

Images in medicine

A characteristic image in Joubert syndrome: molar tooth sign

Mouna Sghir^{1,&}, Wassia Kesomtini¹

¹Unit of Physical Medicine and Rehabilitation, University Hospital Tahar Sfar, Mahdia, Tunisia

[&]Corresponding author: Mouna Sghir, Unit of Physical Medicine and Rehabilitation, University Hospital Tahar Sfar, Mahdia, Tunisia

Key words: Joubert syndrome, molar tooth, aplasia

Received: 15/05/2015 - Accepted: 22/05/2015 - Published: 28/05/2015

Pan African Medical Journal. 2015; 21:69 doi:10.11604/pamj.2015.21.69.7068

This article is available online at: <http://www.panafrican-med-journal.com/content/article/21/69/full/>

© Mouna Sghir et al. The Pan African Medical Journal - ISSN 1937-8688. This is an Open Access article distributed under the terms of the Creative Commons Attribution License (<http://creativecommons.org/licenses/by/2.0>), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.

Image in medicine

Joubert syndrome is a relatively rare autosomal recessive congenital disorder; it is characterized by cerebellar vermis hypoplasia or aplasia. Characteristic clinical symptoms and signs include motor and respiratory abnormalities. It is currently included in the malformation spectrum of cerebello-oculo-renal syndromes (CORS). An image known as a "molar tooth sign" is typically observed in cerebral magnetic resonance imaging (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. We report the case of a 4-year-old male, referred to our rehabilitation unit with a history of hypotonia and delayed psychomotor development. Physical examination found macrocephaly, frontal bossing and triangular upper lip and arched palate. Ocular examination revealed a bilateral divergent squint and inability to track objects with eyes. All aspects of his development were delayed. He had a generalized hypotonia but deep tendon reflexes were normal. There were important negative signs including: Regular breathing pattern, no organomegaly and no polydactyly or syndactyly. With these findings, a brain MRI was requested, which showed the classic "molar tooth sign" which led to the clinical diagnosis of Joubert syndrome. In complementary studies, the audiogram revealed a bilateral sensorineural hearing loss, the ophthalmology assessment and laboratory studies were

normal. We have prescribed a stander and hearing aid. A rehabilitation program was started consisting of: joint mobilization, muscle strengthening, occupational and speech therapy.

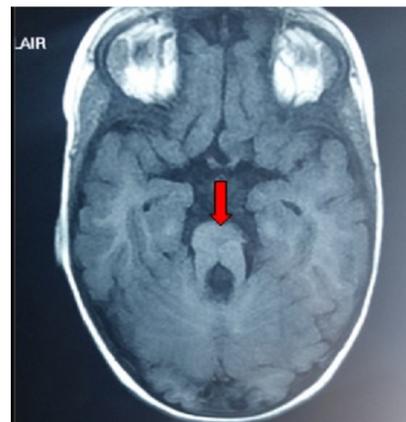


Figure 1: cerebral MRI showing agenesis of cerebellar vermis causing the "molar tooth sign"