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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-oculorenal 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 vermis³. 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



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

elongated superior cerebellar peduncles, as well as hypoplasia or agenesis of the cerebellar vermis¹.

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 avoided⁶. Genetic counselling for this syndrome is necessary because there is a 25% risk of recurrence for each subsequent pregnancy.

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