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Case Report

Pontine tegmental cap dysplasia with a duplicated internal auditory canal

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ABSTRACT

Pontine tegmental cap dysplasia (PTCD) is a rare neurological syndrome that results in a hypoplastic ventral pons, tegmental cap at the dorsal pons, and cranial nerve dysfunction. The most common symptoms are hearing loss and speech problems. We present a case of a 9-month-old male who presented with developmental delay and hypotonia. Magnetic resonance imaging revealed ectopic dorsal transverse pontine fibers and a cap-like protrusion of the dorsal pons. Diffusion tensor imaging showed that the ventral pontine fibers were absent. The cause of PTCD is undiscovered, but proposed hypotheses include dysfunction in axonal guidance, neuronal migration, and ciliary protein function. PTCD is a rare neurological disorder, but the diagnosis can be suggested with MRI using diffusion tensor imaging as an aid.

Introduction

Pontine tegmental cap dysplasia (PTCD) is a rare neurological syndrome with a distinct hindbrain malformation and cranial nerve (CN) dysfunction [1]. Approximately 40 cases have been reported [1–8]. Magnetic resonance imaging (MRI) with diffusion tensor imaging (DTI) plays an essential role in diagnosing PTCD [9].

PTCD is characterized by hypoplasia of the ventral pons and a protrusion of the dorsal pons from the tegmentum into the fourth ventricle. Varying deficits in CN V, VI, VII, VIII, and IX [1] are known. The symptoms vary in scope and severity among patients ranging from mild cognitive impairment to severe dysfunction, but most patients present with deafness and speech difficulty [1–10].

The treatment and prognosis of PTCD depends on the scope and severity of symptoms. Cochlear implants have treated hearing and speech problems with variable efficacy and have been shown to improve quality of life in some patients [11–13]. Case reports have been described in patients

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aged 6 months to more than 48 years [5,14]. The spectrum of PTCD is wide, necessitating individualized treatment depending on the symptoms.

The cause of PTCD is unknown, but several explanations have been proposed. These include defects in axonal guidance, neuronal migration, and ciliary proteins [7]. While a genetic component is likely, there is no specific genetic test and no known inheritance pattern [7].

Because PTCD is a very rare condition, it is important to describe more cases to raise awareness, strengthen knowledge, and facilitate radiographic diagnosis of the disorder.

Case report

A 9-month-old Hispanic boy presented with developmental delay and hypotonia. He did not start rolling over until 8 months and could not sit up independently at the time of presentation. He could transfer toys from hand to hand, smile, laugh, and babble. Cerebellar hypoplasia had been diagnosed prenatally by ultrasound. Bilateral sensorineural hearing loss was uncovered at 7 months via audiometry, and he was known to have dry eyes and corneal ulcers diagnosed at 7 months. He was fed by gastrostomy due to poor oral intake. Additional comorbidities included an imperforate anus with rectovesicular fistula, atrial septal defect, lumbar vertebral segmentation anomalies, and fusion of several ribs.

On physical exam, his muscle tone was found to be hypotonic, but his CN exam was grossly normal. Healed corneal ulcers with stitched outer epicanthal corners were appreciated.

MRI without contrast and DTI of the brain were performed. T1-weighted images demonstrated a posterior bulge of the midportion of the pons which partially effaced the fourth ventricle, an abnormally diminutive pons, and accentuation of the pontomesencephalic fissure (Fig. 1). The corpus callosum was mildly thinned. DTI showed an ectopic transverse fiber tract along the dorsal pons (Figs. 2 and 3). The bilateral hippocampi were incompletely rotated (Fig. 4). The diagnosis of PTCD was assigned based on these characteristic imaging findings and concordant clinical history, specifically, the morphology of the brainstem and ectopic transverse fiber tract across the dorsal pons.

Subsequent MRI of the internal auditory canals demonstrated bilateral absent cochlear nerves and a duplicated internal auditory canal on the right (Fig. 5). Cochlear implantation was not attempted due to the lack of cochlear nerves.

The patient underwent posterior sagittal anorectoplasty and remains developmentally delayed. While his chronic conditions will require long-term management, his prognosis is favorable. He will need special education and speech therapy as he gets older.

Discussion

The term PTCD was coined by Barth, et al. in 2007 in his report of 4 cases [10], though 2 potential cases of PTCD with similar hindbrain malformations and clinical findings were reported prior to 2007 [15–16].

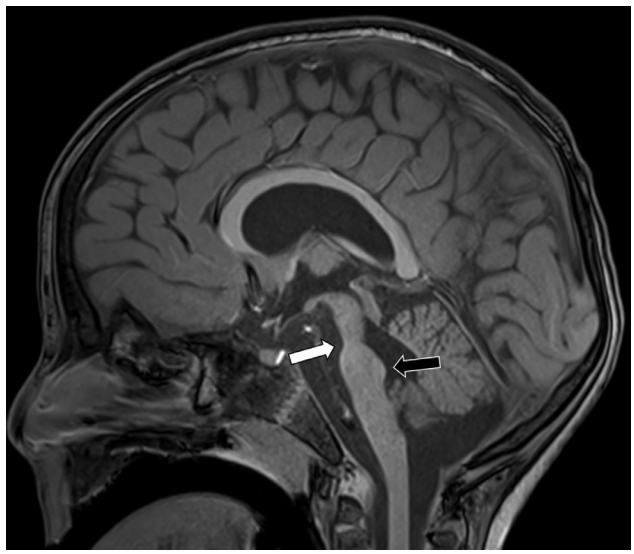

Fig. 1 – Sagittal T1-weighted image of the brain demonstrates a diminutive pons with an exaggerated pontomesencephalic fissure (white arrow). A dorsal bulge of the pons protrudes into the fourth ventricle (black arrow).

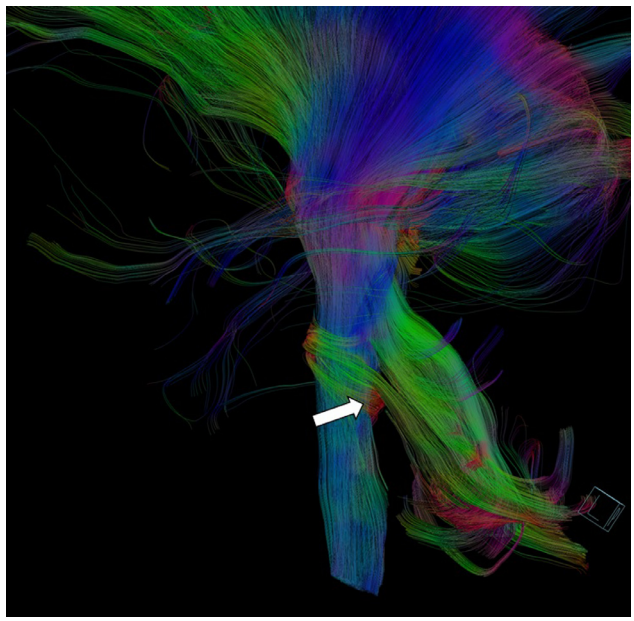

Fig. 2 – Fractional anisotropy (FA) map shows an abnormal transverse fiber bundle (arrow) throughout the pontine bulge described in Fig. 1.

On MRI, the hindbrain malformation is described as a flattened, hypoplastic ventral pons with a tegmental cap, a curved protrusion of the dorsal pons from the middle third of the tegmentum into the fourth ventricle [17]. The protrusion has been characterized as cap-like or beaklike [14]. There is often absence or hypoplasia of the middle or inferior cerebellar peduncles which sets this apart from Moebius syndrome, another neurological disorder that may present similarly [9,17].

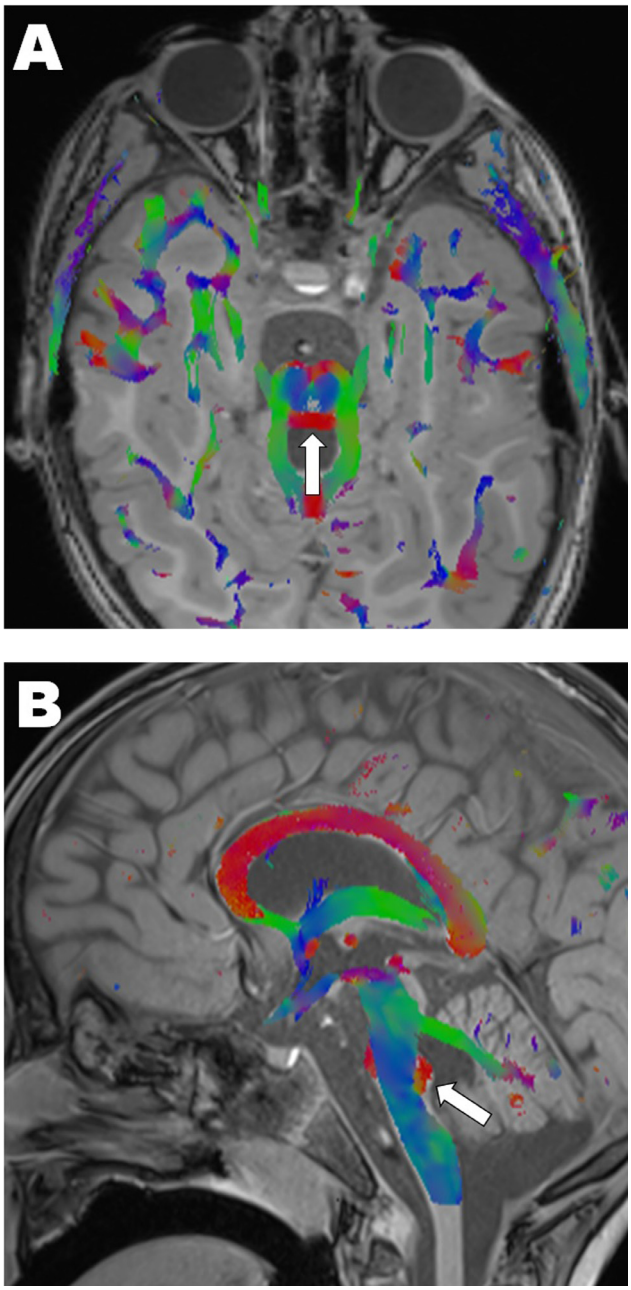

Fig. 3 – The FA map is superimposed on the axial (A) and sagittal (B) T1-weighted images with the abnormal transverse pontine fibers depicted (arrows).

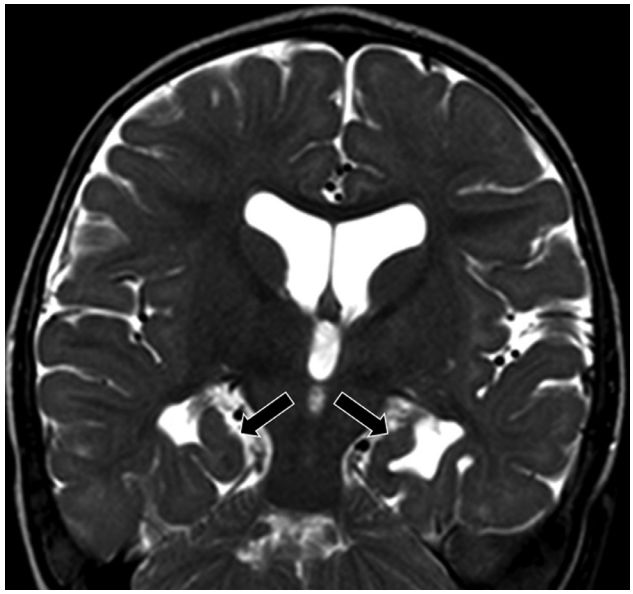

Fig. 4 – Coronal T2-weighted image reveals incompletely rotated hippocampal heads (arrows).

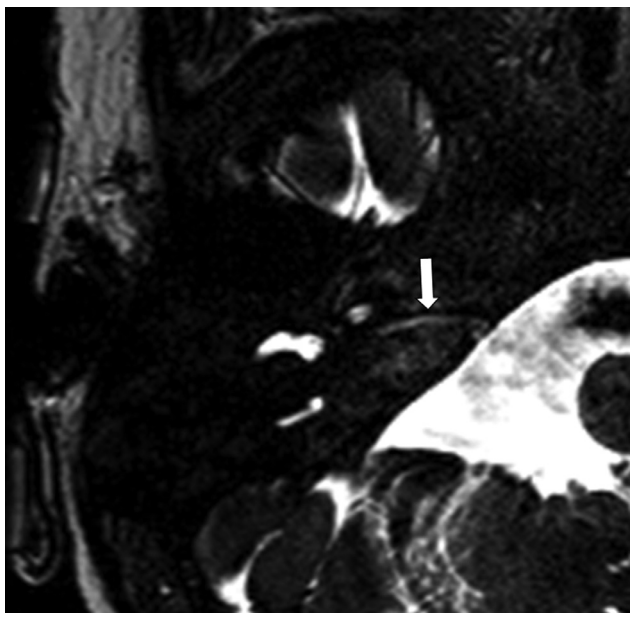

Fig. 5 – The facial nerve is present within a duplicated internal auditory canal (IAC) on this axial heavily T2-weighted image of the right IAC.

Additional imaging findings include a molar tooth appearance of the pontomesencephalic junction, absent inferior olivary prominences, and variably absent CNs V, VI, VII, VIII, and IX [1,17]. In a study by Nixon et al., 94% presented with duplication of at least 1 internal auditory canal, with vestibulocochlear nerve canal stenosis or atresia in the duplicated internal auditory canals and ipsilateral vestibulocochlear nerve aplasia [8]. While many of these imaging findings are present in PTCD, the most specific characteristic is the tegmental cap [17].

DTI reveals ectopic transverse fibers in the tegmental cap of the dorsal pons which may or may not be connected to the middle cerebellar peduncles [7,18–19]. There are also absent or abnormal transverse fibers in the ventral pons and absent or reduced decussation of the superior cerebellar peduncles, possibly contributing to the hypoplasia of the ventral pons [7].

The symptoms of PTCD vary in scope and severity. When it affects the trigeminal nerve (CN V), it can cause decreased facial sensation and corneal ulceration. When the abducens

nerve (CN VI) is affected, paralytic strabismus can ensue. Facial nerve (CN VII) involvement results in decreased facial expression and taste. The most common CN to be affected is the vestibulocochlear nerve (CN VIII) which can cause deafness.

Gastrointestinal defects such as dysphagia or gastroesophageal reflux have been reported in other cases [12–20]. The case presented here uniquely depicts an association with imperforate anus and severe gastrointestinal dysfunction.

Cochlear implantation has been reported in 4 patients, and 3 showed at least moderate improvement in hearing and speech while 1 showed minimal response [11,13]. Vestibulocochlear nerve aplasia may decrease the response to cochlear implantation, and as in our case, MRI to evaluate the status of the CNs may help to determine candidacy for cochlear implantation [8].

Three etiologies have been advanced for PTCD: axonal guidance, neuronal migration, and ciliary dysfunction. Barth et al. suggested dysfunctional axonal guidance because experiments in mice showed that inactivation of the *NTN1* or *DCC* genes that guide axons results in apoptosis of pontine neurons and an aberrant commissure [10]. However, genetic testing of these patients did not reveal a reproducible variation in these genes [10]. Jissendi-Tchofo, et al. argued that defective neuronal migration may result in an imbalance of neurons with fewer in the ventral pons and more in the tegmentum because the *UNC5H3* receptor of *NTN1* is also involved in neuronal migration [14]. In the only case with neuropathological data, there was pontine, dentate, and olivary dysplasia [7]. This supports the neuronal migration hypothesis because these neurons all originate from the rhombic lip. Jissendi-Tchofo also proposed abnormal ciliary proteins as seen in Joubert syndrome, because brain imaging in both PTCD and Joubert syndrome are associated with the molar tooth sign [14]. In 1 case, there was a 2q13del mutation in the *NPHP1* gene which is related to Joubert syndrome type 4 [20].

PTCD is a rare neurological syndrome in children characterized by a distinct hindbrain malformation and CN deficits.[1] This case of PTCD, diagnosed on MRI and DTI, shows the value of imaging in making the diagnosis as well as the need to increase familiarity and understanding of PTCD.

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