Joubert syndrome with associated Dandy-Walker complex

A 6-year-old female child with a history of delayed developmental milestones presented with convulsions for 4 months. She was evaluated with a magnetic resonance imaging study of the brain.

**Imaging Findings:**

A large posterior fossa cyst was demonstrated, which communicated with the fourth ventricle. There was hypoplasia of the inferior cerebellar vermis with increased brainstem-vermis and brainstem-tentorium angles. The cyst caused marked anterior compression of both cerebellar hemispheres. There is elevation of the straight sinus, torcular herophili and tentorium (Fig. 1). There is fronto-temporo-parietal atrophy with mild communicating hydrocephalus. (Fig. 3) There is marked atrophy of the mid brain and flattening of pontine belly. (Fig. 1) A widened interpeduncular fossa with 'Molar tooth' appearance is seen on axial images at the level of the mid brain and superior cerebellar peduncles. (Fig. 4) This is associated with partial agenesis of corpus callosum. (Fig. 1, 2, 5).

**Discussion:**

Joubert syndrome is an autosomal recessive cerebellar and brain stem malformation giving a distinctive 'molar tooth' appearance on axial images. It is characterized by aplasia or hypoplasia of the cerebellar vermis, fragmentation or hypoplasia of the dentate nuclei, inferior olivary nuclei, solitary nuclei and tracts, trigeminal nuclei and tracts, and nuclei gracilis and cuneatus with complete absence of pyramidal decussation. [1, 2] The molar tooth appearance is due to a widened interpeduncular fossa with normal or enlarged superior cerebellar peduncles, and middle cerebellar peduncle atrophy. It is characterized clinically by hypotonia and developmental delay. Another entity called as Joubert syndrome and related disorders (JSRD) is used to describe individuals with Joubert syndrome who have additional findings including retinal dystrophy, renal disease, ocular colobomas, occipital encephalocele, hepatic fibrosis, polydactyly, oral hamartomas, and endocrine abnormalities. In a few cases it is associated with lateral ventriculomegaly and callosal dysgenesis. [3]

Dandy Walker complex is a group of disorders which includes the Dandy-Walker malformation, Dandy Walker variant, Blake's pouch cyst, and fourth ventriculocoele. The prevalence of Dandy Walker complex is about 1 in 25,000 to 30,000 births. [4, 5] This represents the morphological spectrum of anomalous cerebellar vermis and adjacent cerebellar hemispheres. The cerebellar vermis is usually hypoplastic with atresia of the fourth ventricle.
outlet, resulting in abnormal fourth ventricular dilatation. The posterior fossa appears expanded which leads to upward displacement of the tentorium and torcular herophili. In Dandy-Walker malformation, there is absence or hypoplasia of the cerebellar vermis which is associated with hypoplasia of the cerebellar hemispheres. The enlarged fourth ventricle opens dorsally into a CSF-containing cyst, following which the posterior fossa is enlarged with a high insertion of the tentorium, consequently the transverse sinuses are higher in position than normal. Associated anomalies include hydrocephalus (75-80%), corpus callosum agenesis (20-25%), heterotopias, polymicrogyria, schizencephaly and occipital cephaloceles. In Dandy Walker variant, there is mild enlargement of fourth ventricle and upward rotation of cerebellar vermis with normal position of tentorium and torcular herophili. [4, 6]

Both of these conditions are present with global developmental delay, convulsion, cognitive impairment with reduced IQ, hypotonia and ataxia.

The diagnosis of Joubert syndrome and Dandy-Walker complex is based on the presence of characteristic magnetic resonance imaging (MRI) findings described above. Joubert syndrome is also diagnosed by genetic studies hence genetic counselling is necessary. [1, 2]

Management is usually symptomatic with treatment of convulsions and surgery for hydrocephalus. Other management includes occupational therapy and physiotherapy.

**Differential Diagnosis List:** Joubert syndrome with associated Dandy Walker Complex., Joubert syndrome related disorders, Retrocerebellar arachnoid cyst

**Final Diagnosis:** Joubert syndrome with associated Dandy Walker Complex.

**References:**

Description: Sagittal T2W image shows a large posterior fossa cyst communicating with fourth ventricle with elevation of straight sinus, torcula herophili, and tentorium, with midbrain atrophy, flattening of pontine belly and hypoplastic inferior cerebellar vermis. Origin: Radiology department, P.D.U. Medical college and hospital, Rajkot.
Description: Sagittal T2W image shows atrophy of fronto-temporo-parietal lobes with dilated lateral ventricle. Origin: Radiology department, P.D.U. Medical college and hospital, Rajkot.
**Description:** Axial inversion recovery image shows "molar tooth" appearance of midbrain. **Origin:** Radiology department, P.D.U. Medical college and hospital, Rajkot.
Description: Coronal T2W image shows partial agenesis of corpus callosum with moose head appearance and mild communicating hydrocephalus. Origin: Radiology department, P.D.U. Medical college and hospital, Rajkot.