Otological malformation in the context of cat's eye syndrome

Clinical Case

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Abstract

Background and objective: Congenital aural atresia is a congenital defect that occurs in 1 in 10,000 to 15,000 live births. It can occur in isolation or associated with a polymalformative syndrome, including cat eye syndrome .
Clinical case: This is a male born at term with anorectal malformation, bilateral microtia with agenesis of the right external auditive conduct and dysmorphic features are observed. The audiological evaluation performed demonstrates the existence of bilateral moderate-severe conductive hearing loss. The genetic study demonstrates the presence of a trisomy of chromosome 22, a genetic alteration responsible for cat eye syndrome .
Discussion: Cat eye syndrome is a rare disease which presents with ocular coloboma, anal atresia and ear defects. Definitive diagnosis is based on peripheral blood karyotype. Comprehensive patient approach by a multidisciplinary team is essential, however, prognosis is usually good.
Keywords: congenital aural atresia, karyotype, hearing loss

Introduction

Congenital aural atresia (CAA) is a birth defect that occurs in 1 in 10,000 to 15,000 live births. It can occur alone or in association with a polymalformative syndrome, including cat eye syndrome. We present a clinical case of a patient with cat eye syndrome in which the associated otological malformations that are documented radiologically stand out.

Case Description

We present the case of a term born boy with anal atresia, bilateral microtia grade I with agenesis of the right external auditory canal (EAC) (images 1 and 2) and dysmorphic facial features consisting of palpebral fissures with inferior deviation, hypertelorism and thin upper lip. There are no other alterations. Pregnancy and childbirth were unremarkable. There is parental consanguinity of the fourth

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Image 1
Right pinna dysplasia and EAC agenesis

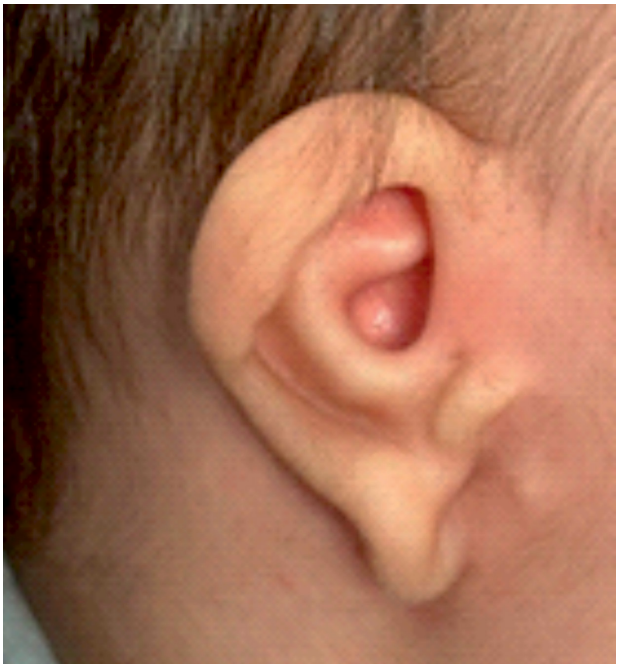
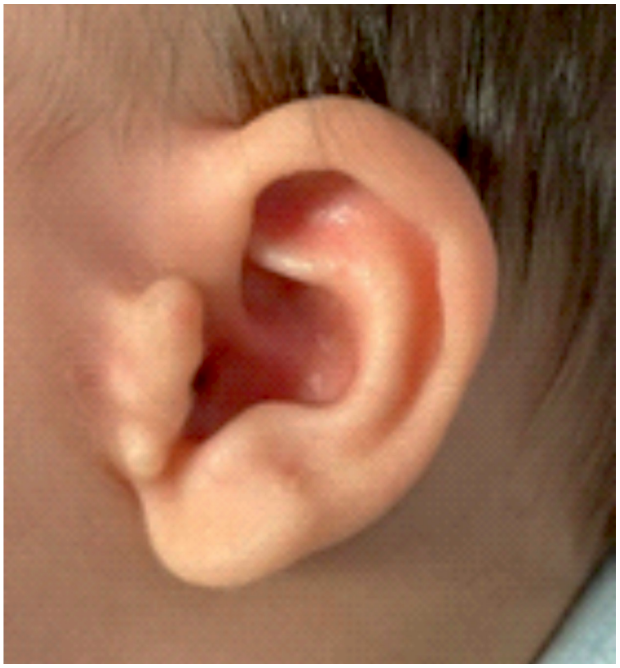


Image 2
Left preauricular appendage and EAC stenosis



degree. At 4 months of age the patient was referred to our center for diagnostic and therapeutic management.

The audiological study using auditory evoked potentials (air and bone conduction) showed moderate-severe bilateral transmission hearing loss with thresholds of 60-70 dB in (air) and 10 dB (bone) (image 3). Evoked brainstem potentials using a Sentiero Advanced transducer (SOH100360) with EC-03 (air) and B71 (bone) headset both with alternating "chirp" stimulus. No acoustic otoemissions were performed due to craniofacial malformations and alterations in both external auditory canals.

The genetic study of the karyotype in peripheral blood showed the presence of a bisatelite supernumerary microchromosome involving a partial mosaic triplication of the proximal region on the long arm of chromosome 22 (22q11). The karyotype presented is: 47, XY, +mar [16] / 46, XY [9] (image 4), genetic alteration responsible for cat eye syndrome (CES).

Computed tomography (CT) of temporal bones showed agenesis of the right external auditory canal (EAC) in its membranous and bony portions and severe stenosis of the membranous portion of the left EAC, with its bottom filled by soft tissue densities (image 5). In addition, bilateral ossicular chain dysplasia with bilateral malleus and incus fusion is described (image 6), and on the right ear a fusion of both to the lateral wall of the tympanic cavity. Both stapes are clearly observed (image 7). We observe also hypoplasia of mastoid cells and tympanic cavity bilaterally (image 6). A dilation of the subarcuate duct was also observed bilaterally (Figure 8). There is no other alterations. The cochleas, vestibule and semicircular canals are unremarkable. And both facial nerves are normal (image 9)

A bilateral adaptation of bone hearing aids coupled with an elastic band and speech therapy support was chosen, which obtained a good auditory response. In the future, when the age and conditions of the patient allow it, the adaptation of bone anchored hearing implants will be considered.

The comprehensive diagnostic and therapeutic management of the patient was discussed in a multidisciplinary committee with pediatricians, pediatric surgeons, otolaryngologists and maxillofacial surgeons.

Image 3
Brainstem evoked potentials. A: Air conduction with thresholds of 60 dB in the right ear and 70 dB in the left ear and increased latency in all waves in both ears. Transductor EC-03, alternating "chirp" stimulus. B: bone conduction with thresholds of 10dB. Transduketor B71, alternating "chirp" stimulus

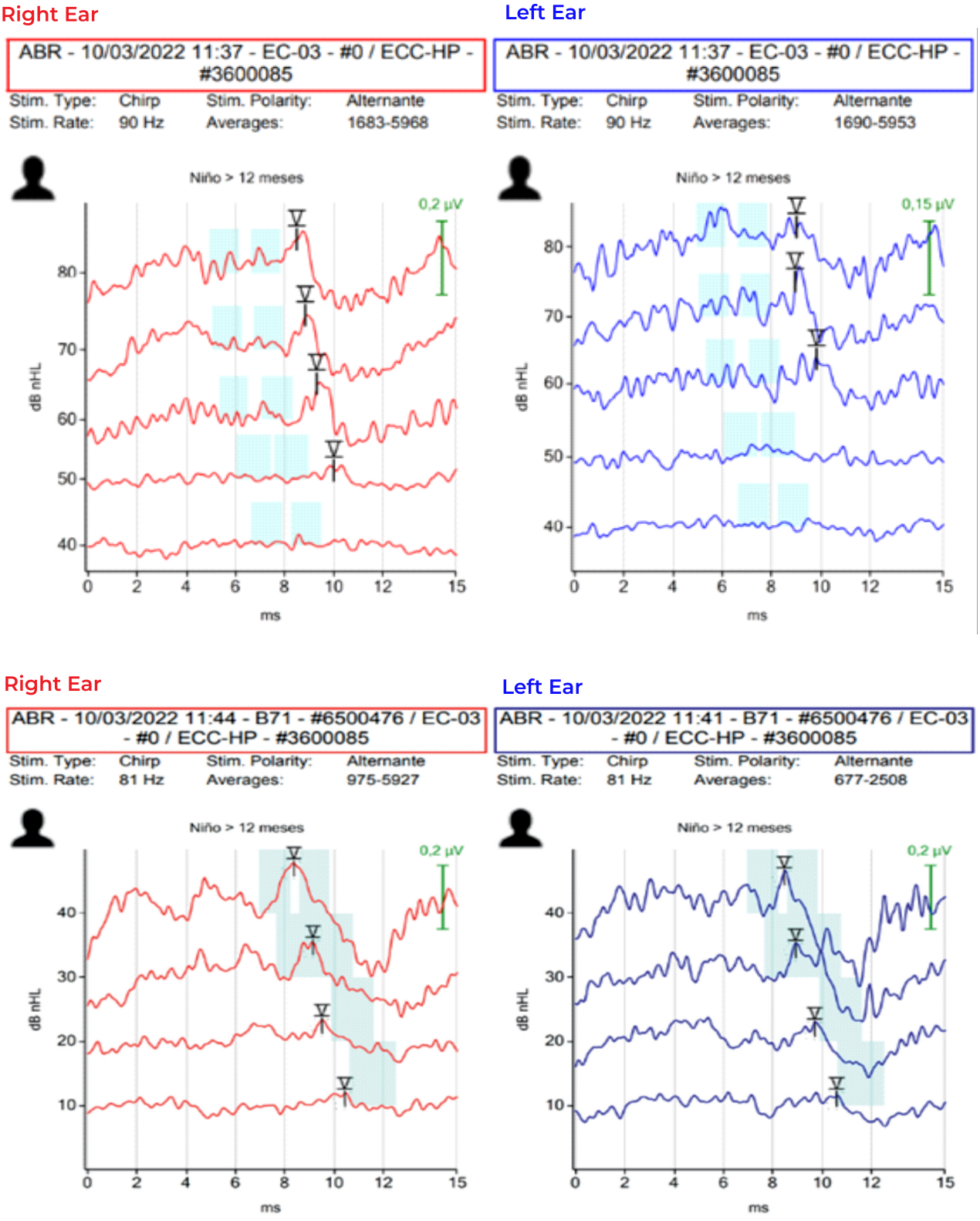


Image 4
Karyotype in peripheral blood 47, XY, +mar [16]/46, XY [9]. Bisatelised supernumerary microchromosome is indicated (arrow)

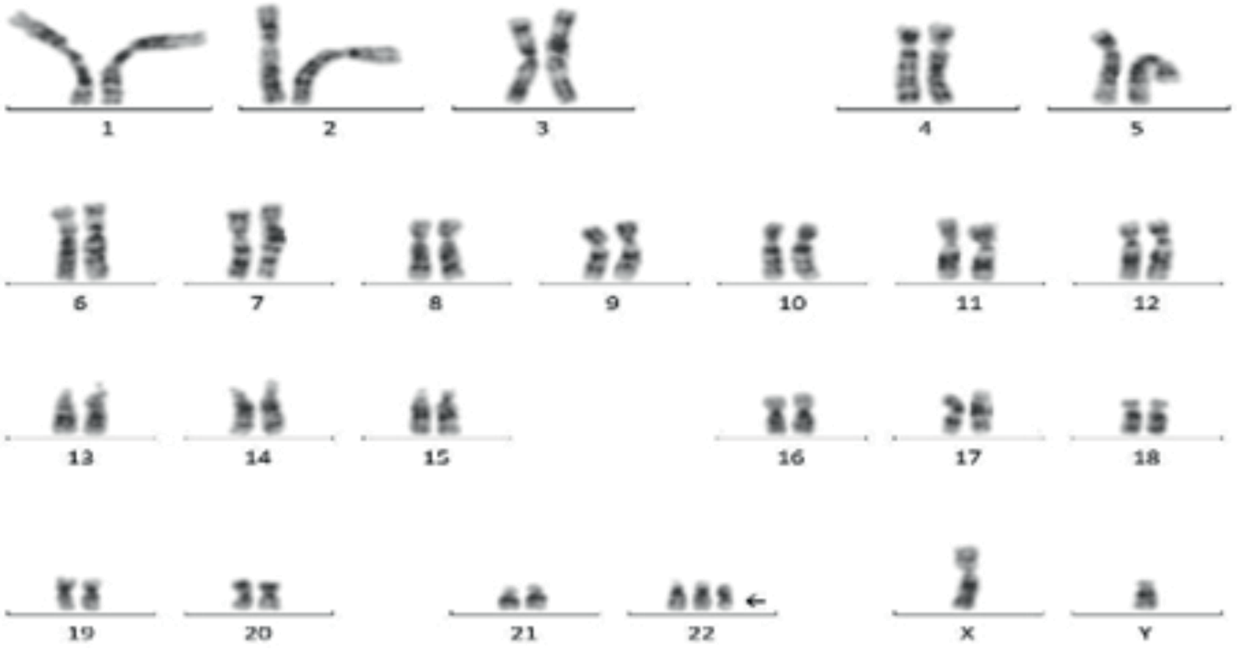


Image 5
Transverse CT image identifying right EAC atresia (black arrow) and stenosis of the membranous portion of the inner ear (white arrow), partial aeration of the left tympanic box and complete occupation of the right are observed. It shows fusion of malleus and incus to anterior wall of the tympanic cavity (arrowhead).

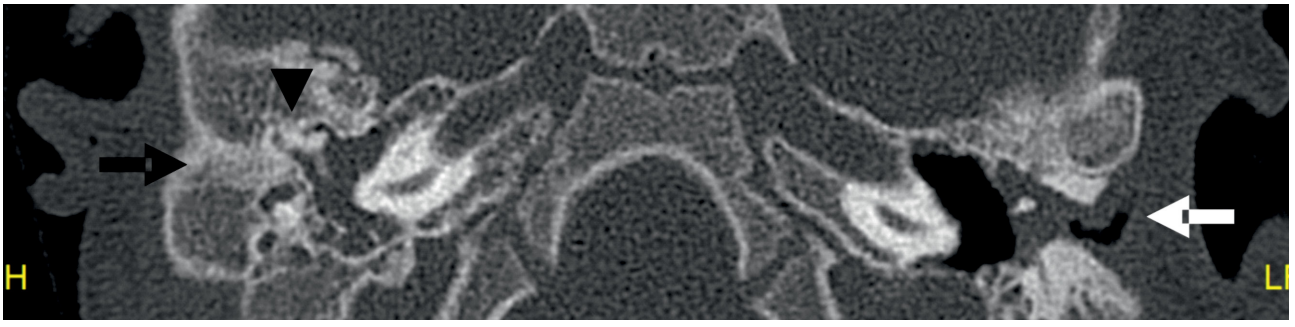


Image 6
Bilateral chain dysplasia with malleus and incus fusion (white arrows) is observed. Hypoplasia of mastoid cells (black arrows). Bilateral tympanic cavity hypoplasia with soft tissue inside (arrowheads).

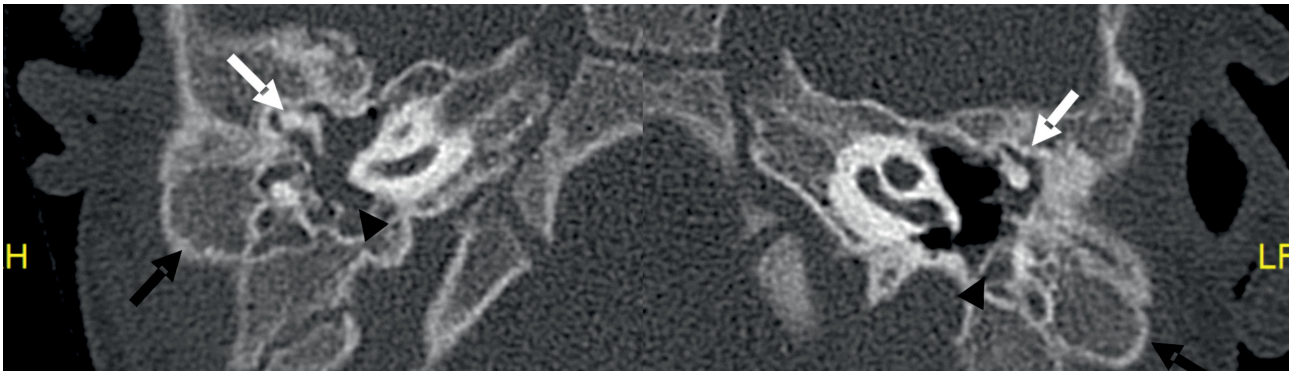


Image 7
Both stapes (white arrows) are observed

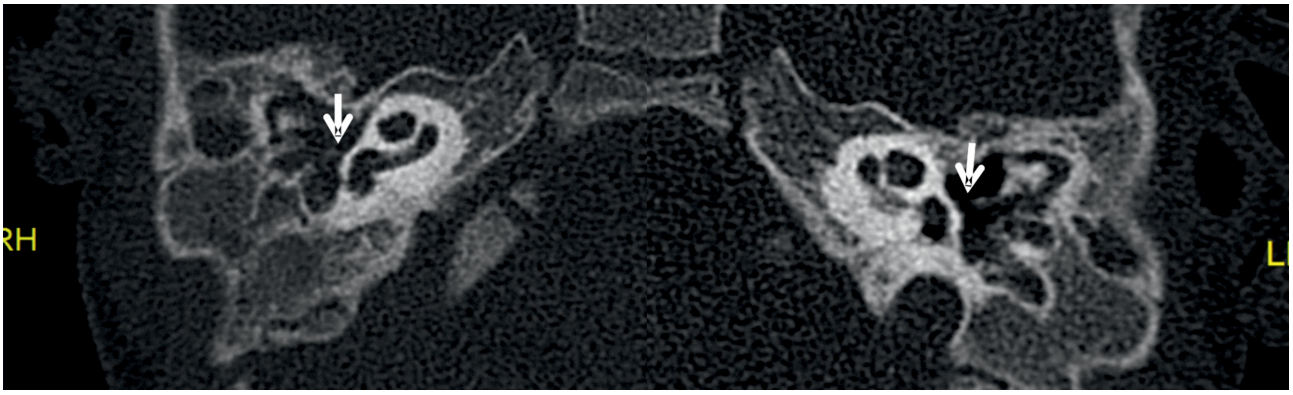


Image 8
Cross-sectional image of CT in bone window. Bilateral and symmetrical dilation of the lateral two-thirds of subarcuate or petromastoid canals (white arrows) and preservation of the proximal third (black arrows) is identified

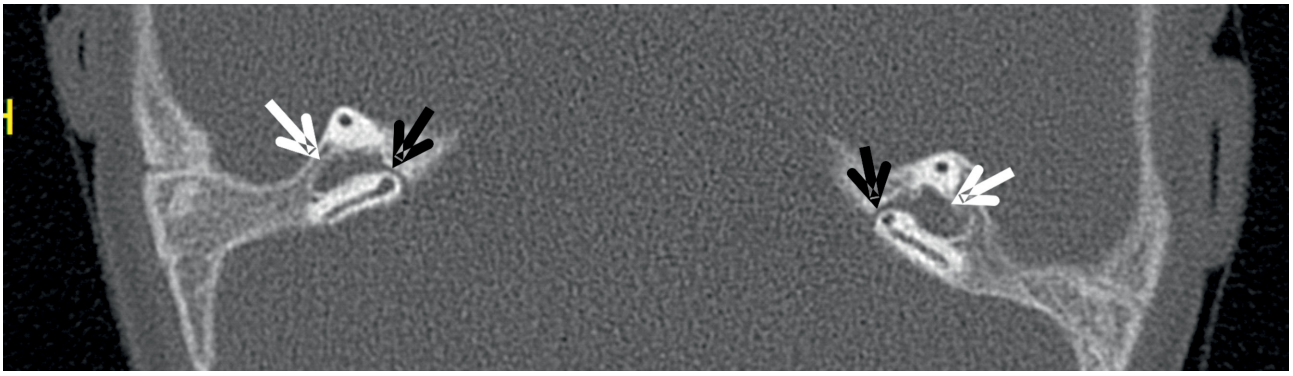
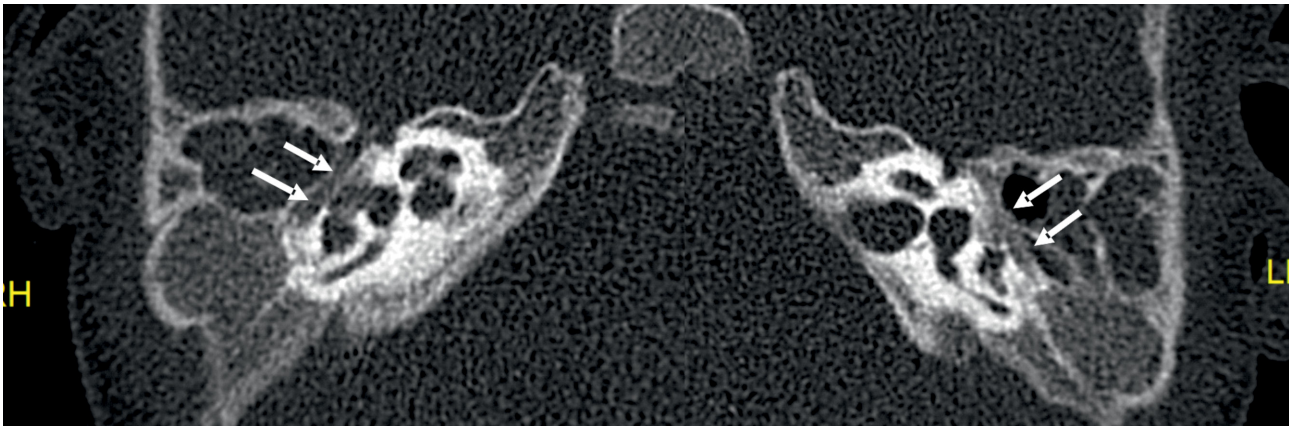


Image 9
Normal and symmetrical direction and thickness of the tympanic portion of both facial nerves (white arrows).



Discussion

CES is a rare genetic syndrome caused by trisomy or partial tetrasomy of chromosome 22. It has an incidence of 1: 150,000 live births[1]. It is clinically characterized by the combination of iris coloboma, anal atresia, down slanting palpebral fissures, preauricular appendages, cardiac and renal malformations, and normal or borderline cognitive function[2]. CES presents a high phenotypic variability even between affected individuals of the same family[2]. This variability leads to frequent underdiagnosis[3,4].

Among the aural malformations described are the low-set ear, preauricular appendage or fistula, hypoplastic lobe, prominent antihelix, and EAC atresia [1] . As we can see, most of the aural anomalies found in literature[1] are minor malformations, and in our present case we found, in addition to EAC atresia, alterations at the level of the middle ear shown by CT.

It is common in CES to find a bisatelised supernumerary chromosome derived from chromosome 22 that causes duplication or triplication of the proximal sequences of the long arm of this chromosome (22q11). Interstitial duplication of the 22q11 region has also been described. On some occasions, and so it was in our case, there is the presence of mosaicisms of the supernumerary chromosome.

In the literature there is no consensus on whether there is a correlation between phenotypic severity and the degree of mosaicism. Some authors postulate that there is no such correlation [3], however, a recent study favors its existence[5].

Cases tend to be sporadic although family pattern has been described [6]. Performing a karyotype is the reference diagnostic test. Fluorescence in situ hybridization (FISH) techniques or a hybridization genomic array can also be performed [7].

The comprehensive treatment discussed in multidisciplinary committees is essential to solve the different malformations present in these patients[8]. So it was in the case at hand, it was decided to treat the anorectal malformation first because it was the most urgent, and then to study and treat the rest of the malformations.

The prognosis tends to be favorable but depends fundamentally on the presence or absence of associated renal and cardiac malformations [4]. There are prenatal cases of great severity whose prognosis depends on cerebellar hypoplasia[9].

CES is rare but should be suspected in the presence of associated anorectal, aural and ocular malformations. The genetic study by karyotyping is essential for diagnostic confirmation while the audiological study is essential to correctly assess the auditory involvement and to be able to carry out precise and personalized therapeutic strategies.

Conflicts of Interest

The authors declare that there is no conflict of interests regarding the publication of this paper.

Data Confidentiality

The authors declare having followed the protocols in use at their working center regarding patients' data publication.

Protection of humans and animals

The authors declare that the procedures were followed according to the regulations established by the Clinical Research and Ethics Committee and to the 2013 Helsinki Declaration of the World Medical Association.

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Availability of scientific data

There are no datasets available, publicly related to this work.

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