‘Molar tooth’ sign in Joubert syndrome

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Received: 2 April 2009 / Revised: 29 April 2009 / Accepted: 10 June 2009 / Published online: 7 July 2009
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An 11-month-old girl born to nonconsanguineous parents presented with abnormal eye movements and frequent upper respiratory infections. The immediate postnatal period had been marked by respiratory distress that subsided with supportive measures. When seen she had global developmental delay, nystagmus, and limb hypotonia. Other systems were normal. Laboratory investigations and chest radiograph were normal. Cranial MRI revealed absence of normal decussation of the superior cerebellar peduncles along with its perpendicular extension to the brainstem. The interpeduncular cistern was deeper with decreased anteroposterior midbrain diameter—

Fig. 1 T1-W axial MR image of the brain

Fig. 2 T1-W sagittal MR image of the brain

findings characteristic of a “molar tooth,” a classic sign of Joubert syndrome (Fig. 1).

Joubert syndrome is characterized by a hypoplastic or absent cerebellar vermis (Fig. 2) and abnormal brainstem. Cardinal features include hyperventilation, muscular hypotonia, oculomotor apraxia, mental retardation and ataxia [1]. Although mostly sporadic, autosomal-recessive inheritance can occur. Treatment is mostly supportive.

Reference

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