

Clinicoradiological aspects of pontine tegmental cap dysplasia: Case report of a rare hindbrain malformation

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Abstract

Malformations involving the brainstem are very rare and present with a varied spectrum of clinical symptoms due to multiple cranial nerve palsies and pyramidal tract involvement. Of these, pontine tegmental cap dysplasia is a very unusual malformation, characterized by ventral pons hypoplasia and an ectopic dorsal band of tissue, projecting into the fourth ventricle, from dorsal pontine tegmentum. A 4-year-old male child, presenting with left facial nerve palsy, revealed hypoplastic ventral pons and an ectopic structure on magnetic resonance imaging (MRI). The ectopic structure was isointense to pons, arose from the left side of dorsal pontine tegmentum, at pontomedullary junction and protruded into the fourth ventricle, impinging upon the left seventh and eighth cranial nerves. Diffusion tensor imaging (DTI) depicted abnormal white matter tracts in ectopic tissue with absent transverse pontine fibres and abnormal middle and superior cerebellar peduncles. The typical MRI appearance, coupled with DTI, helped us reach an accurate diagnosis of pontine tegmental cap dysplasia, in a setting of neurological dysfunction.

Key words: Brainstem malformations; cranial nerve palsies; hindbrain malformations; pontine tegmental cap dysplasia

Introduction

Brainstem malformations are extremely rare and account for varied clinical symptoms owing to the presence of multiple cranial nerve palsies and pyramidal tract involvement.^[1] Pontine tegmental cap dysplasia (PTCD) is one such under-explored and unfamiliar hindbrain malformations, characterized by hypoplastic ventral pons, with an ectopic dorsal pontine white matter band (called as “tegmental cap”), protruding into the fourth ventricle from the tegmentum of pons.^[2] Here, we report an interesting case of this rare pontine malformation and discuss the associated clinical and radiological features.

Case History

A 4-year-old male child, born out of a non-consanguineous marriage, through a normal vaginal delivery, presented with facial nerve palsy, deviation of the angle of mouth towards the left, drooling of saliva from the left angle of mouth and abnormal watering from the left eye for the last 1 year. There was no history of any speech or swallowing disturbances. No family history of any neurological deficit was available.

The patient was referred to us for imaging by a pediatric neurologist, to ascertain the cause of facial palsy. Magnetic

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resonance imaging (MRI) was performed using 1.5T scanner (Phillips, Achieva). Axial, coronal and sagittal T1 and T2-weighted images were obtained. An axial heavily T2-weighted DRIVE sequence was acquired for better demonstration of cranial nerves. In addition, diffusion tensor imaging (DTI), with generation of fractional anisotropy (FA) color maps was done, on a 3T MRI scanner (Phillips, Ingenia) using 32 icosahedral directions. Other parameters used were TE 110 ms, TR 3985 ms, FOV 200–230, slice thickness 3 mm and image matrix 256 × 256.

On axial T1/T2-weighted MRI, we observed hypoplastic left side of ventral pons [Figure 1A]. The ventral pons appeared flattened on the corresponding sagittal images [Figure 1B]. On axial and sagittal T1/T2-weighted MRI, an ectopic aberrant structure, isointense to the pons, was noted arising from the left side of dorsal pontine tegmentum, at the pontomedullary junction, protruding into the fourth ventricle posteriorly and inferiorly [Figure 2A-C]. On heavily T2-weighted axial DRIVE sequence, this aberrant exophytic dorsal pontine tissue was seen to impinge upon and compress the left seventh and eighth cranial nerves at their origin [Figure 3]. However, bilateral inner ear structures and internal auditory meati appeared normal.

Additional MRI features observed were abnormal morphology and slight lateral orientation of the superior cerebellar peduncles [Figure 1A and 4A]. The middle cerebellar peduncles appeared hypoplastic on the right side [Figure 4B]. However, inferior cerebellar peduncles were observed to be normal. Cerebellar vermis, cerebellar hemispheres, and medullary olivary nuclei were also normal. Supratentorial structures appeared normal in morphology.

DTI, with the cross-sectional FA color maps depicted elongated and laterally oriented superior cerebellar peduncles [Figure 5A]. The middle cerebellar peduncles appeared hypoplastic, with paucity of white matter tracts, markedly on the right side [Figure 5B]. An abnormal bundle of white matter axons, directed anteroposteriorly and depicting green color on color maps, was noted in the dorsal exophytic band of tissue [Figure 5C]. Significantly, the transverse pontine fibers were observed to be virtually absent [Figure 5D]. The normal transverse pontine fibers in a healthy control are shown in Figure 5E.

Besides MRI, whole spine radiographs were also obtained to exclude any associated vertebral anomalies, which revealed no obvious abnormality.

Discussion

The term PTCB was coined for the first time in literature by Barth *et al.*^[2] This is a rare, nonprogressive disorder, characterized by developmental delays, multiple cranial nerve deficits and a unique hindbrain malformation.

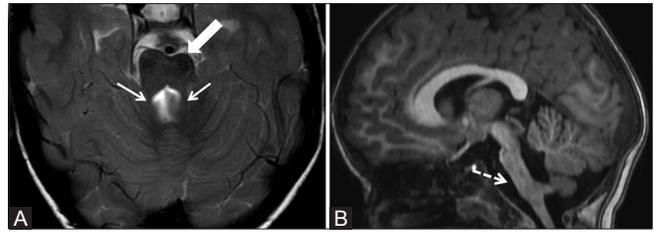


Figure 1 (A and B): Axial T2-weighted image (A) shows hypoplastic left side of ventral pons (thick white arrow), with abnormal lateral orientation of superior cerebellar peduncles (thin white arrows). Sagittal T1-weighted image (B) shows flattened ventral pons (dotted white arrow)

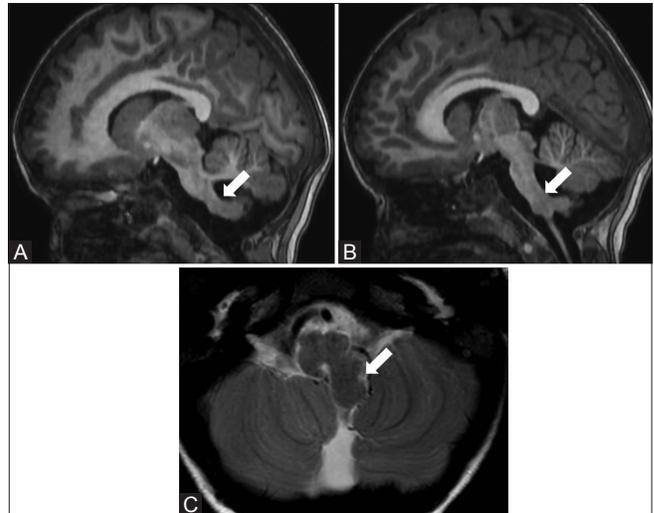


Figure 2 (A-C): An aberrant exophytic structure (thick white arrows) is seen protruding into the fourth ventricle from the dorsal pontine tegmentum at pontomedullary junction on corresponding sagittal T1-weighted (A and B) and axial T2-weighted images (C)

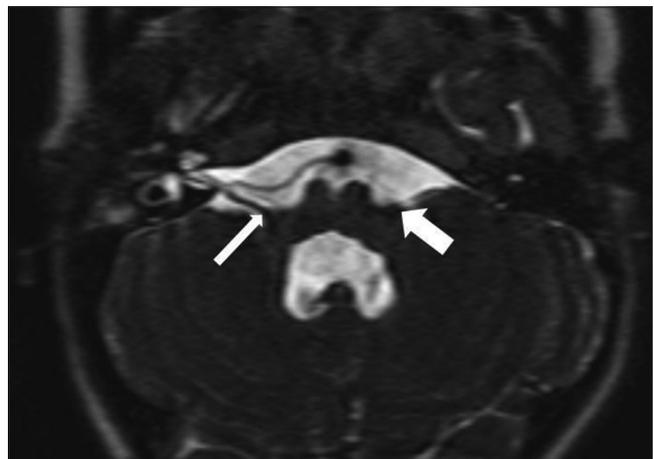


Figure 3: Axial DRIVE sequence shows normal origin of right 7th and 8th cranial nerves (thin white arrow), with impingement at the origin of left 7th and 8th cranial nerve (thick white arrow)

Associated clinical features include sensory neural hearing deafness, ataxia, speech disturbances, facial palsy, spine anomalies, swallowing problems and failure to thrive.^[1,2] The exact etiology of this disorder is yet unexplained, with a possibility of having a genetic origin.^[1,3]

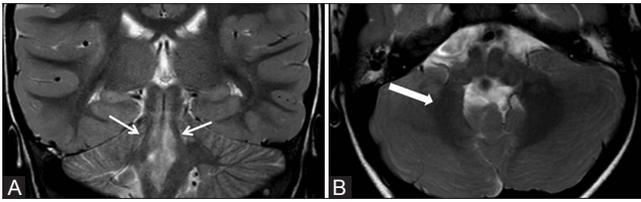


Figure 4 (A and B): Coronal T2-weighted image (A) shows elongated and laterally oriented superior cerebellar peduncles (thin white arrows). Axial T2-weighted image (B) shows hypoplastic right middle cerebellar peduncle (thick white arrow)

MRI plays an essential role in adequate assessment of this rare brainstem malformation.^[1-7] The abnormally hypoplastic ventral pons in conjunction with an exophytic ectopic dorsal tissue projecting from the pontine tegmentum are the key radiological features. Other associated MRI findings are total or partial absence of middle cerebellar peduncles, abnormal morphology and orientation of superior cerebellar peduncles, hypoplastic inferior cerebellar peduncles, distorted appearance of medulla oblongata with absence of medullary olives and hypoplastic cerebellar vermis and/or cerebellar hemispheres.^[1,2]

The abnormal superior cerebellar peduncles, with MRI appearance of a “molar tooth,” at times pose difficulty for the radiologist to differentiate it from Joubert syndrome. However, other typical MRI features seen in PTC, coupled with cranial nerve palsies, helps in distinguishing the two entities.^[2]

In our case study, MRI and DTI revealed similar features described by previous investigators, including virtually absent normal transverse pontine fibres and abnormal white matter axons in the ectopic pontine tegmental cap, that were directed in the anteroposterior direction and depicted green color on FA images.

In the largest reported series of 16 patients reviewed retrospectively and published recently in 2016 by Nixon *et al.*, the authors found associated duplication of one or both internal auditory canals in all but one patient.^[8] Associated cochlear nerve aplasia and either atresia or stenosis of internal auditory canals was also observed in their study. However, our patient did not reveal any temporal bone and cochlear nerve abnormality.

The possible pathogenesis of this rare condition can be explained by abnormal axonal guidance and/or neuronal migration as evidenced by absence of transverse pontine fibres, absent superior cerebellar peduncles decussation and ectopic pontine fibres. All these findings can be better demonstrated on DTI. Jissendi-Tchofo *et al.* hypothesized the development of this rare condition at molecular level in one of the three possible manners. (1) The reduced ventral migration of pontine nuclei may lead to aberrant dorsal pontine white matter fibres. (2) Increased radial migration

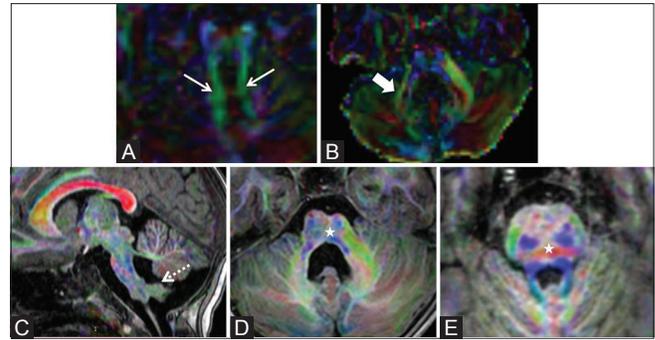


Figure 5 (A-E): Axial FA color images show elongated and laterally oriented superior cerebellar peduncles (A) seen as green (thin white arrows) and hypoplastic right middle cerebellar peduncle (B) (thick white arrow). Sagittal fractional anisotropy color DTI image (C) shows anteroposteriorly directed white matter axons in ectopic dorsal pontine tissue seen as green (dotted white arrow). Axial fractional anisotropy color DTI images show near complete absence of transverse pontine fibers (D) and normal transverse pontine fibers in a healthy control as red (E) (white stars)

of pontine gray neurons which are then relocated on the surface of pontine tegmentum in an abnormal manner. (3) Lastly, normal pontine neuronal migration followed by abnormal axonal guidance away from pontine ventral surface.^[2]

Prognostic correlation of neuroimaging findings in PTC has also been documented. The degree of brainstem dysplasia on imaging correlates well with clinical disability. A rounded bump (so called tegmental cap) reflects a mild disease, whereas a beak-shaped angular brainstem kink, reflects severe form of the disease with poorer outcome.^[9]

PTCD has also been included under the umbrella term “pontocerebellar hypoplasia.” However, an autosomal recessive inheritance in pontocerebellar hypoplasias and an ectopic dorsal pontine tissue in PTC aid in adequate differentiation between the two diagnoses. Besides Joubert syndrome, PTC should also be differentiated from Moebius syndrome. The characteristic MRI findings and substantial developmental delay, seen invariably in all cases of PTC, help in distinguishing it from Moebius syndrome.^[1,2,4,5] In addition, Moebius syndrome is characterized by the presence of bilateral combined 6th and 7th nerve palsies, which is not seen in PTC. Moreover, the differentiating MRI features of Moebius syndrome include predominant hypoplasia of the dorsal pons with absence of medial colliculus at the pontine level, causing depression in the fourth ventricle and absent hypoglossal prominence.^[5]

Although similar clinical and imaging features of PTC have been published previously by a few investigators, a PubMed search did not reveal any report from the Indian investigators. Our study, thereby adds to the literature in describing the radiological and clinical features of this rare pontine malformation.

We conclude that the peculiar hallmark MRI appearance of PTCB, together with the abnormal axonal findings on DTI, can help radiologists, in reaching an accurate diagnosis in an appropriate clinical setting of neurological dysfunction.

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Conflicts of interest

There are no conflicts of interest.

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