

of asymptomatic fat embolism. Brain MRI during the acute phase is the most sensitive imaging technique for diagnosis: DWI reveals lesions arranged in a characteristic "starfield" pattern in a high percentage of patients.<sup>3</sup> Cytotoxic oedema associated with CFE lesions is transient; therefore, the pattern typically seen on DWI is only observed in 18% of patients during the subacute phase.<sup>4,5</sup> A considerable percentage of patients undergo MRI during this phase for numerous reasons, including the use of an external fixator (which is not compatible with MRI) or patient instability. In the light of the above, SWI constitutes a highly useful diagnostic tool<sup>6</sup> and should always be included in the assessment protocol, since it is more sensitive than DWI and T2\*-weighted sequences, which have traditionally been used to rule out bleeding.<sup>7,8</sup> Around 60% of patients with CFE show diffuse petechial haemorrhages, predominantly affecting the white matter; the lesions usually appear on the first day and persist until the chronic phase.<sup>5,9</sup> Two main hypotheses have been proposed to explain the pathophysiology of CFE: the first suggests that lesions result from mechanical vascular occlusion due to embolism, whereas the second postulates that lesions result from the proinflammatory effect of fatty acids; an interaction between both mechanisms is also plausible.<sup>8,10</sup> The usefulness of MRI for determining the association between lesion extension and long-term neurological progression has not been studied, but should be addressed in future research.

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S. Gil-Perotin<sup>a,\*</sup>, J. Carreres-Polo<sup>b</sup>

<sup>a</sup> *Servicio de Neurología, Unidad de Investigación en Esclerosis Múltiple, Hospital Universitario i Politécnico La Fe, Valencia, Spain*

<sup>b</sup> *Área Clínica de Imagen Médica, Sección de Neuroradiología, Hospital Universitario i Politécnico La Fe, Valencia, Spain*

\* Corresponding author.

*E-mail address:* sara.garcia@uv.es (S. Gil-Perotin).

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## Kearns-Sayre syndrome: Absence of clinical response to treatment with oral folinic acid<sup>☆</sup>

## Síndrome de Kearns-Sayre: ausencia de respuesta clínica al tratamiento con ácido folínico oral

*Dear Editor:*

Mitochondrial DNA deletion syndromes constitute rare processes resulting from defects in oxidative phosphorylation.<sup>1</sup>

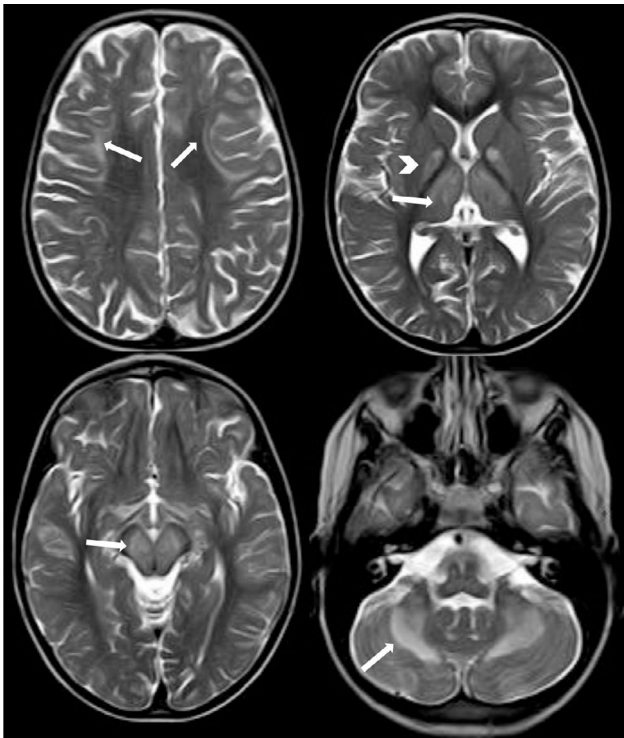


The incidence of these processes is unknown, but is estimated at up to one case per 5000 live births, with the majority being sporadic processes.<sup>2</sup> This syndrome includes 3 overlapping phenotypes: Pearson syndrome, progressive external ophthalmoplegia, and Kearns-Sayre syndrome.<sup>1,3–6</sup>

Kearns-Sayre syndrome is characterised by the development of progressive external ophthalmoplegia, ptosis, and retinitis pigmentosa with onset before the age of 20.<sup>4,6</sup> It is usually associated with such other neurological manifestations as cerebellar ataxia and sensorineural hearing loss. Up to 50% of patients present heart disorders, with the most frequent being conduction disorders. Patients may also display such endocrine manifestations as short stature, hypoparathyroidism, gonadal failure, and diabetes mellitus.<sup>7–9</sup>

Diagnosis is clinical, and based on the identification of the classic clinical characteristics together with one or more of the following factors: high cerebrospinal fluid (CSF) protein levels (> 100 mg/dL), atrioventricular block, or cerebellar

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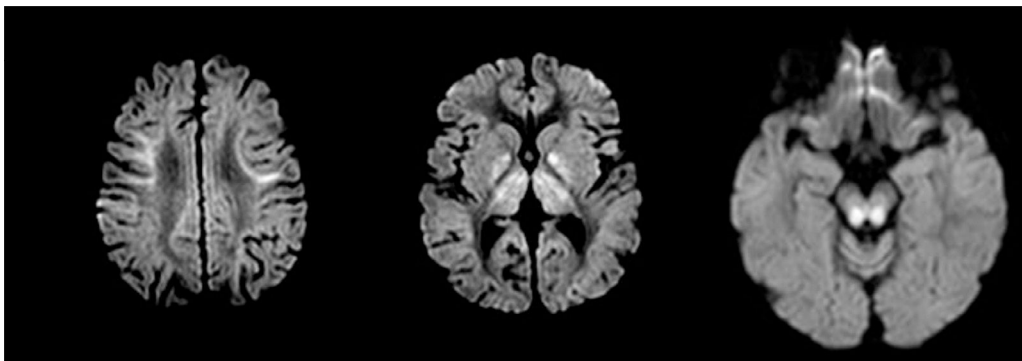
**Figure 1** Axial T2-weighted TSE sequence showing signal hyperintensity in the subcortical white matter (U-shaped fibres), which is more pronounced in the frontal lobes (arrows in the upper left image), globi pallidi (arrowhead in the upper right image) and thalami (arrow in the upper right image), brainstem (arrow in the lower left image), and cerebellar white matter, with bilateral, symmetrical involvement.

ataxia.<sup>5</sup> Muscle biopsy and genetic studies help confirm the diagnosis.<sup>5</sup>

There is currently no specific treatment for this entity, but clinical improvement has been described after the administration of folinic acid to patients with Kearns-Sayre syndrome,<sup>4</sup> since this condition has been associated with cerebral folate deficiency.<sup>4,5</sup>

We present the case of an 8-year-old boy displaying difficulties in gross motor skills and intentional tremor, with onset several months earlier. He had been assessed by the paediatric neurology department at the age of 2 due to initially unilateral ptosis which progressed bilaterally. Neuromuscular disorder was ruled out on several occasions (creatinase kinase and anti-acetylcholine receptor antibody determinations, negative results for anti-MuSK antibodies, and negative Tensilon test). The patient's personal history also included thrombocytopenia secondary to bone marrow hypoplasia, hypoparathyroidism, short stature, and primary adrenal insufficiency. Physical examination revealed bilateral ptosis predominating on the right side, limited facial mimicry, proximal muscle weakness, abolished deep tendon reflexes, and action tremor in both hands, hindering his ability to write. We requested a brain magnetic resonance imaging study, which revealed mild cerebral and cerebellar cortical atrophy with bilateral, symmetrical involvement of subcortical fibres of the supratentorial region; the globi pallidi; the thalami; the cerebellar white matter; and very extensive brainstem atrophy (Fig. 1). Areas of increased diffusion were observed in the cerebellar lesion, with restriction in the other areas, possibly representing more acute involvement (Fig. 2). Given the radiological findings supporting the clinical suspicion of Kearns-Sayre syndrome, we requested a blood mitochondrial DNA study, which detected a single 6.4-Kb deletion and a heteroplasmy rate of 27%, confirming the diagnosis. Laboratory testing revealed a low CSF L-5-methyltetrahydrofolate (5-MTHF) level (1.4 nmol/L; normal range: 47-90); we therefore started treatment with oral folinic acid. Although CSF 5-MTHF levels normalised at one year of treatment, the patient presented no clinical or radiological improvement; unlike the cases reported in the literature, we have observed progressive deterioration of his baseline situation, with muscle weakness and cerebellar ataxia increasing in severity.

During follow-up, he developed bilateral sensorineural hearing loss; complete atrioventricular block, which required an emergency pacemaker implantation; retinitis pigmentosa; and progressive cerebellar syndrome with gait loss.



**Figure 2** Diffusion-weighted sequence (DWI b: 200) showing diffusion restriction in the affected areas.

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E. Pardo Ruiz<sup>a,\*</sup>, D. Maturana Martínez<sup>a</sup>,  
M. Vázquez López<sup>b</sup>, Y. Ruiz Martín<sup>c</sup>

<sup>a</sup> *Servicio de Pediatría, Hospital General Universitario Gregorio Marañón, Madrid, Spain*

<sup>b</sup> *Servicio de Neurología Pediátrica, Hospital General Universitario Gregorio Marañón, Madrid, Spain*

<sup>c</sup> *Servicio de Radiología, Hospital General Universitario Gregorio Marañón, Madrid, Spain*

\* Corresponding author.

E-mail address: [esther.pardo14@gmail.com](mailto:esther.pardo14@gmail.com) (E. Pardo Ruiz).

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## Isolated sixth nerve palsy: an unusual manifestation of preeclampsia<sup>☆</sup>



### Neuropatía aislada del sexto nervio craneal. Una manifestación inusual de preeclampsia

Dear Editor:

Preeclampsia is a pregnancy complication of unknown aetiology with an estimated frequency of 2%–8%. It is characterised by hypertension after week 20 of gestation in women with previously normal blood pressure, together with proteinuria, or in the absence of proteinuria but with presence of thrombocytopaenia, kidney failure, altered liver function, pulmonary oedema, or neurological symptoms.<sup>1</sup> The most frequent neurological symptoms include headache and visual alterations, with isolated oculomotor palsy being exceptional.<sup>2</sup> We present the case of a primiparous woman (37 weeks pregnant) whose first neurological symptom of preeclampsia was sixth nerve palsy; we also assess the possible action mechanisms.

Our patient was a 31-year-old woman assessed in the emergency department due to progressive onset of binocular diplopia in the horizontal plane, with no headache, fever, or other focal neurological signs. The neuro-ophthalmological evaluation revealed 20/20 Snellen visual acuity in both eyes, and the eye fundus examination showed well-defined papillae and spontaneous venous pulsation; the confrontation visual field test yielded normal results; esotropia was observed in the primary gaze position with slightly impaired adduction of the left eye. No other neurological alterations were observed. The general examination detected no fever, blood pressure values of 150/90 mm Hg, and distal oedema in both lower limbs; a blood analysis revealed thrombocytopaenia with 100 000 platelets/ $\mu$ L, increased LDH levels (418 IU/L), and proteinuria (Table 1). In view of these symptoms, and considering the diagnosis of preeclampsia and findings suggestive of isolated sixth nerve palsy, we requested an MRI study before labour was induced. The MRI study ruled out an underlying intracranial lesion, and labour was induced 24 hours after the patient's admission to hospital; a healthy baby was born with no complications. After delivery, the patient presented normal blood pressure values and diplopia progressively improved, fully resolving in 48 hours.

Isolated sixth nerve or abducens nerve palsy during pregnancy is exceptional, although isolated cases have been reported.<sup>3–6</sup> The action mechanism by which preeclampsia causes neuropathy remains unknown, although the absence of other findings and previous published data in the literature suggest that it may cause vasospasm of the vasa

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