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Case report

Cochlear implantation in a child with subtelomeric 1q deletion syndrome and Dandy–Walker malformation

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ABSTRACT

Subtelomeric 1q deletion syndrome is a rare disorder characterized by severe mental and growth retardation, microcephaly, distinct facial features and corpus callosum abnormalities. Senserineural hearing loss is not common in this syndrome. We report a 2-year-old boy with subtelomeric 1q deletion syndrome presented with typical craniofacial abnormalities and bilateral senserineural hearing loss. Imaging revealed corpus callosum hypogenesis and Dandy-Walker malformation. Cochlear implantation was successfully undertaken using a transmastoid facial recess aproach. After 12-months post-implantation, good audiological outcomes were obtained. Cochlear implantation can be considered for hearing rehabilitation in patients with subtelomeric 1q deletion syndrome and Dandy-Walker malformation.

1. Introduction

Subtelomeric 1q deletion syndrome is a rare disorder, first described by Mankinen et al. [1]. Patients with this syndrome are characterized by mental and growth retardation with prenatal onset, microcephaly, seizures, short neck, hand and foot abnormalities, hypospadias and a variety of other midline defects, including corpus callosum, cardiac, genital, and gastro-esophageal abnormalities [2]. The facial features of patients include a full, round face with prominent forehead, upward slanting palpebral fissures, epicanthic folds, a short, broad nose with a flat nasal bridge, thin lips, downturn corners of mouth, micrognathia, low set ears, and an abnormal palate (sometimes cleft palate) [3].

In addition to these features, inner ear abnormalities and hearing loss have been described in only a few number of reports [2,4,5]. To our knowledge, this is the first case report of cochlear implantation in a child with subtelomeric 1q deletion syndrome.

2. Case report

A 2-year-old boy with subtelomeric 1q deletion syndrome was admitted due to prelingual hearing loss. He was the only child of healthy unrelated parents. At 37 weeks of gestation the patient was born with a weight of 2800 g (−2.5 SD). He had typical clinical features of subtelomeric 1q deletion syndrome including severe mental, microcephaly, micrognathia, short neck, a full, round face, thin limbs, downturn corners of mouth, epicanthic folds, and flat nasal bridge (Fig. 1). Severe developmental delay was noticed in the first year of his life. Audiological assessment by Auditory Brainstem Recordings (ABR) revealed no reproducible waveforms and profound hearing loss in both ears. There was no cochlear microphonics on both sides. He had bilateral type A tympanogram and absence of acustic reflexes. Distortion product otoacoustic emissions (DPOAE) and transient evoked otoacoustic emission (TEOAE) testings were negative. The temporal bone computed tomography (CT) revealed the pattency of cochlea, effusion in the middle ear and mastoid air cells. A magnetic resonance imaging (MRI) demonstrated corpus callosum hypogenesis, Dandy Walker malformation, cerebral atrophy, periventricular leukomalasia and bilateral cochlear nerve patency (Fig. 2). Cochlear implantation with Nucleus 6 CI (Cochlear, Melbourne, Australia) was applied to the right ear of

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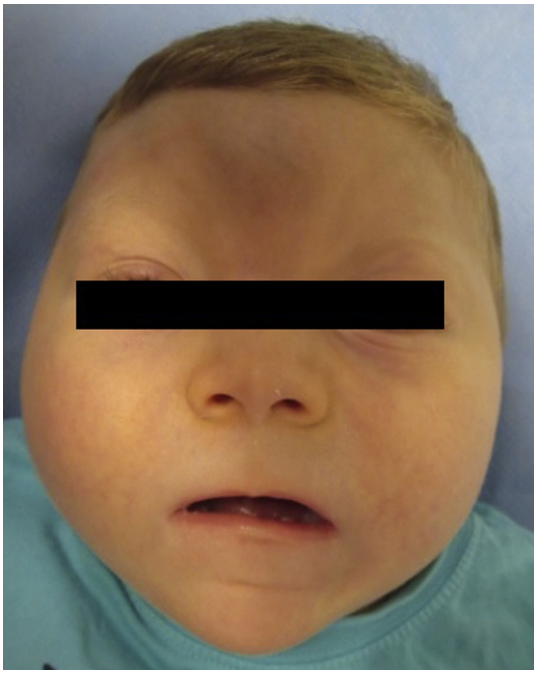

Fig. 1. Typical apparence of the face of a patient with subtelomeric 1q deletion syndrome.

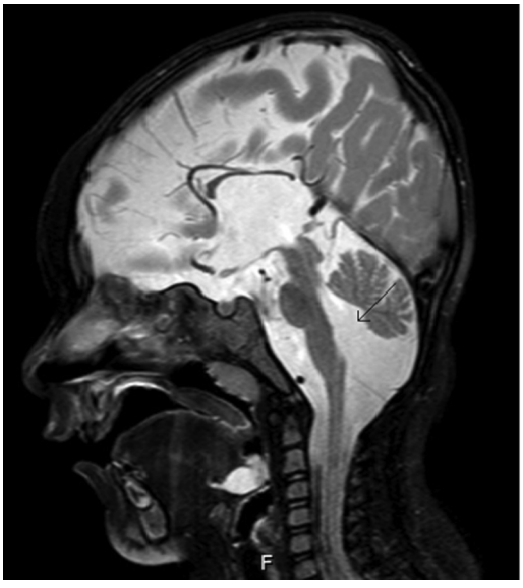

Fig. 2. Sagittal T2 weight cranial MR image of the patient. Note the cystic dilatation of the fourth ventricle (black arrow).

the patient under general anesthesia by using transmastoid facial recess approach. During the operation it was seen that there was an aberrant internal carotid artery was traversing over the promontorium without a bone covering on it. The stapes was absent and there was no oval or round window. The bone on the promontorium corresponding to the imaginary area of the round window was drilled posterior to the carotid artery, and a cochleostomy was performed. The scala timpani was exposed, and the implant electrode was fully inserted (Fig. 3). Postoperative period was uneventful. One year after implantation, the hearing threshold was 40 dB HL at the speech frequencies (Fig. 4). The child could detect

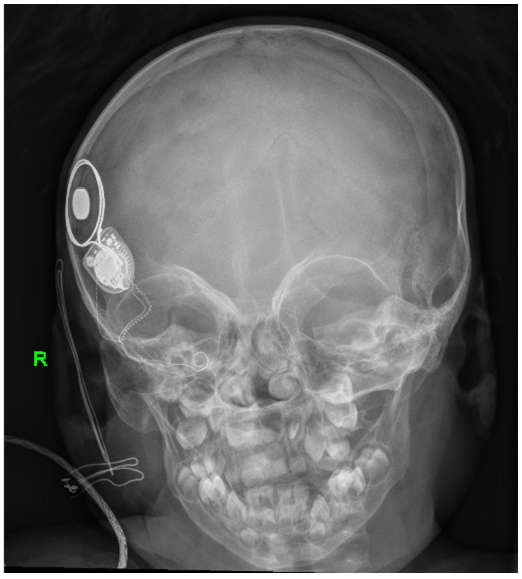

Fig. 3. Fully inserted cochlear implant is shown by transorbital X-ray.

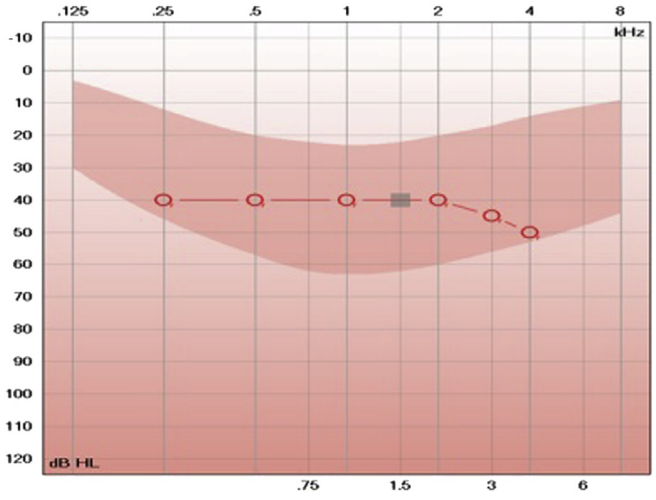

Fig. 4. Hearing threshold of the patient after one year follow up.

all ling sounds. He could also detect sound and speech without visual cues. The vocalization was increasing. He had syllable production (/ba–ba/,/ne–ne/).

3. Discussion

Several cases of subtelomeric 1q deletion syndrome have been reported [2–6]. Hearing loss is a rare feature in this rare syndrome [2,4,5]. However, cochlear implantation has not been reported to date in this syndrome.

For the first time, Rooms et al. described two patients with hearing impairment in subtelomeric 1q deletion syndrome [4] and then Roberts et al. reported a case with recurrent otitis media and mild hearing loss [5]. Subsequent to, Van Bever et al. presented a case who had an abnormal configuration of the auditory ossicles and hearing loss [2]. Our patient had several ear abnormalities like absence of stapes, and oval and round windows as well as an aberrant internal carotid artery traversing on the promontorium.

Structural abnormalities of brain are often present in

subtelomeric 1q deletion syndrome. Corpus callosum abnormalities are commonly described in many patients with this syndrome [6]. In accordance with this, corpus callosum hypogenesis was defined in MRI of our patient. Also our patient had Dandy–Walker malformation (DWM) which is unusual in this syndrome. DWM is the most common cerebellar malformation, described by hypoplasia and upward rotation of the cerebellar vermis, cystic dilation of the fourth ventricle, and enlarged posterior fossa with upward displacement of the lateral sinuses and tentorium [7,8]. DWM is a genetically heterogeneous entity that may be diagnosed as an isolated malformation or in association with genetic syndromes, single gene disorders, chromosomal abnormalities, or other multifactorial conditions [9]. To best of our knowledge, there are only two reports associating subtelomeric 1q deletion syndrome with DWM [2,10]. The first report was presented by Poot et al. and no sensorineural hearing loss was described in this case. Second, in a patient of Van Bever et al., abnormalities of ossicles in the middle ear and hearing loss existed.

Reportedly, there are only two patients with DWM who also had profound bilateral sensorineural hearing and received a cochlear implant [11,12]. First, Cushing et al. presented a child with Kerotosis, Ichtiosis and Deafness syndrome and bilateral profound sensorineural hearing loss was associated with a novel heterozygous missense D50A connexin 26 mutation. DWM and hypoplastic cochlea and a deficient roof of the superior semicircular canal on the left and a small modiolus on the right was revealed by radiological imaging [11]. Afterward, Oliveira et al. described a case with DWM who had profound bilateral sensorineural hearing loss and received a cochlear implant [12]. Our patient is also an additional case of cochlear implantation in DWM.

4. Conclusion

Profound bilateral sensorineural hearing loss can be seen in subtelomeric 1q deletion syndrome. Cochlear implantation can be performed successfully in this syndrome as well as DWM, and can yield a reasonable hearing and speech development. However, cochlear implant surgery can be challenging due to severe middle ear anomalies. Cochlear implantation should be considered in similar patients who has additional developmental delays in an attempt to increase the quality of life of both the patients and parents.

Conflict of interest statement

All authors declare that there are no conflicts of interest associated with this manuscript.

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