Two cases with Pontine Tegmental Cap Dysplasia: A rare hindbrain anomaly which may be misdiagnosed as Moebius Sequence

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INTRODUCTION

Pontine tegmental cap dysplasia (PTCD) is a rare hindbrain anomaly characterized by ventral pons hypoplasia, cap-like structure of the dorsal upper pons protruding towards the fourth ventricle. All reported cases are sporadic, and no specific genetic cause has been yet identified (1).

OBJECTIVES

PTCD shares similar neuroradiologic findings with Moebius sequence (MBS) and pontocerebellar hypoplasia; and it has been suggested that PTCD may be more common than realized, and patients may be misdiagnosed with MBS or pontocerebellar hypoplasia.

Here, we aimed to define the clinical and radiologic findings of two patients with PTCD.

RESULTS

Patient 1: She is a 14-year-old girl. She was born by cesarean section as a twin with a birthweight of 2450 g at 38 weeks of gestation, and was hospitalised in NICU for one month for feeding problems. She had bilateral sensorineural hearing loss, and had no meaningfull words. She had multiple operations for corneal ulcers. She also had funtional one kidney, and a history of vesicoureteheral reflux in the atrophic kidney. She could only take liguid food. Examination revealed severe cognitive impairment, spastic diparesis, and multiple cranial nerve deficits including bilateral V, VII, VIII, IX and X. Corneal reflex was absent, and uvula was not observed. Mouth opening was limited.

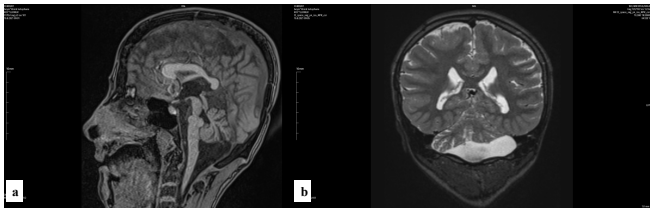

Figure 1: Cranial magnetic resonance imaging of patient (a-Sagittal T1 sequence, b-coronal T2 sequence) shows dysplasia of corpus callosum, flattening of ventral pons, cerebellar atrophy, and cap like structure, protruding to fourth ventricule

RESULTS

Patient 2: He is a 3.5-year-old boy. He was born small for gestational age (2600 g) by cesarean section at 39 weeks of gestation, hospitalized for a week after delivery due to respiratory distress and feeding difficulty, and discharged with a nasogastric tube. Facial asymmetry was present from birth, and he was diagnosed with right peripheral facial palsy. At the age of 6 months, sensorineural hearing loss was detected. The cochlear implant was placed at 14 months of age, but did not provide any clinical benefit. He was hospitalized several times with the complaints of feeding difficulties, vomiting, and poor weight gain, and Nissen fundoplication was performed with the diagnosis of gastroesophageal reflux.

He presented to our clinic, for severe hypertension. He had a pulse rate (PR) 120/min, respiratory rate 18/min, BP 124/86 mm Hg. He had generalized hypotonia, global developmental delay, and multiple cranial nerve deficits including bilateral V, VI, unilateral VII, bilateral VIII, IX, and X. Both axial and appendicular hypotonia was present. He could not sit without support. Pyramidal signs were positive.

The patient's blood pressure (BP) was labile with significant BP elevations or surges with parallel increases in pulse rate (PR) that lasted for hours, accompanied with restlessness, sweating, and headache. Differential diagnosis included pheochromocytoma or paraganglioma, producing catecholamines, symptomatic with flushes, sweating, and palpitations, leading to severe episodic or persistent arterial HTN. However, plasma catecholamine concentrations were normal and abdominal computed tomography (CT) showed no pathological masses. We assumed that the cause of volatile HTN in our case was "afferent baroreflex failure" due to CN XI and/or CN X dysfunction.

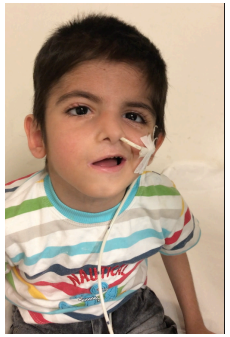

Figure 2

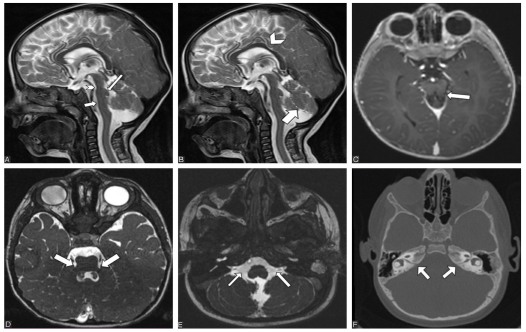

Figure 3

CONCULSIONS

PTCD is a rare anomaly which may be misdiagnosed as Moebius sequence or pontocerebellar hypoplasia. Pons hypoplasia may be present in both of these disorders; but distinctive radiologic findings including cap-like structure of the dorsal upper pons protruding towards the fourth ventricle is specific to PTCD.

The findings in our patient including hypotonia (75.4%), develop- mental delay (70.3%), swallowing di culties (67.2%), CN V palsy (25%), CN VIII (30.8%), CN IX (43%), CN X (53%), pons hypoplasia (60%), and cerebellar vermis hypoplasia (5%) were all reported in MBS

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Figure 1: Patient 2 at the age of 3 years and 9 months. The nasolabial sulcus was at on the right. A nasogastric tube was inserted for oromotor dysfunction and malnutrition

Figure 3: Cranial MRI and CT findings of patient 1 at one years old: **a)** Midsagittal T2-weighted image shows flattening of the ventral pons (thick arrow). The dorsal upper pons has a cap-shaped dysmorphic appearance and protrudes towards the fourth ventricle (thin arrow). Mesencephalon pons junction level (isthmus) is hypoplastic (arrowhead). **b)** In sagittal T2-weighted series, hypoplastic cerebellar vermis separated by dotted lines is observed. The corpus callosum is thin and dysmorphic (arrowhead). **c)** Axial post-contrast T1-weighted image shows 'molar tooth appearance' of elongated superior cerebellar peduncles (arrow). **d)** Axial Constructive Interference in Steady State (CISS) sequence shows hypoplastic middle cerebellar peduncles (arrows). **e)** Axial CISS sequence shows hypoplastic vestibulocochlear and facial nerves (arrows). **f)** Narrow and hypoplastic internal acoustic canal is observed in axial CT images (arrows)