Ausência congénita de estribo e janela oval com nervo facial mal posicionado e tuba auditiva anómala.

Congenital absence of the stapes and oval window with malpositioned facial nerve and anomalous eustachian tube.

Mercedes Álvarez-Buylla Blanco • Miguel Álvarez-Buylla Camino

ABSTRACT

Introduction: Since in 1995 the first case of absence of stapes was described, several cases were reported. However, its etiology remains unknown. Some authors suggest a genetic cause, without excluding the possibility of embryopathy due to infections or chemical agents. The existence of an associated palatal cleft, in this case, reinforces the hypothesis of a multifactorial origin.

Objective: Describe a rare condition in order to know how to suspect it. This condition is frequently associated with facial nerve malposition, that difficult the surgery and so, the most frequent treatment is providing hearing aids or a bone anchored hearing aid.

Case report: We present a case of a 10 years old girl who complains of left hearing loss since childhood, accompanied by tinnitus and frequent left ear otorrhea episodes. We studied symptoms, complementary exams, treatment and course. Discussion and conclusions: A middle ear malformation should be suspected with the presence of a history of conductive hearing loss since birth or more frequently between the 7 and 12years old, fixed-type, which often affects conversational or low frequencies, which are more intense than other acquired hearing loss, with no history of ear infections or without improvement despite different kinds of treatments, and with a family history of hearing loss.

Conclusion: Congenital absence of stapes and oval window associated with anomalous course of the facial nerve is a rare entity. It presents as a conductive hearing loss non-progressive with 60 dB tone air threshold, often presented during childhood. Diagnosis is based on clinical suspicion by a correct medical history and audiological examination, confirmed by Computed Tomography (CT) scan. The anomalous course of the facial nerve supports the diagnosis and guides treatment. Initial treatment with hearing aids provides good hearing gain with adequate adaptation.

Keywords: Congenital, stapes, oval window, facial nerve, hearing loss.

Mercedes Álvarez-Buylla Blanco
Hospital Vital Álvarez-Buylla - Mieres - Spain

Miguel Álvarez-Buylla Camino
Hospital Vital Álvarez-Buylla - Mieres - Spain

Correspondência:
Mercedes Álvarez-Buylla Blanco
Plaza de la Constitución, n6, 1A. CP33600. Mieres. Asturias. Spain
+34669656386
e-mail: mercedesabb@msn.com

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INTRODUCTION

Minor aplasias are congenital malformations that affect almost exclusively the middle ear and ossicle chain[1]. The isolated malformations of the ossicular chain are rare, presenting in a more frequent way in the stapes[2]. Since in 1995 the first case of absence of stapes was described, several cases were informated[3]. However its etiology remains unknown[4]. Some authors suggest a genetic cause[5], without excluding the possibility of embriopaty due to infections or chemical agents[1]. The existence of an associated palatal cleft in this case reinforces the hypothesis of a multifactorial origin[6].

Stapes, oval window, and facial nerve embrionary development are temporal and spatially related. The stapes and facial nerve derive from the second branchial arch, the footplate and oval window from the otic capsule and the Eustachian tube from the first pharyngeal pouch[1]. Despite the different embryologic origin of these structures, their anomalies appear associated because of their chronologic embriological development relationship occurring the defect between 4 and 10 week[2,6]. Stapes and oval window congenital anomalies occurs during the 5-7 week of embriological development due to anterior displacement of the facial nerve that stands between the otic capsule and the stapes, avoiding the necessary contact between both structures, for the adecuate formation of the oval window and correct development of the stapes superestructure[2].

In 51 cases series of congenital absence of the oval window published in 1976 by Jarsdoefer, 96% of the patients had a stapes malformation (43% absent stapes and 41% malformed stapes) and 76% had a facial malformation. Lambert published 7 cases with congenital absence of oval window all with absent stapes or severely malformed, and 45% with facial anomalies association. The most frequently were malposition or dehiscence of the horizontal segment of the facial nerve[2].

This association of the anomalies of the stapes and oval window with the anomalous development of the facial nerve present a great importance for an adequate surgical planning. 76% in the Jarsdoefer series and 50% in the Lambert series presented associated facial nerve malformation[2]. The most

frequent anomalies are inferior facial nerve, at the expected location of the oval window or below it; dehiscent facial nerve and wider or larger facial nerve; or less frequently there may be absenced of the second portion of the facial nerve with a timpanic cord thicker that is presumed to present fibers of the facial nerve[2].

OBJECTIVE

Describe a rare condition in order to know how to suspect it. This condition is frequently associated with facial nerve malposition, that difficult the surgery and so, the most frequent treatment is providing hearing aids or a bone anchored hearing aid.

CASE REPORT

We present a case of a 10 years old girl who complains of left hearing loss since childhood, accompanied by tinnitus and frequent left ear otorrhea episodes.

Presented cleft palate at birth which was surgically treated when she was one year old, and also adenotonsillectomy was performed at 4 years old. She presents family history of conductive hearing-loss.

Physical examination revealed a normal bilateral external ear and tympanic membrane. The impedance exhibits a normal compliance and bilateral absence of the acoustic reflex. Audiometry study shows bilateral conductive hearing loss, especially in the left ear with air tone auditory threshold at 70 dB and in the right ear at 40 dB only in low frequencies. Speech audiometry study shows the maximum threshold discrimination at 80 dB in the left ear and at 30 dB in the right ear.

A Computed Tomography (CT) scan revealed a congenital absence of stapes and bone obliteration of the left oval window. (Figure 1), malposition of the second portion of the facial nerve (Figure 2) and the right fallopian tube bone communicates with the apex of the temporal bone and its membranous part by an aberrant communication with the right sphenoid sinus. MRI of the internal auditory canals didn´t show any abnormalities. Under the diagnosis of congenital absence of stapes and oval window with malposition of the facial nerve in a child with unilateral hearing loss without language delay, we recommended hearing aid adaptation in the left ear getting very good tolerance and benefit with the device.

DISCUSSION

A middle ear malformation should be suspected with the presence of a history of transmission hearing loss from birth or more frequently between the 7-12-year-old, fixed-type, which often affects conversational or low frequencies, which are more intense than other hearing loss adcquired, with no history of ear infections or without improvement despite different kinds of treatments, and with a family history of hearing loss[4].

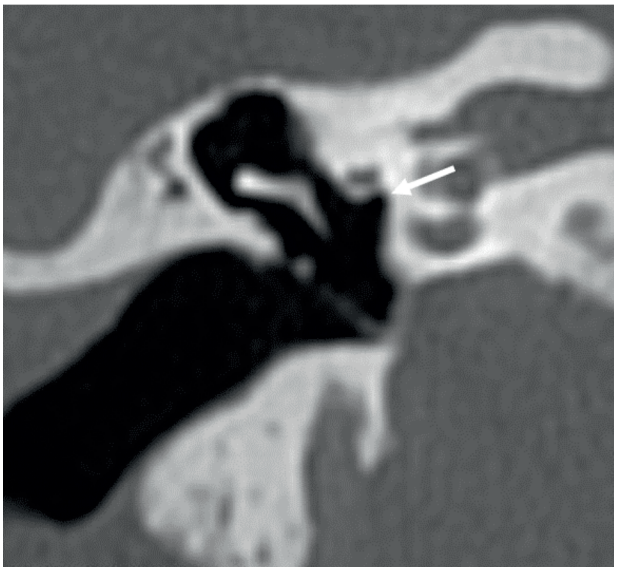
The delay in diagnosis is due to the fact that often these patients present unilateral hearing loss, and although it may be severe, the patient, family and professors recognize it later. Also the delay in the diagnosis is due to the absence of anomalies associated in pavilions or auditory canal as they occur in the major aplasias, and the difficulty of perform a conventional tonal audiometry before the kid has 4-5 years old. The etiology remains unknown with hereditary transmission defended for some authors and acquired etiology for another's[1,4,5]. The association with cleft palate in this case reinforces the theory of multifactorial origin.

The audiological exam shows a conductive hearing loss fixed at 60-70 dB, detected at birth, or more frequently during scholar age, affecting conversational or low frequency, and/or with family history of hearing loss[1, 4].

The impedanciometry rule out the existence of fluid in the middle ear or a stapedial fixation, finding normal compliance and no stapedial reflex[1].

The reconstruction of the axial plane of the TC with double - oblique orientation allows optimal visualization of the stapes and the oval window[2]. The absence of oval window is reflected as a bone obliteration or as a concentric narrowing which causes a depression along the medial wall of the tympanic cavity. (Figure 1).

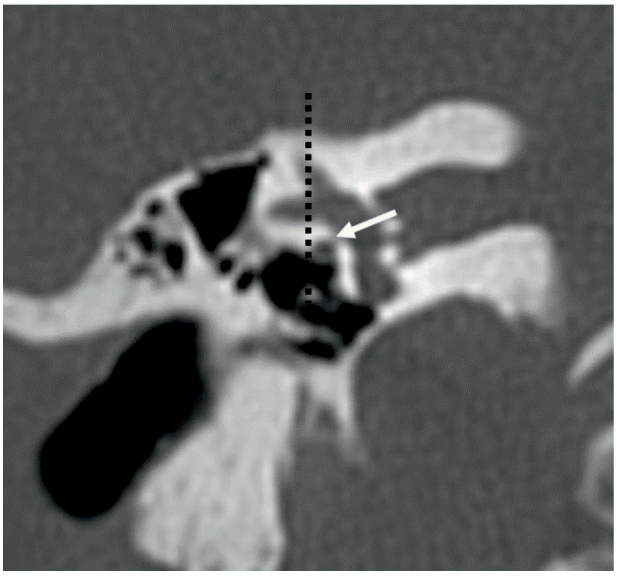
FIGURA 1
Coronal CT shows oval window obliteration (arrow) and stapes absent.



Using TC coronal sections, we can see the anomalous course of the facial nerve placed lateral to a vertical line drawn from the anterior junction of the superior semicircular canal and lateral semicircular canal[2]. (Figure 2).

The differential diagnosis includes traumatic or suppurative ossicular chain disruption. A proper anamnesis, audiological exploration and TC images helps the diagnosis. In the present case, despite the frequent history of otitis, the compliance was normal as well as the grade of ventilation of the middle ear and the state of the middle ear mucosa in the TC, discarding these diagnoses, with impaired compliance

FIGURA 2
Anomaly of the horizontal segment of the facial nerve, placed medial to a vertical line drawn from the anterior junction of the superior semicircular canal and lateral semicircular canal.



and middle ear mucosa disease. The differential diagnosis also include fenestral otosclerosis, very rare in children; and tympanosclerosis with usually presents inflammation changes in the middle ear mucosa and / or ossicular fixation[2].

Malposition facial nerve with suspicious stapes and/or oval window absence TC images are very suggestive in childhood, with no medical history of ear pathology, of congenital stapes and / or oval window absence[4].

Nevertheless, the diagnosis in most cases is casual during exploratory tympanotomy[7]. Surgical treatment is performed whenever the conditions are favorable. Nowadays the most used technique is platinotomy using interposition graft and piston placement, after creating a new oval window[1,8]. Hearing aids adaptation is reserved for cases in children with no progressive hearing loss, without language delay, where the surgery involves a surgical risk, and who may benefit from an intervention in adulthood[9].

Thus, treatment should be based on patient age, anatomic conditions that radiological images provides, and the characteristics of hearing impairment.

Surgical results found in the literature are highly variable due to the use of different techniques as semicircular canal fenestration or vestibulotomy, nowadays less used[9,10]. At present, the functional outcome by placing prostheses are satisfactory, with 80 % of cases improvement, while the average gain setting is estimated at 20-24 dB[1,8]. Malposition facial nerve difficult ossicular reconstruction conditioning sometimes reconstructive results[1,2,4,8]. In these cases, the adaptation of hearing aids or bone anchored hearing aid is generally an appropriate option with good functional results[4,8,10].

CONCLUSION

Congenital absence of stapes and oval window associated with anomalous course of the facial nerve is a rare entity. It presents as a conductive hearing loss non-progressive with 60 dB tone air threshold, often presented during childhood. Diagnosis is based on clinical suspicion by a correct medical history and audiological examination, confirmed by CT scan. The anomalous course of the facial nerve supports the diagnosis and guides treatment. Initial treatment with hearing aids or bone anchored hearing aid provides good hearing gain with adequate adaptation.

Protecção de pessoas e animais

Os autores declaram que os procedimentos seguidos estavam de acordo com os regulamentos estabelecidos pelos responsáveis da Comissão de Investigação Clínica e Ética e de acordo com a Declaração de Helsínquia da Associação Médica Mundial.

Confidencialidade dos dados

Os autores declaram ter seguido os protocolos do seu centro de trabalho acerca da publicação dos dados de doentes.

Conflito de interesses

Os autores declaram não ter nenhum confito de interesses relativamente ao presente artigo.

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