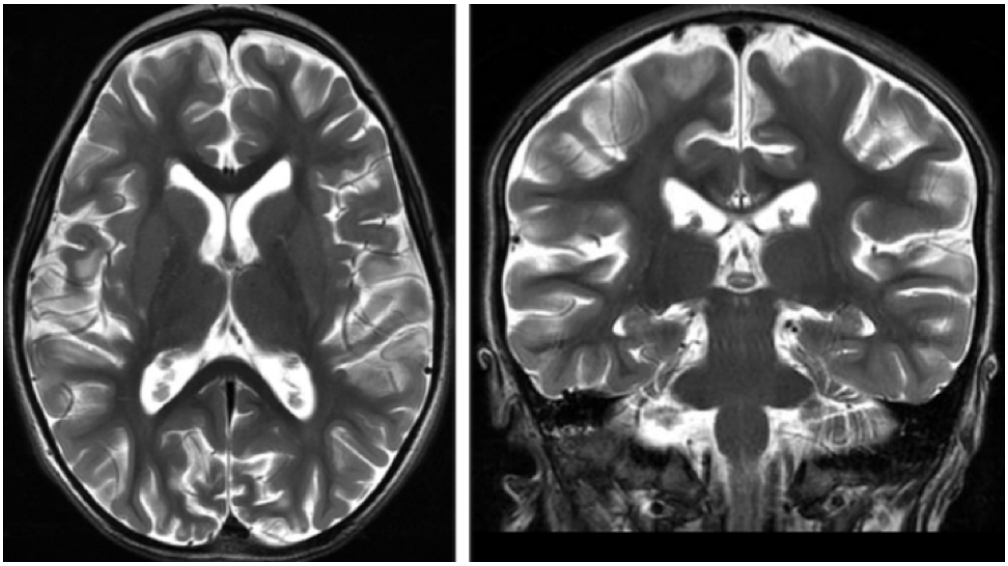
Figure 2

a



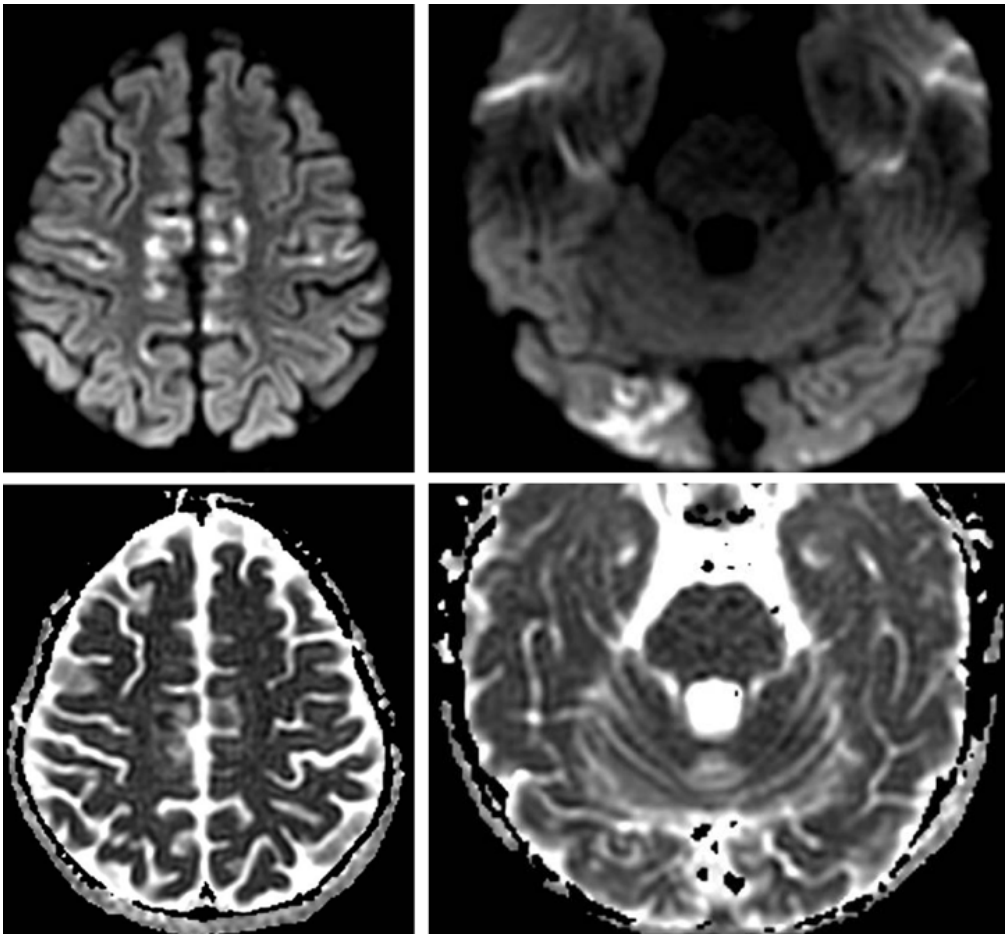
Description: AXIAL T2 FLAIR images show abnormal hyperintensity with cortical and juxtacortical involvement predominantly on both sides of the hemispheric parasagittal convexity, perisylvian and perirolandic areas, as well as the inferior and medial occipital cortical regions. **Origin:** Digital file of Diagnostico Maipu. Buenos Aires. Argentina.

b



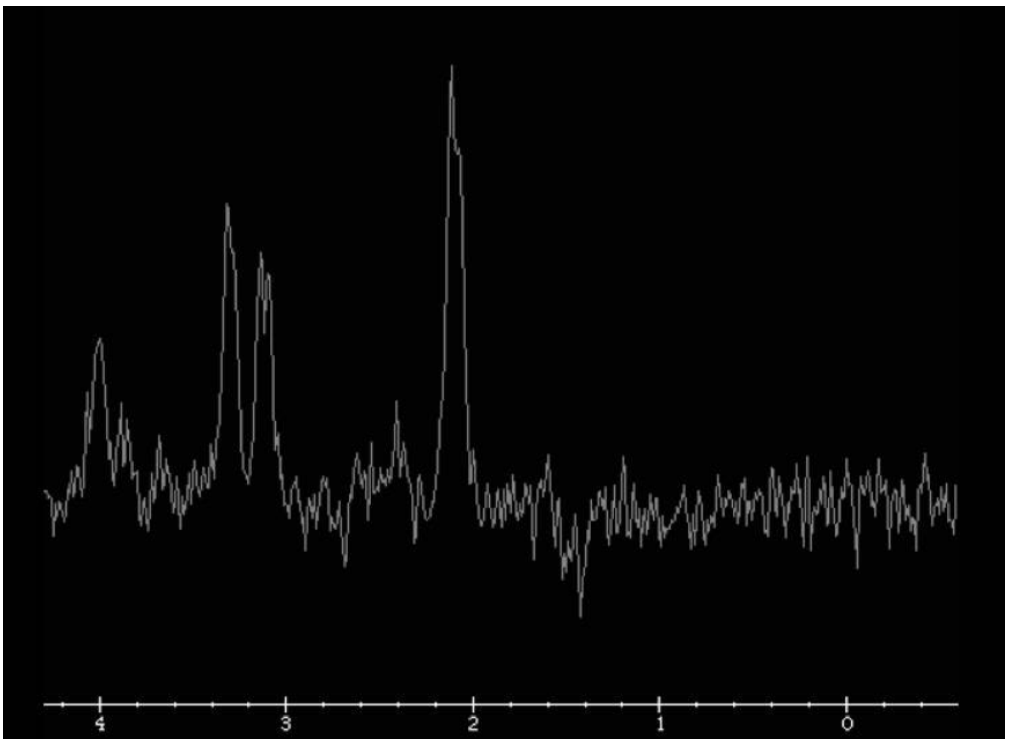
Description: AXIAL and CORONAL T2-weighted MR images show mild to moderate prominence of the sulci and slight dilatation of ventricles. **Origin:** Digital file of Diagnostico Maipu. Buenos Aires. Argentina.

c



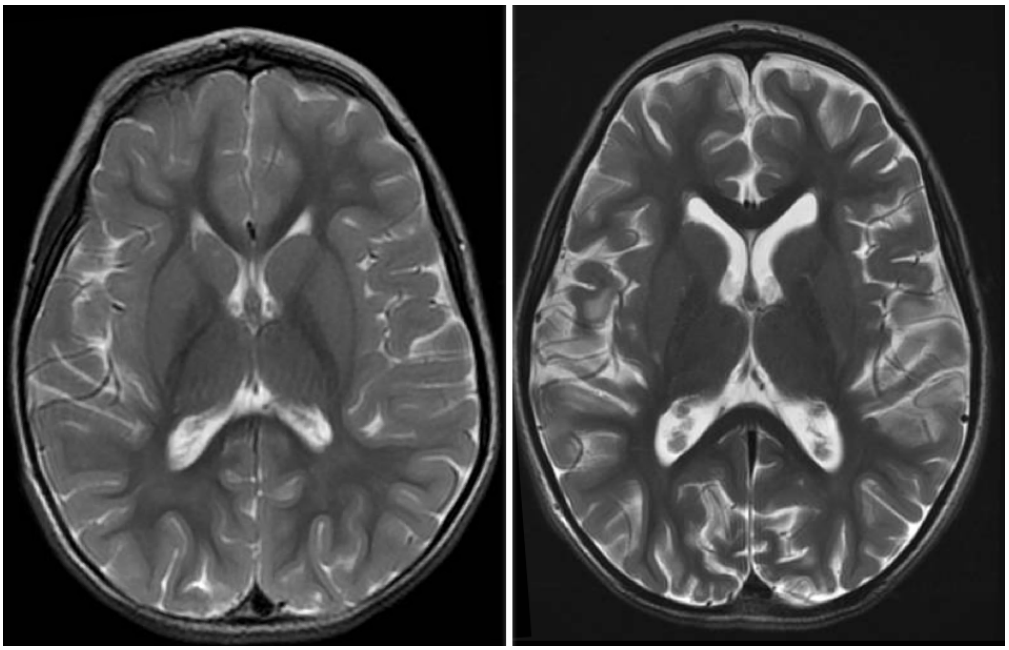
Description: AXIAL-DWI shows hyperintensities with low ADC value on both sides of the cortical hemispheric parasagittal convexity and perirolandic areas, and high signal with high ADC value on inferior and right medial occipital cortical region. **Origin:** Digital file of Diagnostico Maipu. Buenos Aires. Argentina.

d



Description: Spectrum from MR spectroscopy performed with an echo time of 144 msec with the voxel placed over the region of interest (left basal ganglia) reveals slight increase of inverted lactate peak.
Origin: Digital file of Diagnostico Maipu. Buenos Aires. Argentina.

e



Description: T2-weighted MR images, 2 years apart; note the increase of the ventricular system and sulci, consistent with cerebral atrophy. **Origin:** Digital file of Diagnostico Maipu. Buenos Aires. Argentina.