CASE REPORT

Inferior pontine segmentation abnormality in a child with sensorineural deafness: DTI analysis of fiber tracts

Anomalie de segmentation pontine inférieure chez un enfant avec surdité neurosensorielle : analyse DTI des faisceaux de substance blanche

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Introduction

Malformations of the brainstem and cerebellum have been described in various conditions including chromosomal disorders [1], pontocerebellar hypoplasias types 1-5 [2], familial horizontal gaze palsy and scoliosis [3], Mobius syndrome [4] and more recently, pontine tegmental cap dysplasia [5]. Recent advances in neuroimaging and genetics have resulted in a more accurate classification of posterior malformations into disorders involving a) abnormal brainstem segmentation, b) segmental hypoplasia, c) postsegmentation abnormalities and d) abnormal cortical organization [6]. In this report, we present the magnetic resonance imaging (MRI) and diffusion tensor imaging (DTI) findings in a 12-month-old child referred because of sensorineural hearing loss (SNHL). MRI revealed hypoplastic 8th cranial nerves in combination with an inferior pontine segmentation abnormality.

Case report

A 12-month-old Caucasian female presented to our department for MR imaging of the brain and brainstem because of profound bilateral SNHL. She was born at 41 weeks gestation via spontaneous delivery. Pregnancy was uncomplicated. Apgar scores were 5/7/9. At birth she had respiratory dis-
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Figure 1  T1- and T2-weighted (CISS) sagittal MRI scans of the brain shows truncation of the inferior pons (arrow). Corpus callosum and remainder of the midline structures appear unremarkable.

tress which required intubation. A gastric tube was placed because of significant swallow dysfunction.

Otoacoustic emission (OAE) testing at 4.7 months showed no response bilaterally indicating poor outer hair cell function. Follow up testing, at 6 months showed no responses with air conduction to 95 dB bilaterally or bone clicks at 60 dB. Behavioral observation audiometry at 7.5 and 10.4 months confirmed the previous findings. A hearing aid was placed and a questionable response was seen at 80 dB for a 500 Hz warble tone. Repeat testing showed aided speech awareness and pure tone responses at 70 dB. No other consistent aided responses were noted to narrowband noise or warble tones. Based on these results it was unclear whether the patient had any measurable hearing. Based on these results it was unclear whether the patient had any measurable hearing. In addition, she had no obvious response to gentle small amplitude rapid head rotations which suggested possible vestibular hypofunction. No formal vestibular testing was carried out.

Further neurological examination showed significant hypotonia. She was not able to sit up or hold up her head, thus postural instability could not be clearly assessed. She showed the ability to briefly sit on a ball and move around with some vision, she had intact facial nerve motion bilaterally, reduced gag reflex and 2+ reflexes. She also had oropharyngeal dysphagia with delayed onset of swallowing. Electroencephalogram (EEG) findings were normal.

MRI of the brain was performed to exclude retrocochlear pathology (e.g. absence/hypoplasia of the vestibulocochlear nerve) prior to possible cochlear implants. MRI was performed on a 3T MRI scanner using standard departmental pediatric brain MRI protocols including dedicated imaging of the inner ear and brainstem. MRI revealed an abnormal shortened pons, with apparent truncation of the inferior one-half (Fig. 1). The internal auditory canals and 8th cranial nerves were hypoplastic bilaterally (Fig. 2a,b). The remainder of the cranial nerves appeared intact on imaging. The cochlea, vestibule and semicircular canals appeared normal. Otherwise the brain appeared normal.

In addition, DTI sequences were acquired to study the internal neuroarchitecture of the brain and brainstem. Isotropic diffusion-weighted, apparent diffusion coefficient, and fractional anisotropy (FA) maps were calculated. The principal 3D orientation of the major eigenvector was color-coded per voxel according to the red-green-blue convention: red indicating a predominant left-right, green an antero-posterior, and blue a superior-inferior orientation of the anisotropic component of diffusion within each voxel. The color intensity scale was proportional to the measured FA-value. The color-coded FA maps were studied for presence or absence of major white matter tracts within the brainstem. Comparison was made with an age matched healthy control using the identical DTI sequence as well as with the color-coded FA atlas published by Susumu Mori [7]. The fiber tracts were classified as present, absent, or not clearly visualized on multiple levels of the brainstem. In addition, if

Figure 2  A,B: coronal (A) and axial (B) high resolution heavily T2-weighted CISS imaging of the cochlea and vestibulum. The internal auditory canals (arrows) and 8th cranial nerves are severely hypoplastic, right > left. The inferior pons is hypoplastic.
the tracts were identified, the volume of the tracts was estimated based on the cross sectional area as normal or diminished.

Analysis showed that in the inferior, abnormal pons, the corticospinal tracts (CST), corticopontine tracts (CPT), inferior cerebellar peduncles (ICP), middle cerebellar peduncles (MCP) and the pontine crossing tracts (PCT) appeared absent/could not be identified (Fig. 3). In the anatomically normal appearing, superior pons, the CST, CPT, PCT and MCP could be identified but appeared diminished (Fig. 3). No additional malformations or tract anomalies were noted in the supratentorial brain.

Discussion

Only recently studies have started to use DTI data sets to unravel the complex internal neuro-architecture of brainstem malformations in vivo [8—10]. Several studies have focused on the spectrum of molar tooth malformations in small series of patients [9]. In addition, multiple case reports have described the DTI findings in a variety of selected, rare brainstem malformations [5,8,10]. Detailed analysis of color-coded FA-maps, three-dimensional reconstruction of fiber tracts, and comparison with normal DTI maps may help to better understand, classify and group brainstem malformations. In addition, these data may help to better understand neurological symptoms, clinical presentation and could give valuable information about neurological outcome or guide treatment.

This case report would like to add an additional small piece to the complex puzzle of brainstem malformations by presenting and discussing a novel case of an inferior pontine segmentation abnormality accompanied by hypoplastic/absent vestibulocochlear nerves bilaterally.

The distinct brainstem abnormality found in this patient is characterized by the apparent truncation of the inferior portion of the pons. Our findings are best understood by a review of the normal pontine anatomy. The dorsal pons primarily contains cranial nerves V, VI and VII as well as their nuclei, the medial longitudinal fasciculus, the tectospinal tract, the medial lemniscus, the lateral lemniscus, the central tegmental tract, and parts of the superior cerebellar peduncle. The ventral part of the pons, which is the area of abnormality in our case, consists of the descending axons of the CST, CPT, pontine nuclei as well as transverse pontocerebellar axons, which are known as the PCT [6]. DTI analysis of our patient revealed a marked diminishment of the CPT, CST, PCT in the inferior portion of the pons. In addition, the ICP, MCP were diminished at the inferior level of the pons. This observation can be explained by the fact that during development, the PCT normally connect the pontine nuclei to the contralateral cerebellar cortex via the MCPs [8] and therefore abnormalities in the PCT would lead to an abnormality of the MCP.

In addition, conventional MR imaging also demonstrated absence of the 8th nerves and diminutive internal auditory canals bilaterally. We speculate that this may be attributed to an absence/malformation of the cochlear nuclei and/or course of the cochlear tracts within the brainstem. The cochlear nuclei are located lateral and dorsal to the restiform body in the floor of the lateral recess of the fourth ventricle, while the cochlear tract lies along the inferior border of the pons as cochlear nerves. In our case this inferior part of the pons appeared absent/hypoplastic.

Combining the findings of conventional MRI/DTI, the malformation observed in our case could have resulted from an early abnormal pontine segmentation as well as an impairment of cell proliferation of the pons and brainstem [6,8].

While our report may add another small piece of the puzzle to the ongoing research on brainstem malformations, it suffers from a number of limitations. It is a single case report, no neuropathological or detailed neurophysiological information is available, only those fiber tracts were studied that have been described previously. Our case report shows that several of these tracts were not identified in their usual location. It is however unclear if these tracts are absent, hypoplastic or possibly follow a different anatomical course. Future prospective studies using multi-tensor DTI should explore if these tracts are truly absent, follow a different pathway or if other neuronal networks have been recruited.

Conflicts of interest statement

The authors declare to have no conflicts of interest.

References

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