A Case of Familial External Auditory Canal Atresia

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Abstract

Introduction: Congenital aural atresia (CAA) can occur as an isolated congenital malformation or as part of a monogenic and chromosomal syndromes. CAA can be associated with several syndromes. An 18q deletion is frequently seen in CAA. The subject of the study are three individuals from one family (mother, son and daughter). Methods:After taking a detailed history, an examination of ENT organs, tone audiometry testing, auditory brain stem responses, brain CT, external auditory canal and middle ear were performed. Results: The conducted tone audiometry testing and auditory brain stem response found bilateral sound conduction hearing loss. Conclusion: Bilateral atresia is treated surgically. Surgical treatment is difficult and not always successful, it is preferable to perform it at age 4-5.

Keywords: Congenital aural atresia, bilateral sound-conductive hearing loss,

Introduction

Congenital aural atresia (CAA) is a rate malformation of the ear, occurring in 1 of 10,000 newborns (3). Its characteristics may vary from a narrow external auditory canal and hypoplasia of the tympanic membrane, and a cleft in the middle ear to complete absence of middle ear structures and anotia (2). Often, Congenital aural atresia (CAA) can occur as an isolated congenital malformation or as part of a monogenic and chromosomal syndromes. CAA can be associated with several syndromes, including Treacher Collins, Goldenhar, Klippel-Feil, Branchio-Oto-Renal (BOR) and Hemifacial Microsomia Crouzon syndrome, Pierre Robin syndrome, etc. (4, 5).

An 18q deletion is frequently seen in CAA. In 1964, De Grouchy described individuals with an 18q deletion who, in addition to atresia of the external auditory canals, had a wide range of associated features, including short stature, characteristic facial features, intellectual disability, reduced white matter myelination, and foot deformities. Veltman et al. studied children with isolated auditory canal atresia. In all patients, a ~5 Mb deletion was found on the long arm of chromosome 18 (18q22.3-q23), the gene for congenital atresia of the auditory canal.

Various classifications of Congenital aural atresia – CAA have been introduced. In 1955, Altman first described a classification of CAA, which has been modified over the years by other scientists. CAA type I is classified as bony or fibrous atresia of the lateral part of the external auditory canal and a nearly normal medial part and middle ear. CAA type II is the most common and is characterized by partial or complete aplasia of the external auditory canal. In type IIA, the external auditory canal is affected by complete bony atresia of its medial part or partial aplasia, ending in a blind fistula that leads to a rudimentary tympanic membrane. CAA type IIB is characterized by bony stenosis of the entire length of the external auditory canal. CAA type III is characterized by bony atresia of the external auditory canal and a very small or absent middle ear cavity (2).

The subject of our study are three individuals from one family (mother, son and daughter) with isolated congenital malformation – atresia of the external auditory canal.

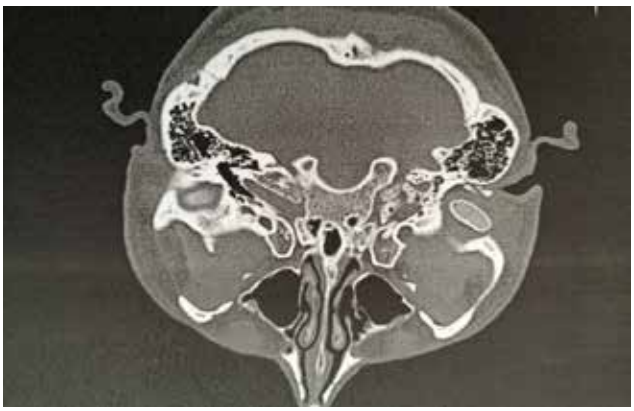
After taking a detailed history, an examination of ENT organs, tone audiometry testing, auditory brain stem response, brain CT, external auditory canal and middle ear were performed.

Case 1

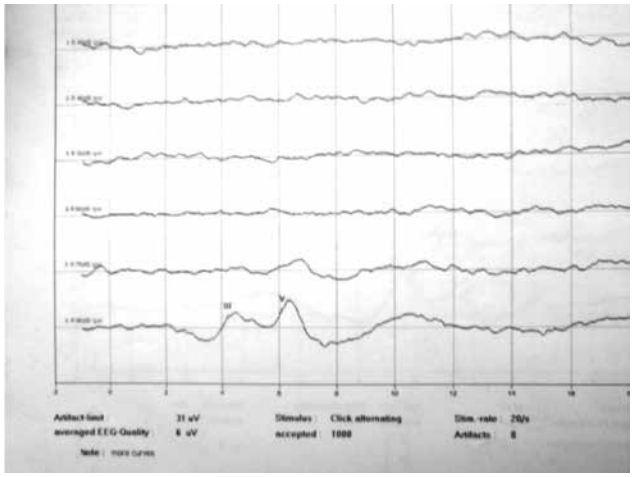
Subject 1 is a 55-year-old female born at term. One year after birth, atresia of the external auditory canals was found bilaterally. At the age of 6, following tonal threshold audiometry, bilateral acoustic hearing loss was found. She was successfully fitted with a hearing aid at a later age.

Examination of the ENT organs showed normally developed pinnae. Otoscopy revealed bilateral atresia of the external auditory canals type II as per the SCA classification. No abnormalities were observed in the other ENT organs. The tonal audiometry testing revealed bilateral sound-conductive hearing loss with right ear systolic hearing loss with Moderate Hearing Loss of 58 dB in the right ear and Moderate Hearing Loss of 55 dB in the left ear.

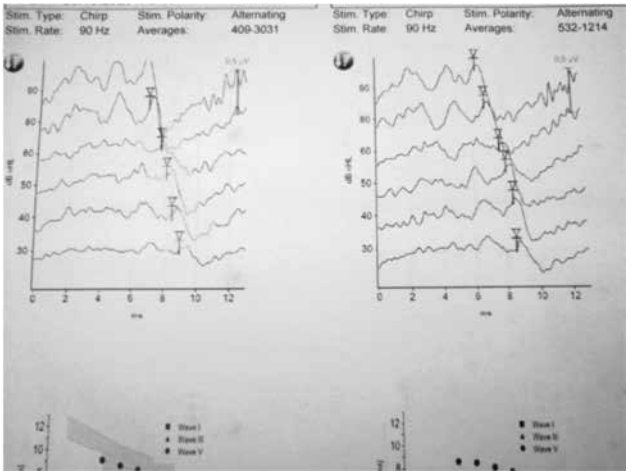
The performed computed tomography showed evidence of atresia of the two external auditory canals. No abnormality in the middle and inner ear structures.



Air-conducted brainstem auditory evoked potentials revealed typical threshold waves of 80, 90 dB. Bone-conducted brainstem auditory evoked potentials revealed typical threshold waves of 30 dB.



Air-conducted ABR

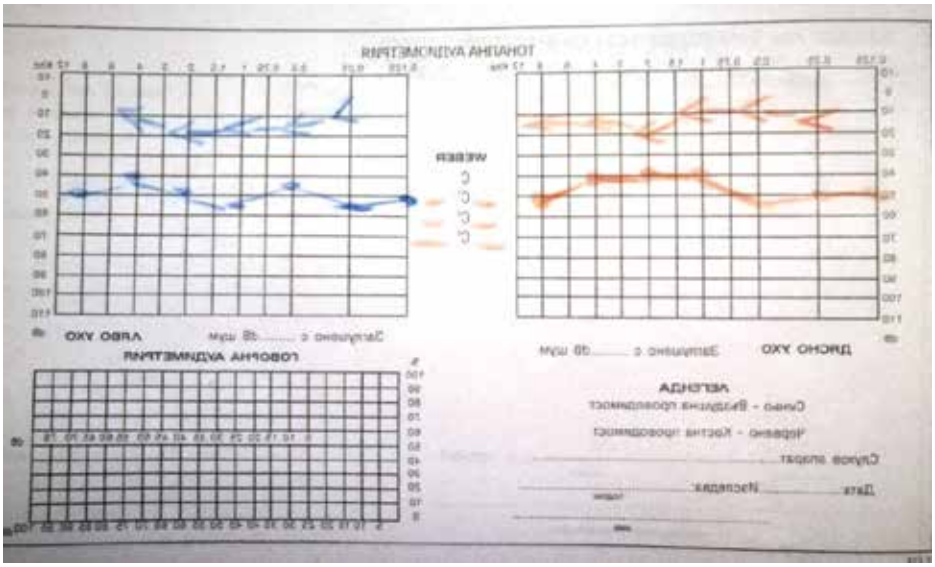


Bone-conducted ABR

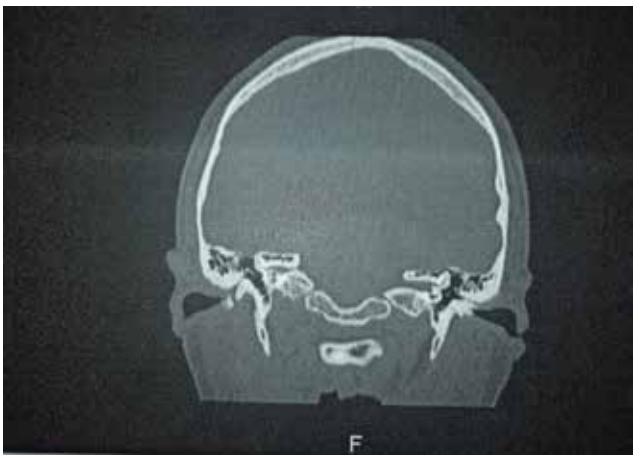
Case 2

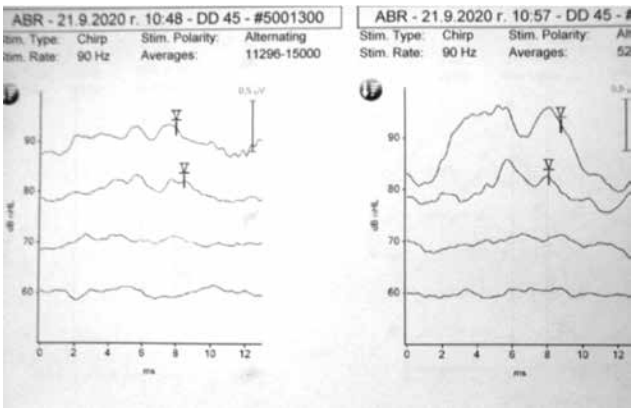
Subject 2 is the daughter of Subject 1 and her healthy husband. Born following a normal pregnancy at term, with normal birth parameters. During the examination of ENT organs, bilateral atresia of the external auditory canals was found, no other abnormalities were found.

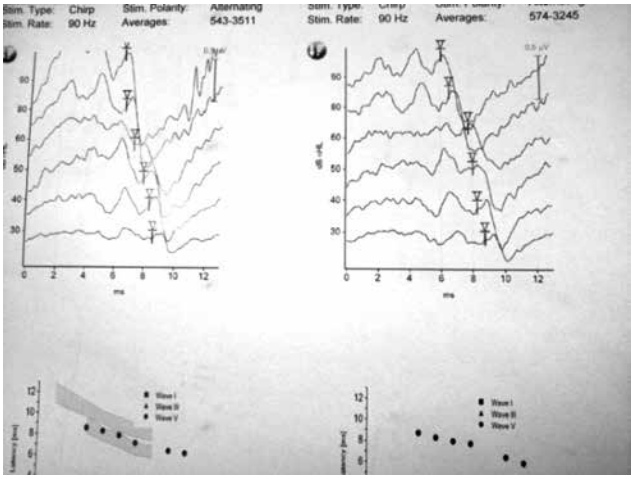
The tonal audiometry testing revealed bilateral sound conduction hearing loss. Moderate Hearing Loss of 48 dB in the right ear 48dB, Moderate Hearing Loss of 48 dB in the left ear.



The performed computed tomography showed atresia in the bony part of the external auditory canals, with a normally developed mastoid and inner ear.



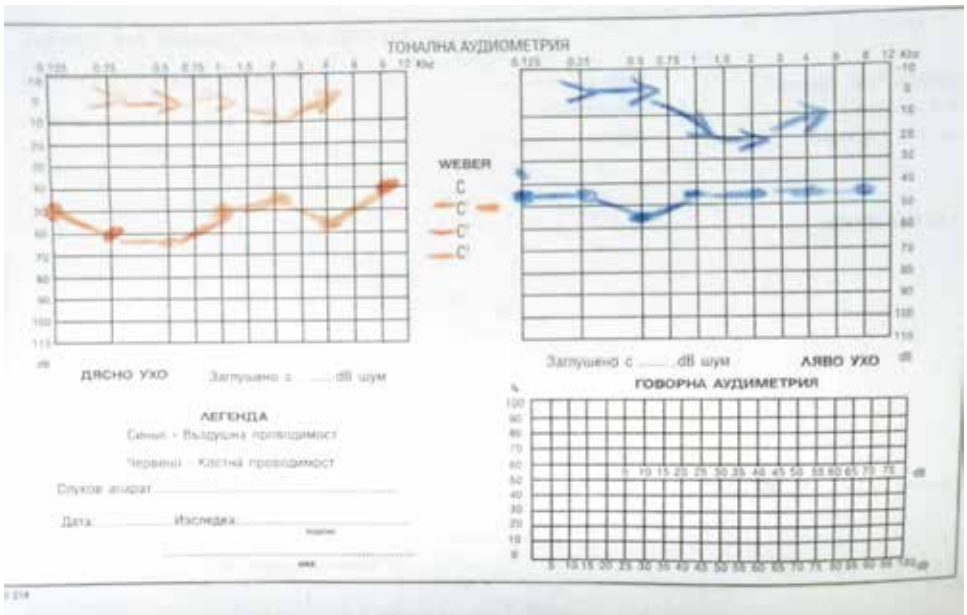




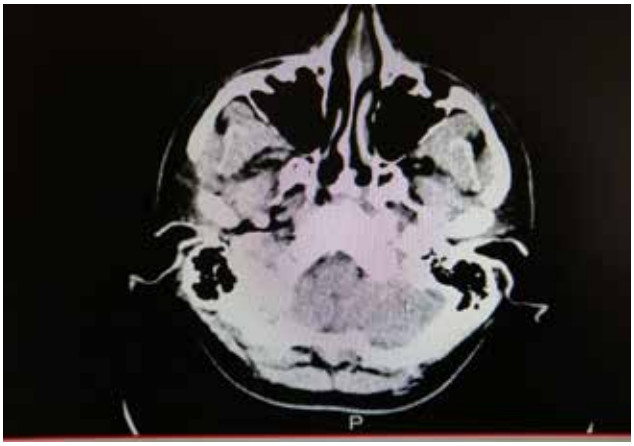
Air-conducted brainstem auditory evoked potentials revealed typical threshold waves of 80, 90 dB. Bone-conducted brainstem auditory evoked potentials revealed typical threshold waves of 30 dB.

Case 3

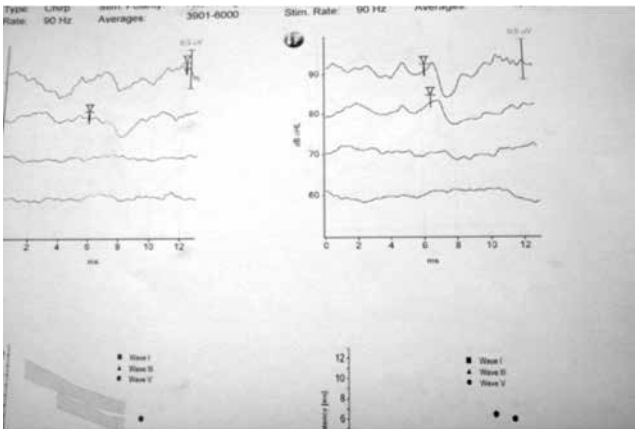
Subject 3 is the son of Subject 1 and her healthy husband. Born following a normal pregnancy at term. CAA was diagnosed after birth, without any other anomalies. Similarly to the mother and the daughter, the subjective and objective audiometry demonstrated that the patient has bilateral conductive hearing loss, in the case of right ear 53 dB and 53 dB for the left ear.

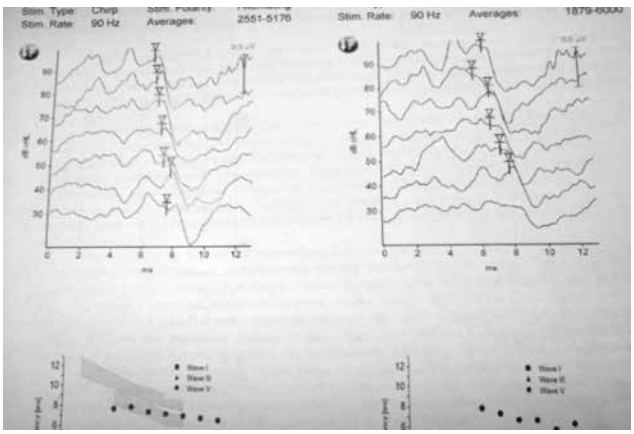


The computed tomography showed atresia in the bony part of the external auditory canals, with a normally developed mastoids and inner ear.



Air-conducted brainstem auditory evoked potentials revealed typical threshold waves of 80, 90 dB. Bone-conducted brainstem auditory evoked potentials revealed typical threshold waves of 30 dB.





Result

Atresia of the external auditory canals was found in all three subjects, and no other abnormalities were observed. The conducted tone audiometry testing and auditory brain stem response found bilateral sound conduction hearing loss. All three cases are classified as CAAII according to Altman (the external auditory canal is affected by complete bony atresia of its medial part or partial aplasia, ending in a blind fistula that leads to a rudimentary tympanic membrane).

Conclusion

The diagnosis of congenital atresia of the external auditory canal requires a team approach and very close contact with the parents. Bilateral atresia is treated surgically. Surgical treatment is difficult and not always successful, it is preferable to perform it at age 4-5. Long-term follow-up is important and requires cooperation between the otosurgeon and the patient.

References

1. Al-Qahtani Khalid, FRCS , Mohammed Aleisa, MD , Ahdi Albader, MD ,Vale´rie J. Brousseau, MDCM , Mary Lessard, FRCSC b, Melan Schloss, FRCSC and Tahera Islam, MD External auditory canal atresia: Surgical correction compared with bone anchored hearing devic. Journal of Taibah University Medical Sciences Volume 9, Issue 4, December 2014, Pages 307-310. DOI: 10.1016/j.jtumed.2014.05.005
2. Feenstra Ilse, Lisenka E.L.M. Vissers, Mirjam Orsel, Ad Geurts van Kessel, Han G. Brunner, Joris A. Veltman, Conny M.A. van Ravenswaaij-Arts Genotype–phenotype mapping of chromosome 18q deletions by high-resolution array CGH: An update of the phenotypic map American Journal of Medical Genetic 13 July 2007. https://doi.org/10.1002/ajmg.a.31850
3. Jacob R, S. Gupta, B. Isaacson, J.W. Kutz, Jr,[b] P. Roland, Y. Xi, and T.N. Booth High-Resolution CT Findings in Children with a Normal Pinna or Grade I Microtia and Unilateral Mild Stenosis of the External Auditory Canal Author information Article notes Copyright and License information Disclaimer AJNR Am J Neuroradiol. 2015 Jan; 36(1): 176–180. doi:10.3174/ajnr.A4067
4. Nguyen V.T, Paek G. Hu J. Presurgical CT Evalution on Congenital Aural Atresia. North Health Journal Articles 2015. https://academicworks.medicine.hofstra.edu/articles
5. Shah K, Knight B, Shermetaro C. External Ear Aural Atresia Publication Details
6. Yellon Robert F.Congenital external auditory canal stenosis and partial atretic plate Internacional of Journal of Pediatric Otorhinolaryngology Volume 73, Issue 11, November 2009, Pages 1545-1549. https://doi.org/10.1016/j.ijporl.2009.07.021

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