Dysmorphism of the middle ear: case report

Dismorfismo dell'orecchio medio: revisione della letteratura e descrizione di un caso

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Key words

Middle ear malformations • Cranio-facial dysplasia • Conductive hearing loss • Surgical treatment

Parole chiave

Malformazioni dell'orecchio medio • Displasia cranio-faciale • Sordità trasmissiva • Trattamento chirurgico

Summary

Although there are numerous publications in the literature describing the wide range of diagnosis, classifications and treatment of malformations of the hearing apparatus, even more variations can be found in clinical practice. Indeed, each individual case is unique as far as concerns pathogenesis, clinical course and treatment. The case reported herein describes a 12-year-old boy affected by cranio-facial dysmorphism and monolateral conductive hearing loss in the right ear: followed from radiological diagnosis – carried out to study a malformation of the ear pinna – to surgical treatment.

Riassunto

Vi sono, in letteratura, numerose pubblicazioni concernenti diagnosi, classificazione e terapia delle malformazioni a carico dell'apparato uditivo, ma ancora più numerose sono le possibili varianti riscontrabili nella pratica clinica. Ogni singolo caso è, per patogenesi, decorso clinico e possibilità terapeutiche, peculiare. Il presente lavoro è l'analisi del caso di un bambino di 12 anni affetto da dismorfismo cranico e ipoacusia trasmissiva destra: lo si seguirà dalla diagnosi radiologica – avvenuta in seguito ad accertamenti effettuati per una malformazione del padiglione auricolare – al trattamento chirurgico.

Case report

J.R. was first seen when he was 12 years old for hearing problems noticed by his mother.
He was born of a 30-year-old mother and a 35-year-old father: both parents were in good health and there was no family history of malformations.
The child was the product of a normal pregnancy, labour and delivery and there was no evidence of foetal infections during the pregnancy. Audiometrically, the patient was found to have a monolateral conductive hearing loss in the right ear. This impairment was more severe for frequencies between 125 and 2000 Hz. He pronounced his first words when he was 12 months old and started walking at 18 months. When he was a year old, he underwent surgery, to remove a cartilaginous formation in the region of the right ear tragus, at 5 he underwent adenoidectomy, and, at 10 appendicectomy. Speech and language milestones were essentially age-appropriate, and there was no evidence of cognitive or behavioural impairment. There was no family history of hearing loss. External inspection revealed a cranio-facial dysmorphism. Computed tomography (CT) scan of the petrous pyramid and mastoid revealed anomalies of the middle and internal ear: "asymmetry of the bony structures at the basis of the skull, particularly of the right petrous pyramid that is dysmorphic, anteriorly displaced and rotated. There is a hypoplasia of the epitympanum; the ossicles are in place with not well-identified stapes" (Figs. 1, 2).
Noteworthy was the internal acoustic canal: i.e., the longitudinal axis was normal in the left ear, while it was rotated by about 90° in the right ear. Mandibular condyles were of different volume and were not at the same level in the frontal section.
Having taken into consideration all these elements, it was decided to proceed with surgical treatment.

Surgical procedure

Given the young age of the patient, an anterior tympanotomy was performed under general anaesthesia. Following incision of the skin between the tragus and helix, the external ear canal was enlarged to allow a better view of the postero-inferior portion of the tympanic membrane that was partially hidden by the abnormal curvature of the canal floor.
The chorda tympani was preserved and the middle ear cavity normal.
The ossicular chain was present, but the stapes had a fixed footplate, a hypoplasic posterior crus and no anterior crus. The malleus handle was displaced anteriorly, while the anterior apophysis of the incus was also hypoplasic.

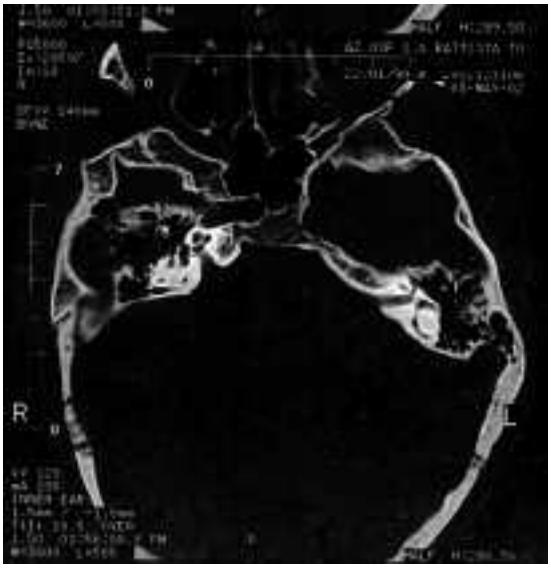

Fig. 1. Axial CT scan showing the asymmetry of the bony structures of the basis of the skull, particularly of the right petrous pyramid that is dysmorphic, anteriorly displaced and rotated. It has to be pointed out the direction of the internal auditory canal of the right side: nearly 90° in comparison to the left one.

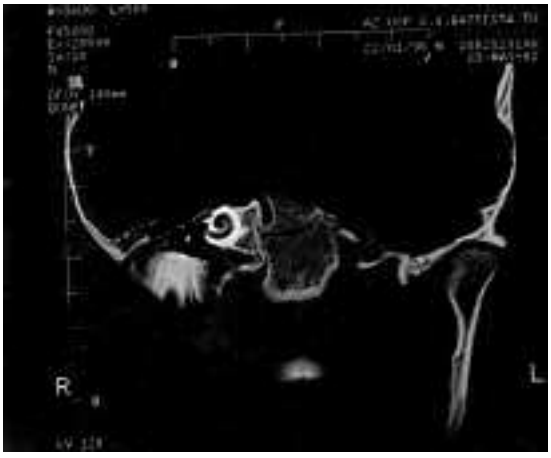

Fig. 2. Coronal CT scan showing hypoplasy of the right epithympanum and a not well identified right stapes. Impossible to have both the petrous bones on the same section.

It was difficult to explore the posterior portion of the oval window as the bony anulus and facial canal were overhanging.
Extensive drilling was performed in the anterior portion of the oval window niche, after which a hole was drilled in order to place a fluoroplastic piston-type prosthesis. The tympanomeatal flap was replaced and the external meatus sutured.
The post-operative course was uneventful and an acceptable threshold was observed at one month, which was subjectively better than the instrumental (Figs. 3, 4).

Discussion

The labyrinth has an embryologic origin which is quite separate from that of the middle and external ear. The inner ear or labyrinth develops from the ectoderm by the invagination of the otic placode. The external and middle ears, as well as the ossicles, develop from the first branchial cleft, the first pharyngeal pouch and the adjacent regions of the first and second branchial arch. Due to their separate origins, anomalies tend to be either of the inner or middle and external ear. Although anomalies of both regions are rare they do occasionally occur in certain syndromes and chromosomal anomalies. Combination anomalies also occur in cases of toxin exposure during early gestation.
The inner ear: the anomalies that affect the bony structure of the inner ear result from problems in the first trimester of pregnancy. The bony inner ear is essentially completely developed by this time. Initially, a small invagination of surface ectoderm forms a small vesicle, which becomes separate from the external surface. The vesicle, called the otocyst, is at first almost round, but soon small pouches form that elongate and further differentiate to form the cochlea, vestibule and semicircular canals. Congenital anomalies may mimic stages in this differentiation or may reflect absence or failure of this differentiation [1].
Middle and external ear: the first pharyngeal pouch extends from the primitive gut to form the eustachian tube and middle ear "en route" to the mastoid region. The branchial cleft extends inward from the external surface of the embryo and joins the pouch in the region of the tympanic membrane. The mesenchyme of the first and second branchial arches, which border the first cleft, form the ossicular chain, their muscles and ligaments. Anomalies often affect both areas due to the intimate relationship of formation of the middle and external ear [2-7].
Numerous studies on embryogenesis of the hearing system structure [8-10] have shown that it differs greatly from one individual to another: indeed, leading to the statement that "no two ears are the same" [11].
Despite this great physiological variability, reports of cases have appeared in the literature in which this difference becomes clinically relevant and, at times, truly pathological [12-16].
A malformation of the ear can be suspected in the presence of a deep conductive hearing loss without

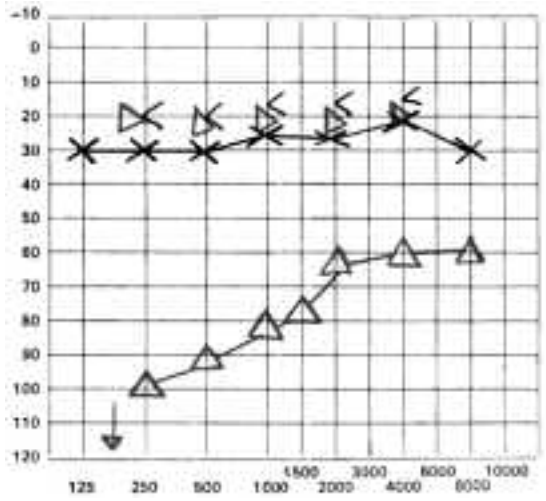

Fig. 3. Pre-operative audiogram showing a right conductive hearing loss for every frequency of the tonal field. The left ear presents an hearing function in the normal values range.

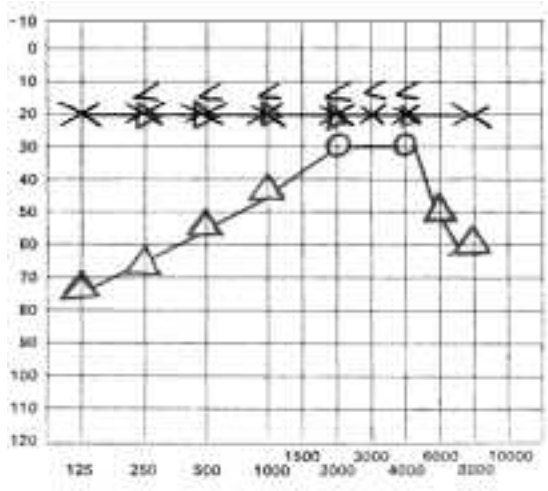

Fig. 4. Audiogram performed three months after the operation. The right ear presents an improvement of the hearing function for all the frequencies. Particularly, the diagram shows good results for the frequencies between 1 and 4 kHz that are the ones mostly interested in the oral communication.

any evident damage in the middle or external ear. When this is the case, as in some microdysplasias (i.e., of the ossicular chain) are not detected either by CT scan [17 11] or X-ray examination [18 19], then explorative tympanotomy is necessary [20-24].

There are numerous reports on congenital malformations of the hearing apparatus[25,26]: from cases of synostosis between the malleus and incus [27], to congenital absence of the incus [28-32], lack of the incudostapedial joint [33-35], or stapedial malformations [36-39]. These malformations are often related to malformations of the external ear. However, it is rare to find reports of malformations with an association of the external, middle or inner ear, such as that described here, in which cranial-facial-somatic dysmorphism with an evident asymmetry of the petrous bones is also present.

It is noteworthy that despite a malformation of the inner ear, there is no sensorineural hearing loss.

The peculiarity of this case resides in the particular distribution of the middle ear structures. Indeed, the surgical procedure routinely used for the fixed stapes in the case of microdysplasia of the ossicular chain was not feasible and, in fact, the surgeon had to try to find a "new" way to perform platinotomy.

Rotation of the whole petrous bone caused a radical alteration in the anatomic relationships between the ossicular chain and the medial wall of the tympanic cavity, resulting in the position of the oval window niche being moved posteriorly/superiorly which meant the surgeon had to enter into the anterior pole in the vestibule.

As the apophysis of the incus and malleus were also displaced anteriorly, it was possible, on account of its elasticity, to use the standard fluoroplastic prosthesis for stapedectomy.

The decision to perform surgery, even in the absence of indications, from a functional point of view (i.e., normal hearing in the other ear), was taken on the basis of the radiological findings (distorted middle ear, but potentially still correctable). Moreover, the family was in favour of all attempts to improve the child's hearing.

If a less favourable outcome is suspected, due to a more severe malformation (dangerous surgery), especially in a patient with monolateral deafness, it is preferable to delay surgery until the patient is of age and able to give his/her own consent or make decisions regarding the "costs/benefits" of the treatment [40-45].

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