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CLINICAL CASE

MELAS syndrome: Would clinical features and tomography lead us to diagnosis before mitochondrial DNA test?

Síndrome de MELAS: ¿Podrían el cuadro clínico y la tomografía llevarnos al diagnóstico previo a la prueba de ADN mitocondrial?

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ABSTRACT

Mitochondrial diseases are not common in clinical practice, some of them are treated as myopathies or just only muscular complications of common disease (diabetes and chronic renal disease). MELAS syndrome (acronym of mitochondrial myopathy, encephalopathy, lactic acidosis, and "stroke-like" episodes) is one of them. We introduce a patient 46 years old, previously with diabetes and hypoacusis who progress to severe acidemia and heart arrest with intensive care unit management, suspecting MELAS syndrome treatment began with improving in few days and discharged with minimal complications. In this case, its clinical features and computed tomography findings lead us to suspect his diagnosis.

Key words: MELAS. Mitochondrial encephalopathy. Hypoacusis. Myopathy. Stroke-like. Lactic acidosis.

RESUMEN

Los trastornos mitocondriales son poco comunes en la práctica clínica, algunas son tratadas como miopatías o complicaciones musculares de trastornos comunes (diabetes, enfermedad renal crónica). El síndrome MELAS (miopatía mitocondrial, encefalopatía, acidosis láctica y episodios ictus-like) es uno de ellos. Presentamos el caso de un paciente de 46 años, diabético, con hipoacusia, el cual progresa a acidemia severa y parada cardiorrespiratoria, se manejó en cuidados intensivos. Sospechando síndrome de MELAS se inicia manejo, con mejoría progresiva, siendo dado de alta con mínimas secuelas. Los hallazgos clínicos y tomográficos iniciales nos permitieron sospechar el diagnóstico y encaminar su estudio.

Palabras clave: MELAS. Encefalopatía mitocondrial. Hipoacusia. Miopatía. Acidosis láctica. Ictus.

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INTRODUCTION

MELAS syndrome (mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes) is an uncommon disease it's reported in England mitochondrial disease a prevalence of 2.9/100,000[1], there are few studies in adulthood, the mean clinical features in MELAS syndrome points to its acronym, but some other features are common in this patients such as sensorineural hearing loss, short stature, and diabetes[2]. Usually, MELAS syndrome has a young onset but there are reports of older onset patients[3,4]. We share our approach in a 46-year-old male based on clinical features and tomography findings.

CLINICAL CASE

A 46-year-old male, unemployed right-handed, originally from Mexico City, smoking denied irregular alcohol consumption for 20 years old (every 3-6 months) with no use of any other illegal substance. Diabetes type 2 of 7 months of onset in current management with metformin 850 mg every 8 h and glibenclamide 5 mg every 12 h refers 2 months of tinnitus first in one site then bilaterally that progress to bilateral hearing loss with left predominance, mood changes. One week before admission to our unit, he began with the left hemiparesis with involuntary distal hand left movements and aphasia and disorientated, in private health-care system requested a brain magnetic resonance and came with us for evaluation.

On his 2nd day of hospital stay, he worsening with dyspnea, two generalized tonic-clonic seizures, and sudden cardiac arrest, with severe lactic acidemia (pH 6.89) with lactic acid of 6 mmol/L, with these findings, diabetic ketoacidosis was suspect, and advanced cardiopulmonary resuscitation began, and insulin treatment and intensive care unit management were solicited for severe diabetic ketoacidosis, a cranial tomography was performed (Fig. 1). There was another finding in computed tomography (Fig. 2); abnormal bilateral symmetric calcifications in the caudate nucleus were found.

There were perform muscle biopsy (with chronic unspecified changes in optic microscopy) and mitochondrial DNA gene mutation m.A3243G in MTTL1 gene in blood and muscular tissue was performed with a magnetic

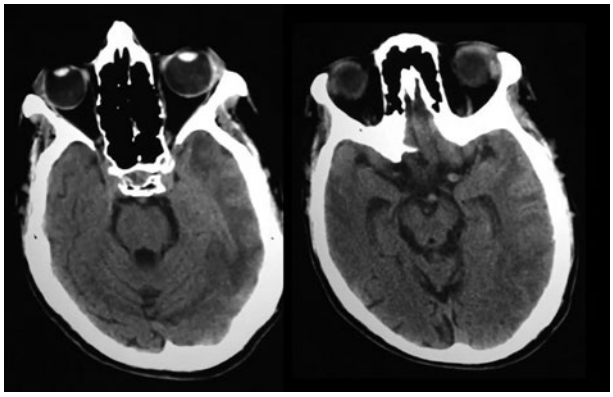

Figure 1. Axial computed tomography with cortical and subcortical white and gray matter changes with lack of specific vascular territory in temporal and occipital left hemisphere.

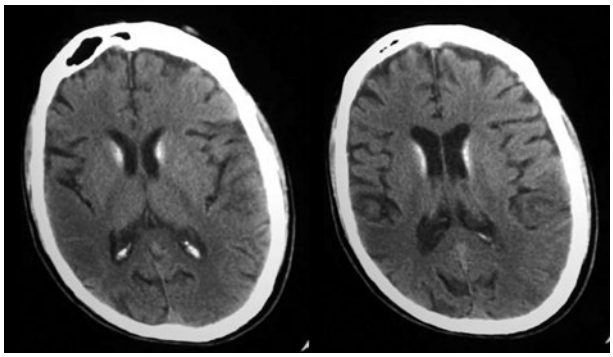

Figure 2. Axial computed tomography with symmetric calcifications in caudate nucleus.

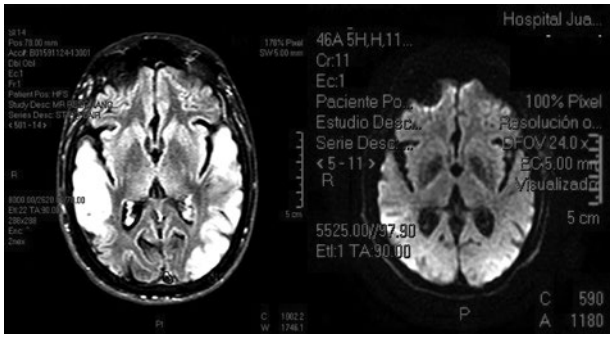

Figure 3. Diffusion weighted imaging magnetic resonance with improve of "stroke-like" lesions 1 week before admission (left) and 2 weeks as inner patient with L-carnitine and arginine management with supportive care with improvement of clinical features (right).

brain resonance (Fig. 3). There were persistently high levels of lactic acid at his hospital stay (in the range of 3-8 mmol/L). There was positive MTTL1 mutation with a heteroplasmic patron in muscular tissue, performed at molecular biology laboratory at Instituto Nacional de Nutrición Salvador Zubiran and we decided to start management with arginine and L-carnitine supplement[5] with the improvement of the clinical features.

CONCLUSIONS

MELAS syndrome is an uncommon disease in our region, it requires a high suspect index to perform this diagnosis, but some features are very useful in computed tomography, caudatus nucleus calcifications are very uncommon (1% of all cranial tomographies); they are common in degenerative disorders and toxic encephalopathies (plumb and carbon monoxide) and metabolic disorders (hypoparathyroidism and calcium disorders) and other uncommon disorders (Fahr disease, Aicardi-Goutieres syndrome, and Cockayne syndrome). There are basal ganglia symmetric calcifications in 30% of MELAS syndrome patients, so in this case with this tomography finding and lactic acidosis and stroke-like imaging in Diffusion weighted imaging of magnetic resonance leads us to this diagnosis[6].

CONFLICTS OF INTEREST

None.

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ETHICAL DISCLOSURES

Protection of human and animal subjects. The authors declare that no experiments were performed on humans or animals for this study.

Confidentiality of data. The authors declare that they have followed the protocols of their work center on the publication of patient data.

Right to privacy and informed consent. The authors have obtained the written informed consent of the patients or subjects mentioned in the article. The corresponding author is in possession of this document.

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