Radiological and Electroencephalographic Findings in Mitochondrial Complex III Deficiency

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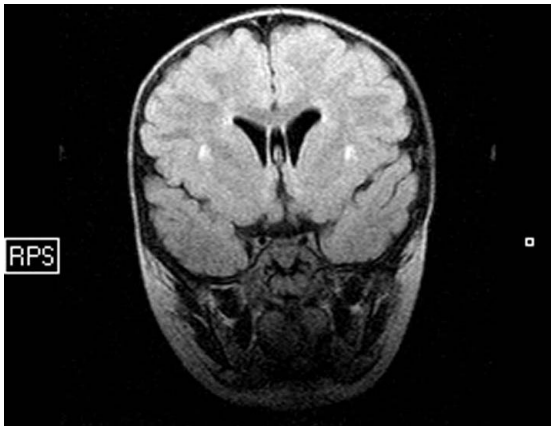

Figure 1. Brain MRI shows an increase of bilateral and symmetrical signal in the anterior portion of both putamens.

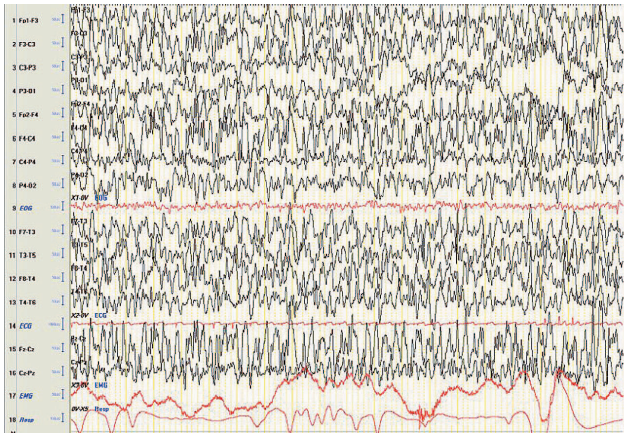

Figure 2. Waking EEG manifests the presence of very disorganized and slowed background activity and persistent spike and multifocal spike-waves paroxysmal. Sens: 7 µV/mm. Time constant: 0,3 sg. High frequency filter: 30 Hz.

A 17 months old girl was in study by psychomotor retardation and phenotypic abnormalities. She presented a severe intrauterine growth retardation and prematurity, respiratory distress, and severe hypocalcemia. The patient obtained a cephalic support at 10 months and sitting at 13 months. Physical examination demonstrated a macrocephaly with width forehead, triangular face and broad nasal root and axial and members hypotonia. No other clinical manifestations were observed. Brain MRI revealed an increase of bilateral and symmetrical signal only in the sequence Flair located in the anterior portion of both putamens. The electroencephalogram showed a very disorganized and slowed background activity and persistent spike and multifocal spike-waves paroxysmal. The most relevant laboratory findings were the presence of hyperlactataemia with increased lactate / pyruvate ratio and an increase in urine beta-aminoisobutiric acid. Muscle biopsy demonstrated a mitochondrial complex III deficiency.

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DISCUSSION

Mitochondrial diseases are disorders caused by alterations in intracellular energy production, affecting tissues with high energy requirements. The nervous system is the most frequently affected[1]. The most representative findings are muscle weakness, hypotonia and psychomotor retardation and seizures and other neurodevelopmental disorders[2]. The syndromic diagnosis of these entities is based on semiologic findings and specific evidence of mitochondrial dysfunction[3] (metabolic studies, and genetic histoenzymatic), being very useful (as the case presented) characteristic alterations in Brain MRI and EEG to guide the clinical suspicion.

REFERENCES

1. Chi CS, Lee HF, Tsai CR, Lee HJ, Chen LH. Clinical manifestations in children with mitochondrial diseases. Pediatr Neurol 2010;43:183-189.
2 Falk MJ. Neurodevelopmental manifestations of mitochondrial disease. J Dev Behav Pediatr 2010;31:610-621.
3. Jesús Eirís Puñal, Carmen Gómez Lado, Manuel Oscar Blanco Barca, Manuel Castro-Gago. Enfermedades mitocondriales. Protocolos Diagnóstico Terapeúticos de la AEP: Neurología Pediátrica. www.aeped.es/protocolos/