Acta Neurologica Belgica
https://doi.org/10.1007/s13760-021-01694-2

NEURO-IMAGES

NEURO-IMAGE: MELAS (mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes syndrome)

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Received: 19 March 2021 / Accepted: 23 April 2021

Keywords MELAS · Stroke mimic · Mitochondrial · MRI · MRS

Case report

A 30-year-old woman with a history of bilateral sensorineural deafness since the age of 14 years, a right occipital ischemic stroke with left hemianopia and epilepsy was admitted to the neurology department with speech disorders and a new epileptic insult. MRI revealed extensive cytotoxic edema of the left temporo–parieto–occipital cortex and mirrored right-sided sequelae (Fig. 1a, b). MR-angiography and MR-venography (not shown) were normal.

The patient's symptoms including stroke-like episodes, epileptic seizures, generalized muscle weakness, short stature, deafness and cardiomyopathy suggested a MELAS syndrome (mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes). High levels of lactic acid in both blood and cerebrospinal fluid (CSF) samples, and reversal of the lactate doublet at 1.3 ppm at long and short TE at proton MR spectroscopy reinforced the hypothesis.

Genetic analysis confirmed the presence of a heteroplasmic m.3243A > G MELAS mutation in the MT-TL1 gene of the mtDNA of leukocytes.

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Comment

MELAS syndrome is a rare multisystem disorder affecting young patients between 2 and 40 years of age. It belongs to a group of mitochondrial metabolic diseases caused by an absence or deficit of subunits of the respiratory chain protein complex due to a mutation in mitochondrial DNA leading to impaired cell function or even cell death [2, 3].

The clinical/paraclinical diagnosis is based on stroke-like episodes occurring before the age of 40 years, encephalopathy with seizures and/or dementia, the presence of elevated levels of lactic acid in blood and CSF samples and ragged red fibers at muscle biopsy [2].

The stroke-like episodes of MELAS are clinically indistinguishable from common ischaemic stroke, thus using MRI can be helpful [1]. MRI shows cortical and subcortical acute damage presenting high signal on T2/FLAIR, restricted diffusion with lowered ADC values as is the case for real ischaemic lesions. These lesions are often located in posterior brain regions, and contrary to ischaemic stroke, their distribution does not follow vascular territories. MR-angiography does not reveal occlusion of a main artery and perfusion-weighted (PW) imaging does not highlight cerebral tissue hypoperfusion. On the contrary, MRA and PWI may display arterial vasodilatation and hyperperfusion [1–5].

Proton MR spectroscopy may add to the diagnosis by revealing elevated lactate peak at 1.3 ppm and decreased NAA one in stroke-like lesions [2].

In some patients, the distribution of the lesions can mimic that of the HSV encephalitis [5]. In such condition, DWI may add to the diagnostic suspicion by demonstrating high signal in only subareas of the acutely damaged brain tissue and not throughout the entire lesions, besides the relevant input of reversal of the lactate doublet at MRS.

Published online: 04 May 2021

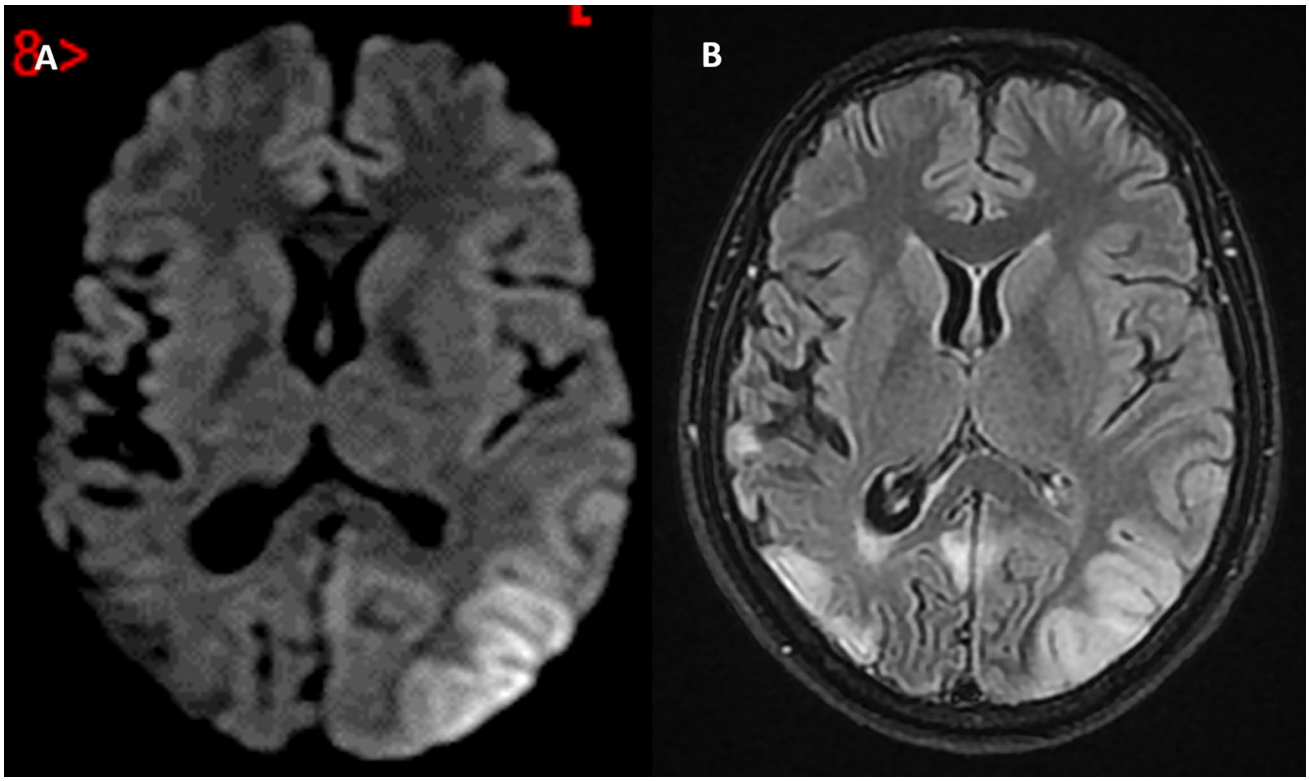

Fig. 1 **a** Axial diffusion-weighted image (DWI) shows restricted diffusion in the left temporo–parieto–occipital junction. **b** Axial fluid-attenuated inversion recovery (FLAIR) image reveals increased signal intensity in the same territory as on the DWI, and mirrored sequelae on the right

In conclusion, MELAS syndrome is a rare IEM (Inborn Error of Metabolism) featured by stroke-like episodes in young or middle-aged people. The MR images in our patient clearly highlight the features of MELAS, such as the coexistence of acutely injured brain areas together with chronic sequelae, the prominent location of lesions in posterior brain areas, the mosaicism of water diffusivity restriction within acute lesions, and the topographic lesions' distribution not matching arterial vascular territories. The MRS can be helpful to confirm the diagnosis besides blood and CSF analyses and ultimately genetic analysis.

Declarations

Ethical approval Not applicable.

Competing interests The authors have no competing interests to declare.

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