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A rare case of Joubert's syndrome with polymicrogyria

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Abstract

Joubert's syndrome has two classical features- 'Molar tooth sign' and vermian hypoplasia or aplasia. This Case report illustrates such a rare case in a young male which was found to be associated with polymicrogyria. Very few such cases have been described in literature. Other Joubert related disorders and differential diagnosis have also been discussed.

Keywords

Joubert's syndrome; polymicrogyria; vermian; molar tooth.

Introduction

Joubert's syndrome is a complex midbrain- hindbrain malformation characterized by 'molar tooth sign' which is due to thickened and elongated superior cerebellar peduncles. There are other conditions also which can show the same sign and must be differentiated from Joubert's syndrome and related disorders. MRI (Magnetic Resonance Imaging) plays a key role in this. All family members of the affected individual must be offered genetic counselling.

Case presentation

A twenty-four-year-old male presented with history of hypotonia and imbalance in the last three months. Neurological examination yielded positive results for hypotonia and gait ataxia. There was no nystagmus and cognitive functions were normal. There were no features to suggest facial dysmorphism. He had no history of seizures. Ocular testing and ultrasound examination of the abdomen were unremarkable. There was no abnormal breathing pattern on examination.

Plain Magnetic Resonance Imaging (MRI) of the brain at the level of midbrain and superior cerebellar peduncles revealed thickened and elongated cerebellar peduncles giving rise to a 'molar tooth' sign. A deep interpeduncular fossa is noted. Midsagittal T1-weighted MR image demonstrates enlargement of the fourth ventricle with rostral shift of the fastigium. Axial T2 weighted sections showed polymicrogyria.

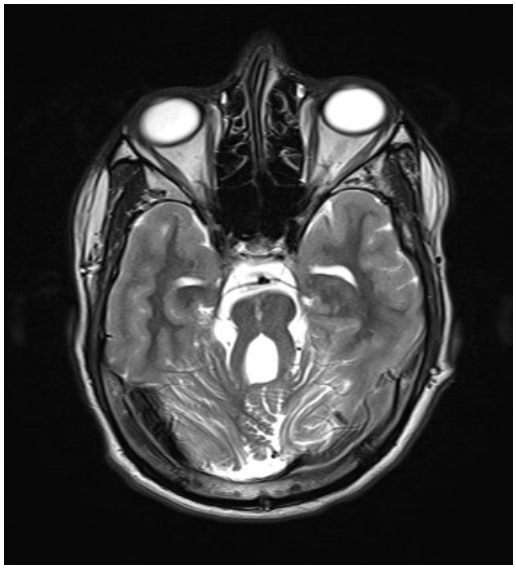

Figure 1: T2 Weighted axial image shows typical neuroimaging findings of 'molar tooth sign' due to thickened and elongated superior cerebellar peduncles and vermian hypoplasia.

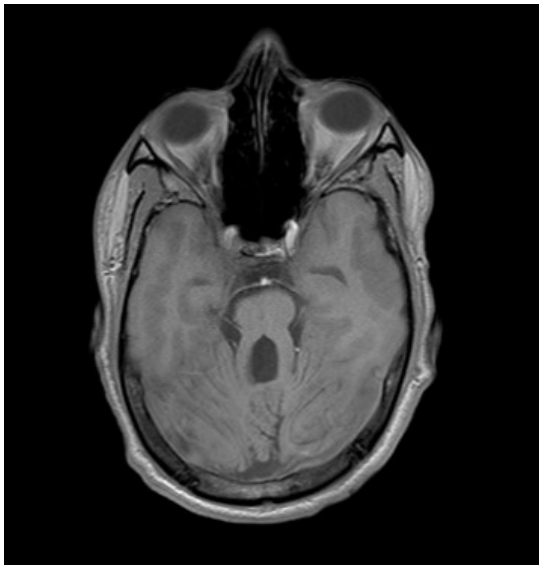

Figure 2: T1 Weighted axial image shows molar tooth sign due to thickened and elongated superior cerebellar peduncles and vermian hypoplasia.

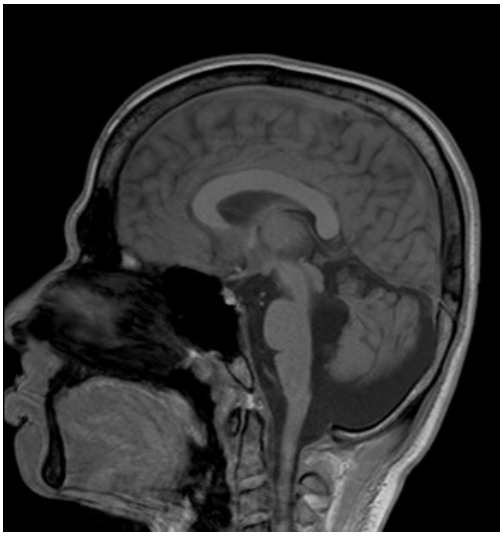

Figure 3: Midsagittal T1-weighted MR image demonstrates enlargement of the fourth ventricle with rostral shift of the fastigium and deep interpeduncular fossa.

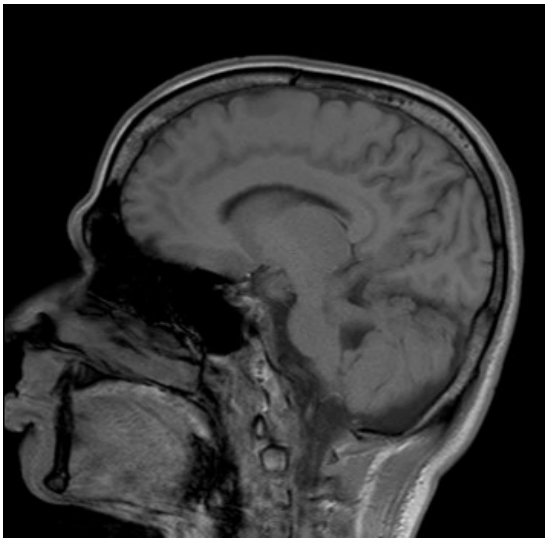

Figure 4: Parasagittal T1-weighted image shows thickened, elongated and horizontally orientated superior cerebellar peduncles.

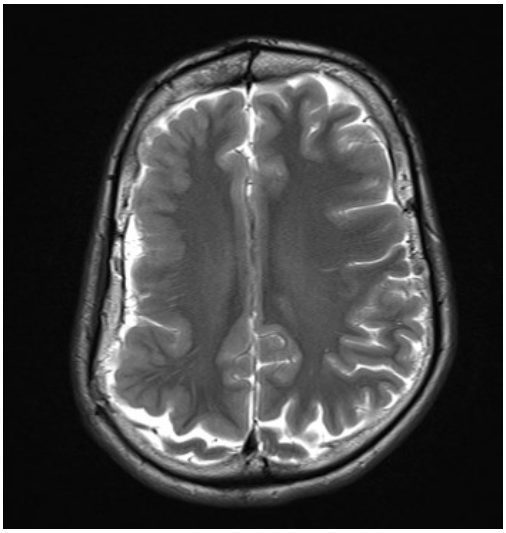

Figure 5: Axial T2 weighted axial image shows polymicrogyria in right frontal and parietal lobes.

Discussion

Joubert Syndrome (JS) is a rare, heterogeneous, midbrain- hindbrain malformation characterized by 'molar tooth sign' with horizontal, thickened and elongated superior cerebellar peduncles. There is aplasia or hypoplasia of cerebellar vermis and abnormally deep interpeduncular fossa. Fourth ventricle can be dilated and may show a 'bat wing' appearance. It shows a genetic overlap with Meckel's syndrome. The term Joubert Syndrome and Related Disorders (JSRD) includes conditions with molar tooth sign such as cerebello-oculo-renal syndrome, Dekaban- Arima syndrome, Orofaciodigital syndrome type VI and Malta syndrome [1]. JSRD shows genetic and clinical similarity with Meckel syndrome which is characterized by encephalocele, posterior fossa anomalies, ductal plate malformation of the liver and polycystic kidney disease [1]. Another overlap can be with Senior Loken syndrome, an oculo-renal ciliopathy, which is characterized by the association of nephronophthisis and retinal dystrophy.

Clinically JS presents with ataxia, hypotonia, oculomotor apraxia, developmental delay and cognitive impairment. Orofaciodigital syndrome type VI shows similar but more severe neuro imaging findings with more frequent supratentorial abnormalities [2]. As JS is associated with multi organ involvement, all patients should undergo a systematic diagnostic workup to rule out systemic involvement [3].

Dilated ventricles and corpus callosal dysgenesis have been described as associated findings in minority of the cases. Retinal dysplasia is highly correlated with renal cystic disease and carries a bad prognosis. Genetic counseling should be carried out. Mutation screening of known gene mutation can identify less than 50% of the cases. Antenatal magnetic resonance of fetal brain also has a role in picking up JS cases [4]. It needs to be differentiated from Dandy Walker malformation and Rhombencephalosynapsis on basis of imaging findings. In Dandy-Walker malformation, there is agenesis or hypoplasia of cerebellar vermis associated with hypoplasia of cerebellar hemispheres. The dilated fourth ventricle opens dorsally into a CSF containing cyst. Thus, the posterior fossa is enlarged with high insertion of tentorium [5]. Posterior fossa size may be variable in JS; an increased retro cerebellar CSF collection can cause it to resemble Dandy Walker malformation. Another differential is COACH syndrome, which includes cerebellar vermis hypoplasia, oligophrenia, ataxia, coloboma and hepatic fibrosis. The lack of oculomotor abnormalities, episodic hyperpnea and hepatic fibrosis in this case differentiates it from the above [1].

Final diagnosis: Joubert Syndrome.

Differential diagnosis: Dandy-Walker variant, Rhombencephalosynapsis, Oral-facial-digital syndrome type VI, Ciliopathies.

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