

## A Child with Abnormal Breathing Pattern

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**Abstract**

We describe the characteristic MRI findings of a child who presented in the emergency with breathing difficulty.

**Keywords:** Joubert's Syndrome; Molar Tooth Sign; Batwing Sign; Buttock Sign; Pontomesencephalic Cleft.

**Case Report**

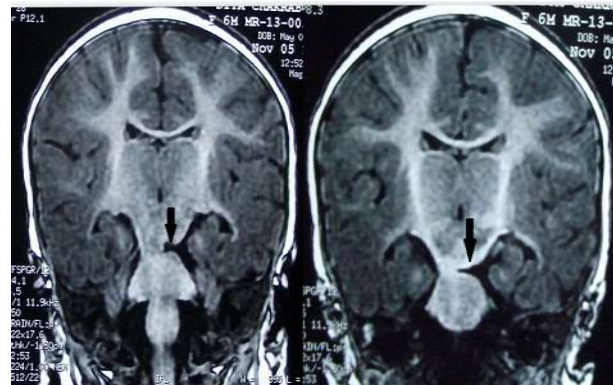
A 6 month old child born at term to non-consanguineous parents presented with delayed milestones and an abnormal breathing pattern. On questioning the parents denied any history of trauma but claimed that the child had episodes of hyperpnoea and bradypnoea since birth which had since decreased in frequency. On examination the child was hypotonic with delayed milestones and pendular nystagmus. There was no cyanosis. Routine blood examinations including serum potassium levels were normal. Chest X ray ruled out any lung pathology.



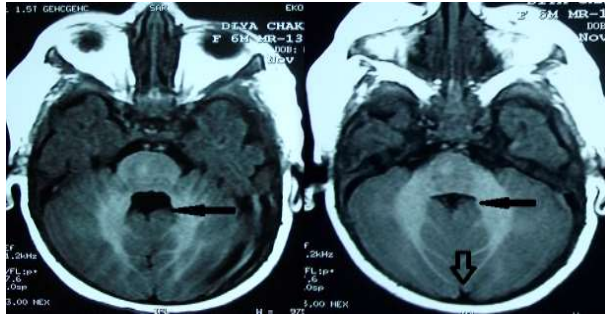
**Fig. 1:** T1 & T2 weighted axial imaging showing 'molar tooth' appearance. The 'roots of the teeth' are formed by the hypertrophied superior cerebellar peduncles (closed red arrows) and the 'crown' is formed by the pons (open red arrows). Hypoplasia of the nuclei on the left side of the pons is also seen.



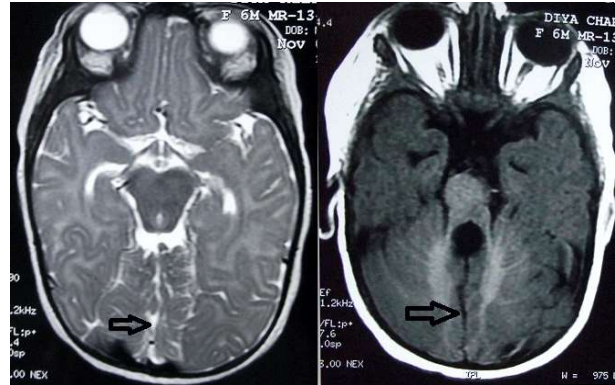
**Fig. 2:** Saggital T2 weighted image showing hypertrophied superior cerebellar peduncle (open arrow) and pontomesencephalic cleft (closed arrow)



**Fig. 3:** Coronal MRI (FLAIR images) showing pontomesencephalic cleft on the left side due to agenesis of the isthmus between the pons and the inferior colliculus (closed arrows).



**Fig. 4:** Axial (FLAIR images) is showing bat wing appearance / umbrella sign of the fourth ventricle due to anterior convexity (closed arrows) and buttock sign (open arrow) due to small cleft between the cerebellar hemispheres consequent to vermian dysgenesis



**Fig. 5:** T2 weighted axial image showing a completely cleft superior vermis at the level of the midbrain. Axial FLAIR image showing cleft vermis at the level of the pons with CSF extending from the fourth ventricle backwards to the cerebellar convexity (both open arrows)



**Fig. 6:** Axial T2 imaging showing deep interpeduncular fossa and part of the 'crown of the teeth' missing due to pontomesencephalic cleft that is caused by the deficient isthmus (closed black arrow)

Echocardiogram was normal. An MRI was done that clinched the diagnosis due to the characteristic findings [Figure 1-6] described below.

#### *Learning Point*

Joubert's syndrome is a genetic disorder described by Marie Joubert in 1969. It is due to developmental anomalies in the cerebellum and brain stem. Associated hepatic and renal abnormalities, seizures and retinal defects may also be found. There is no specific treatment but prognosis depends on the extent of brain malformation.

#### **References**

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