M. Sillero Sánchez*, G. García Domínguez and J.J. Asencio Marchante

Servicio de Neurología, Hospital Universitario de Puerto Real, Cádiz, Spain

*Author for correspondence.
E-mail: miriamsillero@telefonica.net (M. Sillero).

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Thalamic ischaemic stroke: an uncommon aetiology of "startle syndrome"

Ictus isquémico talámico: una etiología infrecuente del síndrome de sobresalto

Dear Editor:

Ischaemic stroke is an uncommon cause of movement disorders (MD) that occurs most often when the lesions affect the basal ganglia, thalamus or subthalamus. Although they can be observed in the acute phase, they usually appear deferred after a variable time interval between the stroke and MD onset[1].

While chorea is the most frequent, postictal MD are highly varied and dystonia, hemiballism, asterixis, tremor and myoclonus have been described.

We describe the case of a 56-year-old woman with a personal history of hypertension of long evolution, who was admitted to the emergency room presenting sudden loss of strength in the right hemisphere and sensation of instability. The general examination was normal. The neurological examination objectified right supranuclear facial paresis, right hemiparesis with muscle balance 3/5 in upper extremity and 4+/5 in lower extremity and right touch and pain hemi-hypesthesia, proprioceptive and vibratory. She presented sudden involuntary jerking of the head and right hemisphere, always triggered by auditory and tactile stimuli, which were mostly isolated, but were occasionally grouped in 2-3 second bursts. In the complementary tests, general biochemistry, CBC, lipid study, thyroid function, autoimmunity (ANA, antimitochondrial antibodies, anti-smooth muscle, anti-parietal cells, anti-LKM, anti-aCL IgM, anti-aCL IgG), and CSF examination were normal. The

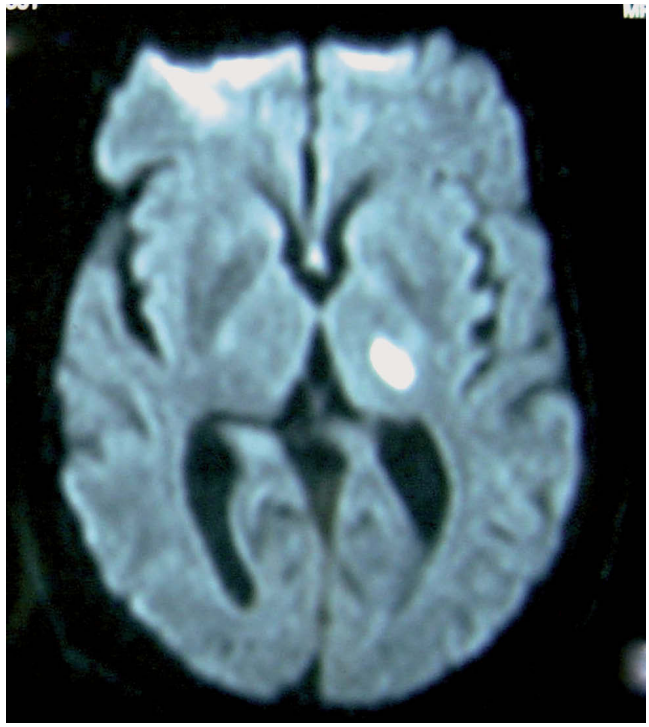

Figure 1 Diffusion sequence of magnetic resonance imaging with hyperintense lesion in the left capsulothalamic region.

electrocardiogram and chest radiography showed no abnormalities. The diffusion sequence of the cranial magnetic resonance revealed a left, capsulothalamic, hyperintense lesion compatible with acute ischemic stroke (fig. 1). Doppler ultrasound of the supra-aortic trunks

showed no significant alterations. The EEG study was normal. The electromyogram showed an excessive response to noise in the retrograde averaging, with no prior cortical edge (fig. 2). Tactile stimulation (not thermal) produced a sharp muscle response that might correspond to subcortical myoclonus, since no prior cortical edge was observed in the retrograde averaging.

The involuntary movements presented by the patient were interpreted as reflex myoclonus, secondary to thalamic stroke. During her admission, in addition to antiplatelet and antihypertensive treatment, we prescribed valproic acid, which achieved an adequate control of symptoms. Three months later, the myoclonus had completely disappeared and medication was gradually reduced until its suppression, with no evidence of recurrence of involuntary movements.

Unilateral MD as a result of focal brain injury are caused by the interruption, at different levels, of the circuits that connect the basal ganglia[1]; cerebrovascular accidents are an uncommon cause of this interruption[1,2]. Different MD have been described after an ischemic or hemorrhagic stroke; the most common being chorea, but dystonias, hemiballism, asterixis and myoclonus can also appear. Most postictal MD are due to a contralateral thalamic lesion[3-6], but strokes with very different topographies have also been described[1,2,7,8].

Myoclonus are involuntary, sudden jerky movements caused by muscle contraction or by its inhibition (negative myoclonus or asterixis)[9]. They have rarely been reported in association with thalamic lesions affecting mainly the ventrolateral nucleus and adjacent structures[7]. Subcortical myoclonus differ from the more common cortical, in that the neurophysiological recording does not show a cortical peak before the muscle response. Subcortical myoclonus include the pathological startle syndrome presented by our patient, which is characterised by an exaggerated startle response to sudden and unexpected stimuli, both tactile and auditory[10]. Its pathophysiology is attributed to the hyperexcitability of the lower brain stem region structures, which are involved in the startle reflex, secondary to the interruption of rubrothalamic pathways that have an inhibitory effect on them[11,12]. There are different forms of pathological startle syndrome: hereditary (in which glycinergic neurotransmission is involved[10]), sporadic and symptomatic. Symptomatic pathological startle syndromes have been associated with strokes and inflammatory lesions of the brainstem, but have also been described after thalamic and subthalamic infarcts, as in our case[12,13].

Three types of pathological startle syndrome have been distinguished based on clinical experience: a first group that would include hyperekplexia, in which a trivial stimulus would produce an exaggerated, inadequate response; a second group that would include startle epilepsy, in which a complex and stereotyped response would appear after the shock; and a third more heterogeneous group, secondary to thalamic injury, posttraumatic stress disorder or other cerebral processes[14] with a response less well characterised, as in the case we have presented.

Postictal MD can occur in the acute phase (as in our observation) or deferred. The period between the stroke and MD presentation varies and, although the mechanisms

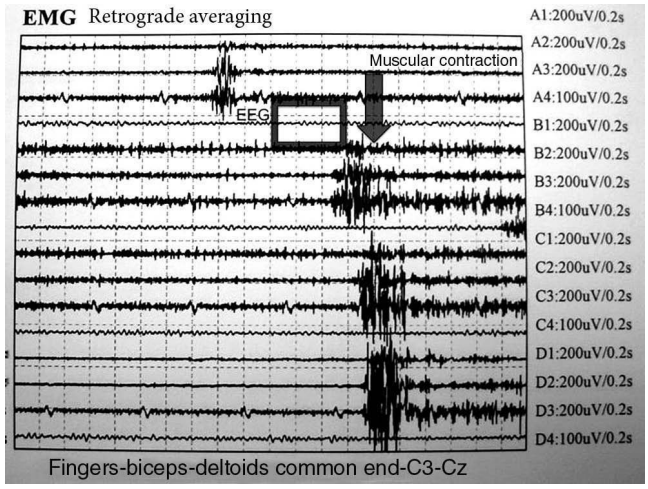

Figure 2 Retrograde averaging without cortical edge prior to muscle shock.

are not fully elucidated, their aetiopathogenesis seems to involve factors including remyelination, inflammatory changes or diaschisis[12].

Although postictal MD may resolve spontaneously, numerous drugs have been used in their treatment, among others, haloperidol, clonazepam and valproate[1]. Clonazepam is the most commonly used treatment in this type of disorders, given its results in hereditary hyperekplexia, a disease in which it is the treatment of choice, because its action on GABA receptors presumably compensates the glycine deficit present in this disease[15].

S. Pérez-Sánchez[a,*], M. Fernández-Recio[a], J.M. López-Domínguez[a], V. Sánchez-Sánchez[a], M. Álvarez-López[b] and G. Izquierdo-Ayuso[a]

[a] *Servicio de Neurología, Hospital Universitario Virgen Macarena, Sevilla, Spain*
[b] *Servicio de Neurofisiología Clínica, Hospital Universitario Virgen Macarena, Sevilla, Spain*

*Author for correspondence.
E-mail: soledad.perez.sanchez@gmail.com (S. Pérez).

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Molar tooth sign: a characteristic image in Joubert syndrome

Signo del molar: imagen característica en el síndrome de Joubert

Dear Editor:

Joubert syndrome (OMIM 213 300) is a rare autosomal recessive disorder, whose locus is on chromosome 9q; it is characterized by ataxia, psychomotor retardation, ocular and respiratory abnormalities related to dysgenesis of cerebellar vermis and mesencephalon. It is currently included in the malformation spectrum of cerebello-oculo-renal syndromes (CORS)[1]. An image known as a "molar tooth sign" is typically observed in cerebral magnetic resonance imaging (MRI)[2].

We report the case of a 2-year-old male, referred with a history of hypotonia and delayed psychomotor development. Physical examination at the genetic consultation found bilateral epicanthal fold, frontal bossing, prominent occiput, triangular upper lip, arched palate, postaxial polydactyly in the left hand, in addition to hypotonia. With these findings, a brain MRI was requested, which showed the classic "molar tooth sign" (fig. 1), by absence of cerebellar vermis, which led to the clinical diagnosis of Joubert syndrome. In complementary studies, the ophthalmology assessment was reported as normal and liver function tests were elevated.

Marie Joubert, a French neurologist, was the first to report this syndrome in five patients who presented breathing disorders and abnormal eye movements, ataxia, mental retardation associated with agenesis of the cerebellar vermis[3]. Maria et al.[4] proposed the diagnostic criteria for Joubert syndrome: hypotonia, ataxia, general delay in developmental and "molar tooth sign".

The "molar tooth sign" is observed in axial neuroimaging cuts, such as cerebral CT and MRI, and is characterised by a deep posterior interpeduncular fossa, thickened and elongated superior cerebellar peduncles, as well as hypoplasia or agenesis of the cerebellar vermis[1].

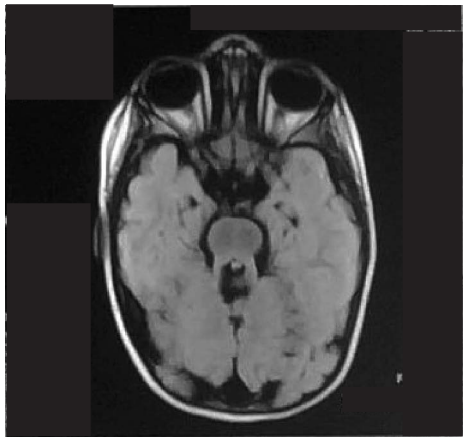

Figure 1 Cerebral MRI showing agenesis of cerebellar vermis and dysgenesis of the mesencephalon, causing the "molar tooth sign" image.

The prognosis of these patients is poor, with a five-year survival rate of only 50%. These patients are more susceptible to respiratory depressant effects of anaesthetic drugs such as opioids and nitrous oxide, so these anaesthetic agents should be avoided[6]. Genetic counselling for this syndrome is necessary because there is a 25% risk of recurrence for each subsequent pregnancy.

P. Hurtado* and H. Pachajoa

Laboratorio de Citogenética, Facultad de Salud, Universidad del Valle, Cali, Colombia

*Author for correspondence.
E-mail: paulamhv@gmail.com (P. Hurtado).