We describe two patients (a Filipino boy aged 2.7 years and a Kuwaiti girl aged 4.8 years) with clinical and MRI findings consistent with the diagnosis of pontine tegmental cap dysplasia (PTCD) and compare them with 23 other cases reported in the literature. Both presented with feeding problems (VII nerve), sensori-neural deafness (VIII nerve) and hypotonia from birth and later developed corneal opacities due to loss of corneal sensation (V nerve). They have severe psychomotor developmental delay. The MRI of their brain showed a flattened ventral pons, vaulted “cap”- like structure protruding into 4th ventricle and a “molar tooth” sign. One of our patients also had Tetralogy of Fallot (TOF) successfully corrected. The other had no extracranial manifestations. The findings in our patients are similar to those reported except for the occurrence of TOF which has not been reported before in association with PTCD.

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not previously reported. We compare our cases with the 23 others reported in the literature.

1. **Case report 1**

A boy S. F. was born at 33 weeks gestation age by spontaneous vaginal delivery as the first child of healthy, unrelated Filipino parents. Pregnancy and delivery were uneventful; birth weight was 1.6 kg. The family history was unremarkable. Immediately after birth he was diagnosed to have Tetralogy of Fallot and in the following 20 months he underwent several surgical procedures and total surgical repair. Hemodynamics improved, and his cardiac function is good. During the surgical procedures he developed seizures, which were treated with antiepileptic drugs with good control. Electroencephalography (EEG) was normal. He has undescended testes, proven by ultrasound. No skeletal malformations were found on X-ray skeletal survey.

He also developed sucking but no swallowing difficulties from birth requiring spoon feeding and soon afterwards was found to be deaf. This was confirmed by auditory evoked response test to be sensorineural in type. There was no history of recurrent apneic attacks. At the age of 6 months he developed marked corneal clouding due to neurotrophic ulcers. When seen at the age of 20 month he had marked global developmental delay: head control was achieved at the age of 18 months, rolling over and sitting with support around 19 months. His speech was undeveloped and his social contact poor. His head circumference was 43.7 cm, <3rd centile, weight 10 kg and height 79 cm, both on 5th centile for age. His eye movements were normal and there was no nystagmus. They were half open during sleep, corneal sensation was decreased and blink reflex absent. He had reduced facial expression and spontaneous movements, generalized hypotonia, but normal deep tendon and plantar reflexes. There was no intention tremor or truncal ataxia. Olfactory and vestibular functions were not assessed.

MRI was done at the age of one month. Findings suggested brain steam involvement, typical for PTCD: flat profile of the ventral side of the pons, vaulted structure protruding into the enlarged 4th ventricle; shortening of the mesencephalic isthmus; hypoplastic cerebellar peduncles – very thin middle cerebellar peduncles and abnormal shape of superior cerebellar peduncles – molar tooth like sign. Medulla oblongata shows absence of the contours of the inferior olivary nuclei (Fig. 1). Supratentorially the MRI showed slight enlargement of peripheral CSF spaces and lateral ventricles.

![Fig. 1](image)

**Fig. 1** – MRI of patient 1 at age 11 months: Sagittal T1-weighted MRI showing flat ventral pons (black arrow), cap-like structure of dorsal pons protruding into enlarged forth ventricle (white arrow), shortening of mesencephalic isthmus (dotted line) (A). Axial T2-weight MRI: very thin cerebellar peduncles (arrows) (B). Axial T2-weight MRI: abnormal shape of superior cerebellar peduncles with molar-tooth-like appearance (arrows) (C). Axial T2-weight MRI showing medulla oblongata without noticeable inferior olivary nuclei (arrows) (D).
2. Case report 2

A Kuwaiti girl N.F. was a product of full term normal vaginal delivery, without perinatal complications and with birth weight 3.3 kg. Parents are consanguineous. Family history is unremarkable. Early after birth she presented with hypotonia, failure to thrive, poor sucking and swallowing. She was fed by nasogastric tube. There was no history of apneic attacks. Later on gastrostomy was performed due to persistent feeding problems. On follow up it was noticed that her vision was poor and bilateral corneal opacities were present due to neurotrophic ulcers caused by decreased corneal sensation. Visual evoked potentials indicated significant postretinal dysfunction affecting visual pathways. Hearing was very poor and the auditory brainstem response waveforms could not be detected in both ears.

At the age of 4 years and 9 month she developed epileptic seizures and sodium valproate was started, with good clinical response. EEG was abnormal with focal epileptiform discharges over left fronto-centro-temporal leads. At that age she was severely developmentally delayed: she could sit without support, but couldn’t stand and walk. She was deaf and had undeveloped speech. She was fed orally and by nasogastric tube. Eye movements were normal and there was no nystagmus. There was hypesthesia of the face and poor facial expression with the eyes half open during sleep. She was edentulous (teeth extracted because of self-mutilation). She was hypotonic with normal deep tendon and plantar reflexes. There was no intention tremor or ataxia. Olfactory and vestibular functions were not evaluated. She had no extracranial malformations.

MRI of the brain showed flattened ventral pons with “cap” structure projecting into 4th ventricle; hypoplastic middle cerebellar peduncles and molar tooth like sign due to abnormal superior cerebellar peduncles (Fig. 2).

3. Discussion

Pontine tegmental cap dysplasia is a relatively newly described entity. Common findings which define this syndrome are present on MRI of the brain: flat profile of the ventral pons, cap-like formation of dorsal pons protruding into 4th ventricle, absence of middle cerebellar peduncles, “molar tooth” appearance of superior cerebellar peduncles, with variable vermis hypoplasia and supratentorial findings. MRI of the brain of both our cases showed typical findings of PTCD.

Etiology is presumed to be genetic but the gene is still unknown. Multiplanar diffusion tensor imaging (DTI) investigation suggested a possible mechanism for the malformation: a defect in axonal growth and guidance resulting in neuronal loss and heterotopic commissures.

All reported cases showed a wide range of symptoms. Clinical findings are variable but affection of some cranial nerve is almost always present (Table 1). Impairment of VIII cranial nerve is reported in all patients with consequential sensorineural deafness. In five reported cases MRI confirmed absence or hypoplastic cochlear nerve. As a consequence all patients had speech impairment which vary from complete mute and sign language to some level of recognizable speech. Three previously reported patients had cochlear implants with improved speech after intervention while one showed minimal response. Neither of our patients developed speech.

Other findings include V, VII, IX and X cranial nerve involvement causing corneal anesthesia, sucking and swallowing difficulties requiring gastrostomy tube feeding in some cases. Both our patients had involvement of V, VII and VIII cranial nerves, and patient no 2 additionally had impairment of IX and X cranial nerves requiring gastrostomy. However, in most patients swallowing and sucking problems improved.
with the time, as was the case in our patient no 1 while the second patient had persistent feeding difficulties. These clinical findings imply involvement of most of the cranial nerves originating from pontine nuclei located in the tegmentum but also cranial nerves originating from nucleus ambiguous in the medulla oblongata.

Ocular movement abnormalities in the form of horizontal gaze palsy, oculomotor apraxia, vertical nystagmus, near-absent voluntary eye movements are described. Abnormal eye movements can be explained by pontine malformations or cerebellar lesion or both.1 Cerebellar signs – ataxia, incoordination, head titubation are also reported.1,3,7,8 Our patients did not have eye movement abnormalities or cerebellar signs other than possible hypotonia. The cerebellar signs may be due to involvement of the corticospinal tracts in the medulla oblongata.

The authors declare no potential conflicts of interest.

Table 1 – Clinical manifestations in 25 reported patients with PTCD.

<table>
<thead>
<tr>
<th>Clinical manifestations</th>
<th>Number of affected patients (n = 25)</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Deafness</td>
<td>25/25</td>
<td>(100%)</td>
</tr>
<tr>
<td>Corneal anesthesia</td>
<td>18/25</td>
<td>(72%)</td>
</tr>
<tr>
<td>Feeding difficulties</td>
<td>20/25</td>
<td>(80%)</td>
</tr>
<tr>
<td>Sucking difficulties</td>
<td>4/20</td>
<td>(20%)</td>
</tr>
<tr>
<td>Chewing difficulties</td>
<td>3/20</td>
<td>(15%)</td>
</tr>
<tr>
<td>Swallowing difficulties</td>
<td>17/20</td>
<td>(85%)</td>
</tr>
<tr>
<td>Abnormal eyes movements</td>
<td>13/25</td>
<td>(52%)</td>
</tr>
<tr>
<td>Cerebellar signs</td>
<td>16/25</td>
<td>(64%)</td>
</tr>
<tr>
<td>Pyramidal signs</td>
<td>19/25</td>
<td>(76%)</td>
</tr>
<tr>
<td>Seizures</td>
<td>13/25</td>
<td>(52%)</td>
</tr>
<tr>
<td>Skeletal anomalies</td>
<td>15/25</td>
<td>(60%)</td>
</tr>
<tr>
<td>Cardiovascular anomalies</td>
<td>10/25</td>
<td>(40%)</td>
</tr>
<tr>
<td>Other anomalies</td>
<td>12/25</td>
<td>(48%)</td>
</tr>
</tbody>
</table>

* Including two patients reported in this paper.

As this is a case report, no ethical review is deemed necessary.

Funding

The author received no financial support for authorship, and/or publication of this article.

Author contribution

Marina Jovanovic prepared manuscript and provided care for patient no 2.
Mohamed Zakkariah was involved in care of both patients.
Majed Elshahat Abdrabon Eilewa provided care for patient no 2.
Ivana Markovic interpreted MRI for both patients.
Sameera Abdulla Sadeq, was consultant in charge for patient no 1.
Allie Moosa, was consultant involved in care of both patients and reviewed the manuscript.

Declaration of conflicting interests

The authors declare no potential conflicts of interest.

References


