Joubert syndrome: A classic case

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Abstract

Joubert syndrome is a rare autosomal recessive disorder characterized by hyperpnoea and eye movements, hypotonia, ataxia, developmental retardation with neuropathologic abnormalities of cerebellum and brainstem including inherited hypoplasia or aplasia of vermis. Cerebellar vermin anomalies are described in other disorders such as Dandy-Walker and rhombencephalon synapsis. These disorders should be distinguished from Joubert syndrome on the basis of imaging. Comparison with typical imaging and clinical findings may be helpful for appropriate diagnosis.

Keywords: Joubert syndrome, molar tooth, open umbrella

Case Report

A 1-year-old female child born to nonconsanguineous parents was admitted with delayed milestones and one episode of seizures. There was no history of birth asphyxia. Pregnancy and delivery were uneventful. The head circumference was normal. There was delay in speech and language development. Abdominal ultrasound was normal. Magnetic resonance imaging (MRI) brain was advised and sent to MRI department for the same. Axial T2-weighted images at the level of mid-brain shows median cleft, which is seen separating the cerebellar hemispheres and communicating with fourth ventricle, producing the typical bat wing (open umbrella) (solid arrow) appearance [Figure 1a].

MRI revealed enlargement of posterior fossa with absence of cerebellar vermis [Figure 1c] and elongated bilateral superior cerebellar peduncle giving molar tooth appearance [Figure 1b]. There was mild colpocephaly seen. Joubert syndrome was hence diagnosed.

Differential Diagnosis

Cerebellar vermin anomalies are described in other disorders such as Dandy-Walker and rhombencephalon synapsis. These disorders should be distinguished from Joubert syndrome on the basis of imaging.[1]

Discussion

Joubert syndrome is a rare autosomal recessive disorder characterized by hyperpnoea and eye movements, hypotonia, ataxia, developmental retardation with neuropathologic abnormalities of cerebellum and brainstem, including inherited hypoplasia or aplasia of vermis, and has a prevalence of less than 1 in 100,000.[1-2] Vermin hypoplasia and abnormalities of the pontomesencephalic junction are the distinguishing features that lead to the diagnosis of Joubert syndrome.[3,4]

The primary MRI features of Joubert syndrome are: (1) small dysplastic or aplastic cerebellar vermis, (2) absence of fiber decussation in superior cerebellar peduncles and cerebellar tracts, (3) abnormal inferior olivary nucleus, and (4) dysplasia and heterotopia of cerebellar nuclei. The
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Posterior fossa typically shows a bat wing-like appearance of fourth ventricle and prominent thickened elongated superior cerebellar peduncles gives characteristic of molar tooth-like appearance. In minority of cases minor lateral ventriculomegaly (6–20% of cases) and corpus callosal dysgenesis (6–10% of cases) are also present.\(^3\)

As the recurrence rate is 25%, prenatal counseling and screening is required.

Prenatal diagnosis of at-risk pregnancies is now possible using serial ultrasounds combined with fetal MRI at 20–22 weeks gestation.\(^5\) These patients are also sensitive to respiratory depressant effects of anesthetic agents such as opiates and nitrous oxide. Hence, the use of these anesthetic agents should be avoided in these patients.\(^6\)

Today, we still do not have a cure therapy for Joubert syndrome, so pediatricians and primary care physicians can help diagnose the disease earlier and offer prenatal diagnosis and genetic counseling to those who have been confirmed by gene mutation. They can advise appropriate physiotherapy and rehabilitation programs that will certainly improve normal motor development and functionality during the treatment processes of Joubert syndrome.

**Conclusion/Summary**

The purpose of this case report is to describe the typical imaging features of Joubert’s syndrome. MRI is the investigation of choice. Cerebellar vermin anomalies are described in other disorders such as Dandy-Walker and rhombencephalon synapsis. These disorders should be distinguished from Joubert syndrome on the basis of imaging. Comparison with typical imaging and clinical findings may be helpful for appropriate diagnosis.

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**Conflicts of interest**

There are no conflicts of interest.

**References**

1. Choh SA, Choh NA, Bhat SA, Jehangir M. MRI findings in Joubert syndrome. Indian J Pediatr 2009;76:231-5.
2. Brancati F, Dallapiccola B, Valente EM. Joubert syndrome and related disorders. Orphanet J Rare Dis 2010;5:20.
3. van Beek EJ, Majoie CB. Case 25 Joubert syndrome. Radiology 2000;216:379-82.
4. Barkovich A. Pediatric Neuroimaging. Philadelphia: Lippincott Williams & Wilkins; 2002. p. 345-6.
5. Doherty D, Glass IA, Siebert JR, Strouse PJ, Parisi MA, Shaw DW, *et al.* Prenatal diagnosis in pregnancies at risk for Joubert syndrome by ultrasound and MRI. Prenat Diagn 2005;25:442-7.
6. Habre W, Sims C, D’Souza M. Anaesthetic management of children with Joubert syndrome. Pediatric Anaesth 1997;7:251-3.