ORIGINAL RESEARCH PAPER
Volume - 12 | Issue - 05 | May - 2023 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

CASES IN SPECTRUM OF POSTERIOR FOSSA MALFORMATION

Radio-Diagnosis

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ABSTRACT

A wide spectrum of congenital anomalies affects posterior cranial fossa structures . The purpose of this exhibit is to demonstrate few rare cases in developmental pathologies of the posterior cranial fossa. Predominantly cerebellar involvement (Vermian): 1. Dandy Walker Malformation Cerebellar and brainstem involvement : 1. Joubert, 2. Chiari 2

KEYWORDS

Posterior Fossa Malformation, Radiology , Case series.

INTRODUCTION:

Congenital posterior fossa malformations can result from genetic or disruptive mechanism. Genetic mutations causing malformations may occur de novo or can be inherited from the parents. A disruption on the other hand is defined as a congenital morphologic anomaly caused by the breakdown of an anatomic structure that had a normal developmental potential.

Evaluation of posterior fossa malformation with significant advances in neuroimaging techniques, both pre- and post-natal, have enabled better definition and classification of the same.

Here we demonstrate few cases in spectrum of posterior fossa malformation where correct differential diagnosis played a paramount importance not only for clinical follow up and prognosis of the patient, but also to appropriately counsel the families for inheritance. Thus, for each disorder, we shall emphasize on the key neuroimaging findings that were needed for the diagnosis.

Case Series:

Case 1

The first case was of a 4 year old female child presenting with hypotonia, coarse facial features, global developmental delay and extrapyramidal features . She was a term child born out of consanguineous marriage with an uneventful birth and family history. She had no polydactyly or oro-facial defects on examination.

After routine investigation, an MRI Brain was performed on 1.5T Philips scanner.

MRI revealed thinned and elongated superior cerebellar peduncle of the midbrain with deepened interpeduncular fossa suggestive of Molar Tooth Sign with Vermian hypoplasia. Other findings included multiple discrete areas of grey matter in subcortical white matter and nodular areas in periventricular region as grey matter heterotropia.

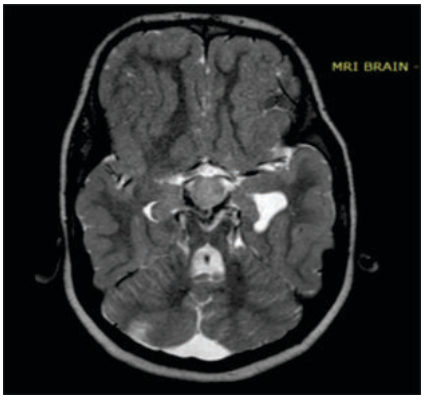

Fig. 1 Thinned & elongated superior cerebellar peduncle of midbrain with deepened interpeduncular fossa.

Molar Tooth sign in Vermian hypoplasia.

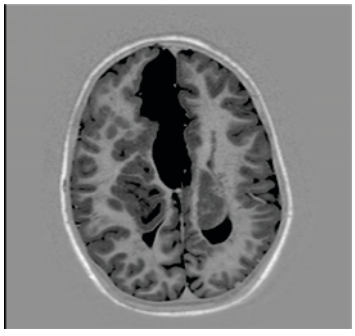

Fig 2: Multiple discrete areas of grey matter in subcortical white matter and nodular areas in periventricular region as grey matter heterotropia.

A Diffusion Tensor Imaging was performed which showed horizontal orientation of superior cerebellar peduncles and absence of decussation of superior cerebellar peduncles.

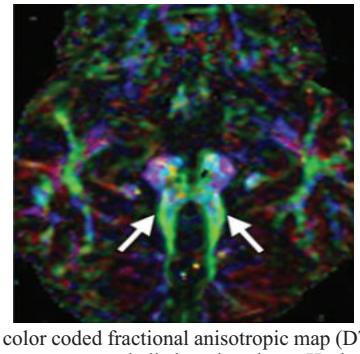

Fig 3. Axial color coded fractional anisotropic map (DTI) obtained at the level of pontomesencephalic junction shows Horizotal orientation of superior cerebellar peduncles (GREEN) (white arrows) and absence of decussation of Superior cerebellar peduncles.

Our diagnosis was Joubert syndrome a.k.a vermian hypoplasia or Molar tooth midbrain-hindbrain malformation Signs and symptoms can vary but commonly include weak muscle tone (hypotonia); abnormal breathing patterns; abnormal eye movements; ataxia; distinctive facial features; and intellectual disability. Dysplasias and heterotopias are common.

Case 2

Second case was a 16-year-old female patient presenting with intermittent episodes of nausea, vomiting and headache. On physical examination, she had movement disturbances. Head circumference was 59 centimeters (cm). She did not have any significant family history of congenital disorders, additionally, her mother did not remember any exposure to drugs or infections during her pregnancy.

After routine examinations , MRI Brain was performed which revealed

a large posterior fossa with a CSF filled cyst extending from fourth ventricle to posterior fossa with absent vermis and hypoplastic cerebellar hemisphere.

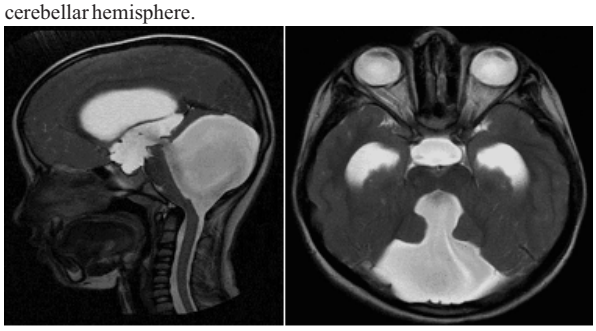

Fig 4: Large posterior fossa with large CSF filled cyst extending from fourth ventricle to posterior fossa with absent vermis and hypoplastic cerebellar hemisphere.

Our diagnosis was Dandy Walker Malformation.
Most common posterior fossa malformation.
Neuroimaging features involve:

a) Hypoplasia of the cerebellar vermis which is elevated and upwardly rotated.
b) Dilatation of the cystic-appearing fourth ventricle which consequently may fill the entire posterior fossa.

Case 3

Third case was a 7 montz old male presenting with depressed gag reflex, involuntary, rapid, downward eye movements, swelling in lower back. Ultrasonography of the swelling revealed a myelo meningocele.

MRI Brain and spine was performed which showed lower lumbar and sacral spinal defect with associated myelomeningocele, MRI brain showed descent of cerebellar tonsils in the cervical canal along with effacement of retro-cerebellar CSF.

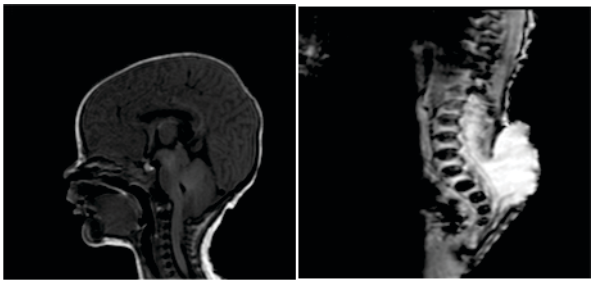

Fig 5:

- Lower lumbar and sacral spinal defect with associated myelomeningocele.
- Descent of cerebellar tonsils in the cervical canal.
- Effacement of retro-cerebellar CSF.

Our diagnosis was Arnold Chiari II malformation which are characterized by myelomeningocele and a small posterior fossa with descent of the brainstem, cerebellar tonsils and vermis through foramen magnum.

Imaging differentials include spinal astrocytoma, Chiari I malformation, Chordoma and Encephalocele.

CONCLUSION:

- Neuroimaging yields detailed anatomic findings and plays a key role in the diagnosis of congenital posterior fossa abnormalities and correct differential diagnosis is of paramount importance not only for clinical follow up and prognosis of patients, but also to appropriately counsel the families for inheritance.
- Well defined neuroimaging based diagnostic criteria are available for the various disorders and should be used by pediatric neurologists, general radiologists, and neuroradiologists.
- Diffusion Tensor Imaging is a promising method for characterizing microstructural changes or differences with neuropathology.

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