CASE REPORT

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Cochlear implantation for severe mixed hearing loss caused by Treacher Collins syndrome – a case report

Primena kohlearnog implanta kod bolesnika sa teškim mešovitim oštećenjem sluha izazvanim sindromom Tričer Kolins

Sanja Ostojić-Zeljković, Mina Nikolić, Sanja Djoković

University of Belgrade, Faculty of Special Education and Rehabilitation, Belgrade, Serbia

Abstract

Introduction. Treacher Collins syndrome (TCS) is a rare genetic condition characterized by typical head and neck malformations occurring in 1:50,000 newborns. Permanent conductive or mixed hearing loss of various degrees is diagnosed in 50% of individuals with TCS. A prerequisite for speech and language development in children with permanent mixed hearing impairment is the application of one of the bone conduction hearing aids. Choosing an adequate hearing aid in this case depends primarily on the degree of hearing impairment and the type of ear malformation. **Case report.** We present a female patient with multiple genetic malformations due to TCS. The patient was, immediately after birth, referred for audiological evaluation because of considerable ear and face malformations. Using a hearing test battery, permanent mixed, predominantly conductive, bilateral hearing loss of severe degree was diagnosed. The use of bone conduction hearing aids (including the Vibrant® Soundbridge middle ear implant) in the patient did not give the expected results – sufficient amplification for adequate speech and language development. Only after cochlear implantation at the age of nine did the patient's hearing threshold stabilize and her communication and academic potential develop to full capacity. **Conclusion.** If a middle ear implant is not sufficient for adequate amplification, cochlear implantation should be considered as an appropriate solution for treating severe permanent mixed hearing impairment in patients with TCS.

Key words:
bone diseases, developmental; cochlear implants; genetic diseases, inborn; hearing loss; prostheses and implants.

Apstrakt

Uvod. Tričer Kolins sindrom (TKS) je retko genetsko oboljenje koje karakterišu prepoznatljive malformacije u predelu glave i vrata, a javlja se kod 1:50 000 novorođene dece. Kod 50% osoba sa TKS dolazi do trajnog mešovitog oštećenja sluha različitog stepena. Preduslov za pojavu govora i jezika kod dece sa trajnim mešovitim oštećenjem sluha je primena nekog od slušnih aparata za koštanu provodljivost. Odabir adekvatnog slušnog pomagala u ovom slučaju zavisi pre svega od stepena oštećenja sluha i tipa malformacije organa čula sluha. **Prikaz bolesnika.** Prikazujemo bolesnicu sa mnogobrojnim genetskim malformacijama nastalim usled TKS. Bolesnica je zbog značajnih malformacija uva i lica odmah po rođenju upućena na audiološku procenu. Korišćenjem baterije za ispitivanje sluha dijagnostikovan je trajni mešoviti, obostrani, pretežno provodljivi gubitak sluha teškog stepena. Primena aparata za koštanu provodljivost (uključujući Vibrant® Soundbridge implantat srednjeg uva) kod bolesnice nije dovela do očekivanih rezultata – dovoljno pojačanje za odgovarajući razvoj govora i jezika. Tek nakon kohlearne implantacije u uzrastu od devet godina, kod bolesnice je došlo do stabilizacije praga sluha i razvoja komunikacijskih i akademskih potencijala do punog kapaciteta. **Zaključak.** Ukoliko implantat srednjeg uva nije dovoljan za adekvatno pojačanje, treba razmotriti kohlearnu implantaciju kao odgovarajuće rešenje za lečenje trajnog mešovitog oštećenja sluha teškog stepena kod bolesnika sa TKS.

Ključne reči:
kosti, bolesti, razvojne; kohlea, implantat; genetičke bolesti, urođene; sluh, gubitak; proteze i implantati.

Introduction

Treacher Collins syndrome (TCS) is a rare genetic condition characterized by typical malformations of the head and neck, occurring in 1:50,000 newborns [1]. Malformations of the outer, middle, and occasionally inner ear in 50% of TCS could cause permanent, usually conductive, hearing loss [2]. Diagnosis and treatment of those patients is

Correspondence to: Sanja Ostojić Zeljković, University of Belgrade, Faculty of Special Education and Rehabilitation, Visokog Stevana 2, 11 000 Belgrade, Serbia. E-mail: snjostojic@gmail.com

overly complex and requires a coordinated multidisciplinary approach and support during their lifetime. The evaluation protocol should include multiple audiological tests for estimating the type and degree of hearing loss to define a rehabilitation plan by a multidisciplinary team (an ear, nose, and throat doctor, an audiologist, and a surdologist) in order to enable normal speech and language development in those patients [3–5]. In TCS children with conductive or mixed hearing loss, adequate amplification using bone conduction hearing aids, such as bone anchored hearing aid (BAHA) or Vibrant® Soundbridge (VSB), is a prerequisite for speech and language development. Cochlear implantation (CI) for TCS patients with permanent severe mixed hearing loss is seldom mentioned in the literature as a suitable solution. Over the last four years, 659 publications have been cited in the Google Scholar database, and only nine of them have analyzed the rehabilitation effects in TCS patients with cochlear implants. In the patient presented in this paper, only CI has provided stable and sufficient amplification and subsequent progress of speech and language skills. Therefore, we report a case of a patient with TCS who showed considerable progress in listening and speech-language development after CI. This paper focuses on seeking an adequate amplification solution in the first nine years of the patient's life.

The research was approved by the Academic Council and Ethics Committee of the Faculty of Special Education and Rehabilitation, University of Belgrade, Serbia (No 89/1-1), and the parents' consent for this case report was obtained.

Case report

Medical status

The patient was a girl, born from a regularly monitored pregnancy, delivered at term with an Apgar score of 7/8. The following severe malformations were observed immediately after birth: microtia and severe stenosis of the outer ear canal, lack of the zygomatic bones, gothic palate, atresia of the anus, labia minor adhesions, and absent nipples. Neurologic findings included microcephaly, craniofacial dysmorphism, and slight hypotonia. The findings were suggestive of TCS. Genetic analysis showed inversion of chromosome 9 [karyotype 46, XX; Inv (9) (p12q13)]. Surgical treatment of congenital malformations commenced soon after birth. Anal atresia was operated on three times, as well as gynecological malformation of the labia minor. The girl had the ileus surgery at the age of ten months. She was treated twice for sepsis using combined antibiotic therapy over a prolonged period, and some of those antibiotics were ototoxic.

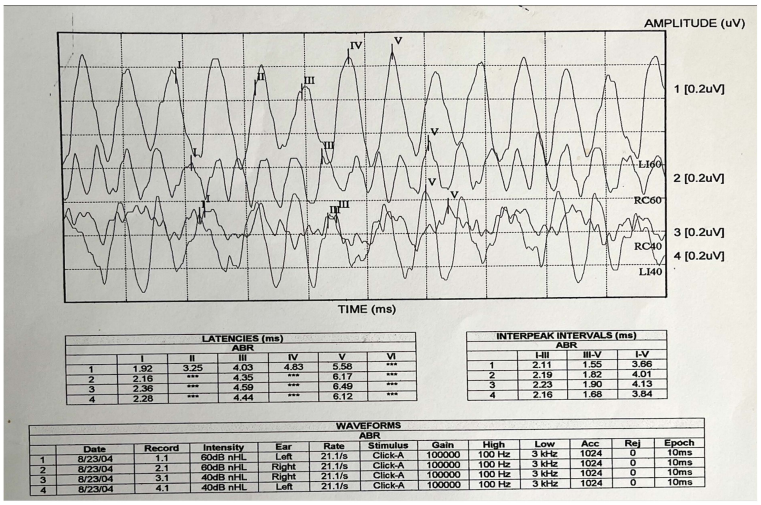
The first audiological evaluation was performed at the age of two months using behavior observation and a battery of electrophysiology tests [brainstem evoked response audiometry (BERA), otoacoustic emissions] (Figure 1A). However, the amplification and rehabilitation process had to be postponed because of the surgical treatment of life-threatening conditions. Periodic audiological check-ups have been conducted once a year using the aforementioned battery of audiological tests. At the age of three years and five months, the BERA test with click stimuli at 40 to 80 dB stimuli intensity confirmed moderate to severe mixed hearing loss with predominant conductive component due to outer and middle ear malformations (Figure 1B). Control BERA at the age of four years and eleven months confirmed previous findings with slight maturation of central auditory pathways (Figure 1C).

Amplification modalities

Although the hearing loss was diagnosed soon after birth, amplification had to be postponed until the age of 18 months due to numerous surgeries and recurrent life-threatening infections in this little girl. The girl used the BAHA® Softband and was enrolled in speech therapy. The device is considered suitable for mild to moderate conductive or mixed hearing loss, but this girl could only identify some isolated sounds with the device. She depended on the lip-reading support for speech understanding and remained quite passive. Surdologist reports based on standardized speech and language tests (Articulation test, Picture describing test, Vocabulary test by Vasić [6]) showed poor results and considerable delay. The delay in speech and language development, as well as in listening skills, became even more obvious over the next three years despite the intensive speech therapy. Since the girl had normal intellectual capacity, it became obvious that she was not amplified adequately. Pure tone audiometry (PTA) at the age of five years and five months showed severe mixed, predominantly conductive hearing loss (Figure 2). The BAHA® Intenso Softband device was provided. Although some progress was observed, surgical implantation of the device was needed to provide optimal amplification.

Eighteen months later, a multidisciplinary team consisting of an otological surgeon, an audiologist, a radiologist, a surdologist, and a psychologist discussed the options for surgical treatment. Multislice computed tomography of the temporal bone has shown good anatomical prerequisites for the implantation of the VSB middle ear implant in the right ear, which could provide better amplification and conditions for further speech and language development. The surgery was uneventful, but the listening improvement after the switch-on was not impressive, as shown in the PTA results. Seven months after the surgery, the hearing threshold started to fluctuate and deteriorate, and the rehabilitation progress was severely affected. That caused severe emotional distress in the nine-year-old girl and her parents. The fluctuation of hearing thresholds with VSB over time is shown in Figure 3 A–D.

Total hearing loss was observed seven months after the switch-on, with partial recovery three months later, but only up to 2 kHz and still with an ascendant curve. Fluctuations of the hearing threshold in PTA were the result of an effort to adjust the hearing map in the VSB processor with the aim of establishing the listening function that was expe-



LATENCIES (ms) ABR

	I	II	III	IV	V	VI
1	1.92	3.25	4.03	4.83	5.58	***
2	2.16	***	4.35	***	6.17	***
3	2.36	***	4.59	***	6.49	***
4	2.28	***	4.44	***	6.12	***

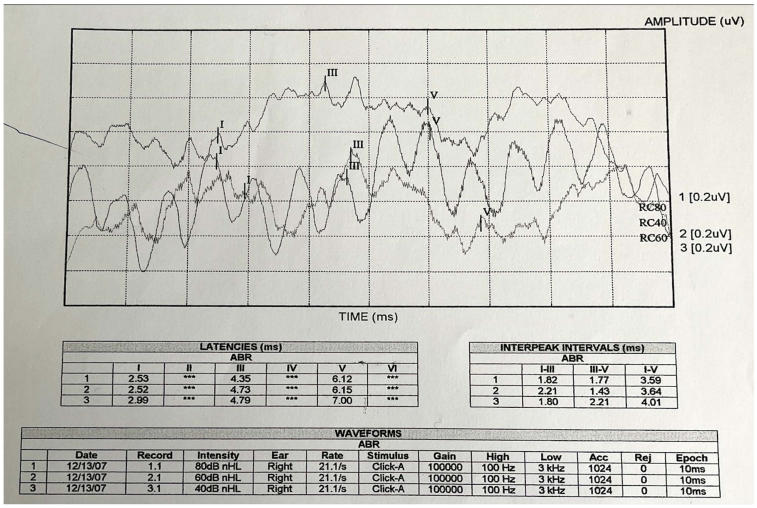
INTERPEAK INTERVALS (ms) ABR

	I-III	III-V	I-V
1	2.11	1.55	3.66
2	2.19	1.82	4.01
3	2.23	1.90	4.13
4	2.16	1.68	3.84

WAVEFORMS ABR

	Date	Record	Intensity	Ear	Rate	Stimulus	Gain	High	Low	Acc	Rej	Epoch
1	8/23/04	1.1	60dB nHL	Left	21.1/s	Click-A	100000	100 Hz	3 kHz	1024	0	10ms
2	8/23/04	2.1	60dB nHL	Right	21.1/s	Click-A	100000	100 Hz	3 kHz	1024	0	10ms
3	8/23/04	3.1	40dB nHL	Right	21.1/s	Click-A	100000	100 Hz	3 kHz	1024	0	10ms
4	8/23/04	4.1	40dB nHL	Left	21.1/s	Click-A	100000	100 Hz	3 kHz	1024	0	10ms

A)



LATENCIES (ms) ABR

	I	II	III	IV	V	VI
1	2.53	***	4.35	***	6.12	***
2	2.52	***	4.73	***	6.15	***
3	2.99	***	4.79	***	7.00	***

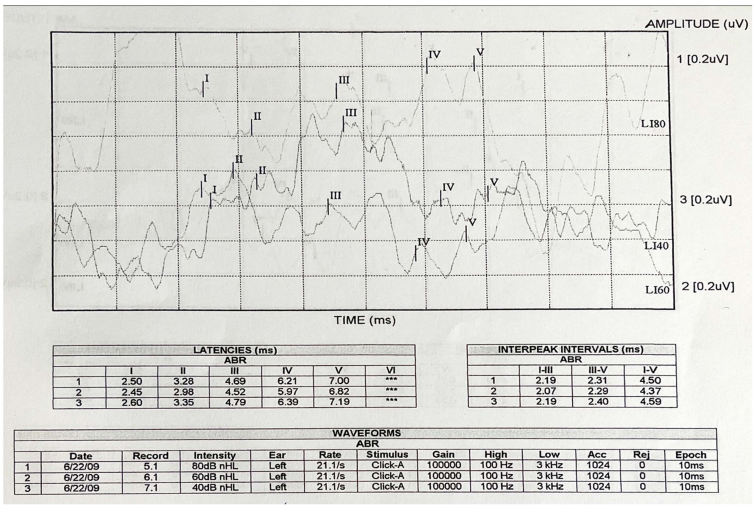
INTERPEAK INTERVALS (ms) ABR

	I-III	III-V	I-V
1	1.82	1.77	3.59
2	2.21	1.43	3.64
3	1.80	2.21	4.01

WAVEFORMS ABR

	Date	Record	Intensity	Ear	Rate	Stimulus	Gain	High	Low	Acc	Rej	Epoch
1	12/13/07	1.1	80dB nHL	Right	21.1/s	Click-A	100000	100 Hz	3 kHz	1024	0	10ms
2	12/13/07	2.1	60dB nHL	Right	21.1/s	Click-A	100000	100 Hz	3 kHz	1024	0	10ms
3	12/13/07	3.1	40dB nHL	Right	21.1/s	Click-A	100000	100 Hz	3 kHz	1024	0	10ms

B)



LATENCIES (ms) ABR

	I	II	III	IV	V	VI
1	2.50	3.28	4.69	6.21	7.00	***
2	2.45	2.98	4.52	5.97	6.82	***
3	2.60	3.35	4.79	6.39	7.19	***

INTERPEAK INTERVALS (ms) ABR

	I-III	III-V	I-V
1	2.19	2.31	4.50
2	2.07	2.29	4.37
3	2.19	2.40	4.59

WAVEFORMS ABR

	Date	Record	Intensity	Ear	Rate	Stimulus	Gain	High	Low	Acc	Rej	Epoch
1	6/22/09	5.1	80dB nHL	Left	21.1/s	Click-A	100000	100 Hz	3 kHz	1024	0	10ms
2	6/22/09	6.1	60dB nHL	Left	21.1/s	Click-A	100000	100 Hz	3 kHz	1024	0	10ms
3	6/22/09	7.1	40dB nHL	Left	21.1/s	Click-A	100000	100 Hz	3 kHz	1024	0	10ms

C)

Fig. 1 – A) Brainstem evoked response audiometry (BERA) at the age of 2 months shows a pathological auditory function with a lowered hearing threshold in both ears, which was determined by a click stimulus with an intensity of 40 and 60 dB. The absolute latencies of the I, III, and V waves are prolonged (normally expected 2, 4, and 6 ms), and the inter-wave intervals of the I–V waves are relatively preserved (up to 4 ms), with worse findings in the right ear. B) and C) The control BERA at the age of 3 years and 5 months and 4 years and 11 months, respectively, shows a moderate to severe hearing loss threshold (click stimulus 40 to 80 dB), with the delay of absolute latency and relatively preserved inter-wave intervals, indicating a mixed hearing impairment with a predominant conductive component.

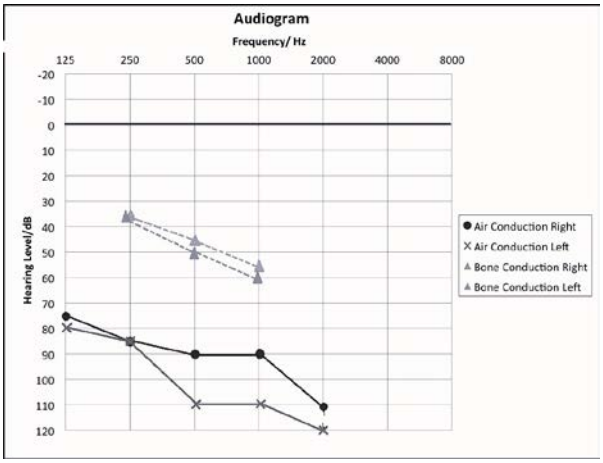


Fig. 2 – Pure tone audiometry at the age of 5 years and 5 months shows severe mixed, predominantly conductive hearing loss.

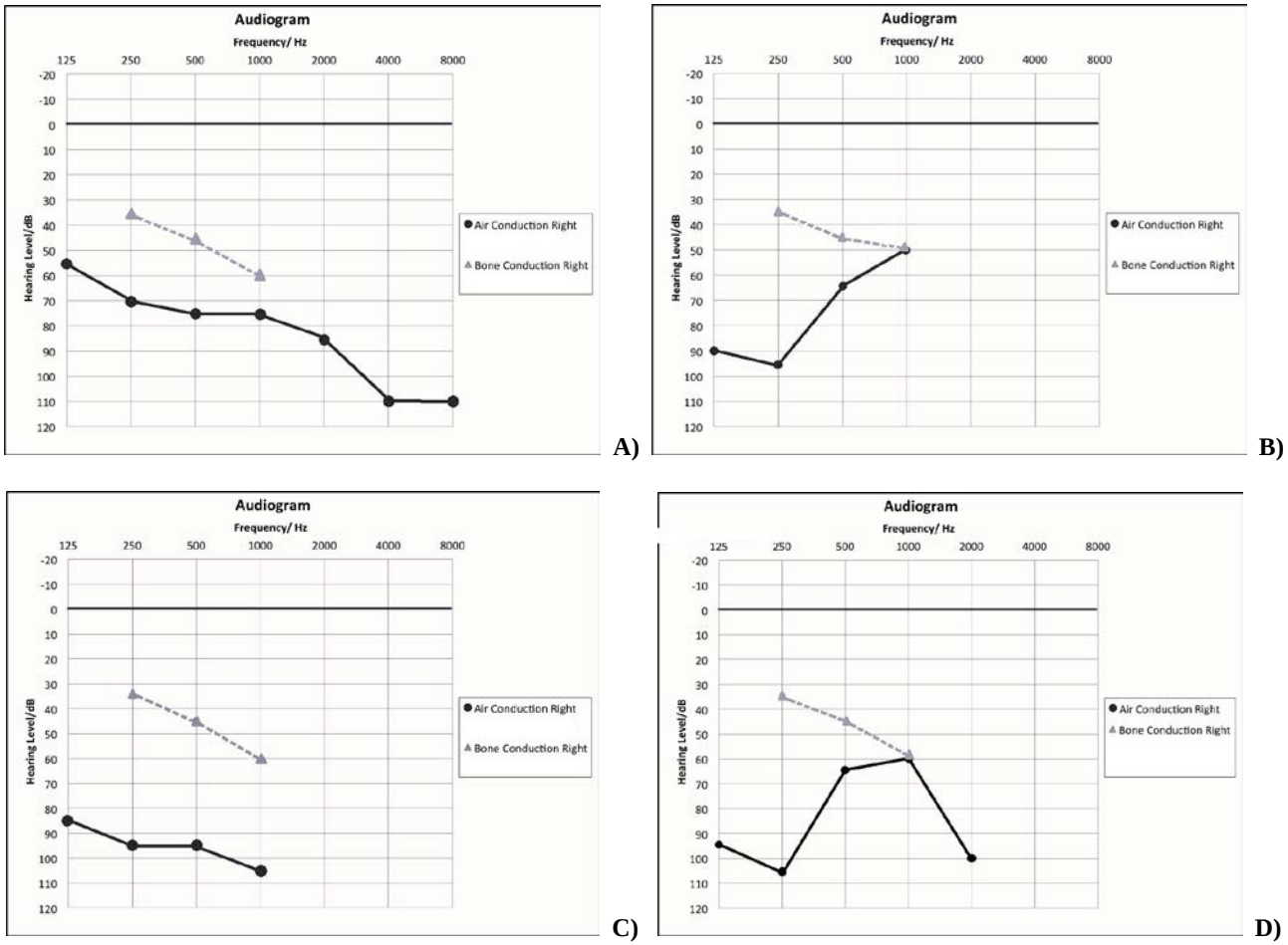


Fig. 3 – Pure tone audiometry of the right ear shows the fluctuation of hearing thresholds with Vibrant® Soundbridge (VSB) over time: A) 2 weeks after switch-on of VSB; B) 4 months after VSB use; C) 7 months after VSB use; D) 10 months after VSB use.

cted after the implantation. This did not provide functional hearing sufficient for speech and hearing development.

That situation called for further consultations of the multidisciplinary team, including engineers from the VSB Med-El® manufacturer, with the parents of the nine-year-old girl with TCS, and a consensus over the fact that CI was necessary was reached. The surgery was successful, and a Med-El® Sonata implant with a shorter Flex 28 electrode was implanted. Four weeks later, the implant was activated, and the Rondo® processor was fitted. The audiometry (Figure 4)

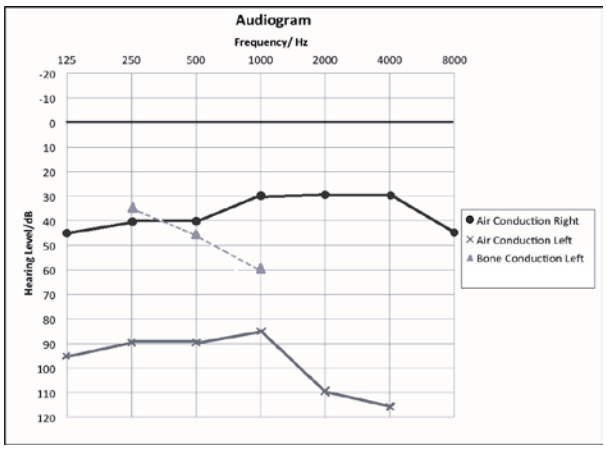


Fig. 4 – Pure tone audiometry with Rondo® processor and cochlear implant in the right ear shows a satisfactory aided hearing threshold (40 to 30 dB).

showed a satisfactory aided hearing threshold (40 to 30 dB) with a processor. Improved aided threshold provided satisfactory hearing and enabled further development of verbal communication. The girl attends a mainstream school with an individual education program and personal assistant.

Discussion

The motivation for reviewing this case is the rare example of CI in a patient with permanent mixed, predominantly sensorineural, with significant conductive component, hearing impairment caused by TCS [6–8]. TCS is a genetic disorder with characteristic malformations of the ears, face, eyes, and jaws, causing the impairment of respiration, speech, and sleep, frequently associated with psychological and behavioral issues due to strange appearance [8, 9]. Expression of various features and symptoms of TCS can differ considerably among patients depending on gene mutations with autosomal dominant or autosomal recessive inheritance [9]. Half of the patients with TCS could have some degree of hearing loss depending on the outer, middle, and/or inner ear malformation [10]. The choice of the amplification device depends on the anatomy of the ear and the degree of the hearing impairment. Bone conduction hearing instruments, external or implantable, are usually the best choice. The first bone conduction aids were mechanical vibrators, but the development of electronic devices improved bone conduction amplification considerably. Most of those devices are surgically implanted in the mastoid bone (BAHA®, Ponto, Bone Bridge) or the middle ear (VSB). Air-conduction hearing aids could be used only occasionally if the anatomy of the auricle and meatus is normal. BAHA® was introduced in 1984, supplying helpful solutions for conductive or mixed hearing loss of mild to moderate degree. Babies and infants could wear the device on a soft band before they are old enough for surgical implantation of the device in the mastoid bone (5–6 years of age) [5]. The surgical procedure is easy, and the percentage of complications is low. Most professionals dealing with the rehabilitation of children and adults with conductive or mixed hearing loss have a positive experience with BAHA® and VSB [11–14]. In most patients, BAHA® and VSB contribute to the improvement of auditory skills, speech, language, and communication development [14].

A cochlear implant is an electronic device suitable for profound sensorineural hearing loss. Over the last 30 years, it has enormously improved the speech and hearing rehabilitation of children with congenital or early acquired deafness. The selection criteria for CI are clearly listed [15, 16], and this patient did not meet the usual requirements from that list. Extended indications for CI in severe to profound mixed hearing loss in progressive otosclerosis have been introduced recently, but the indication for CI in children with similar hearing loss due to congenital malformation of the outer, middle, and/or inner ear has been rarely considered. Unable to achieve satisfactory amplification with a middle ear implant and dealing with constant fluctuation of the hearing threshold, our multidisciplinary team concluded that CI would be the only possible solution in this specific case. In children with congenital or early acquired deafness, the implantation age is a critical factor for the successful outcome of CI [17]. According to the Joint Committee on Infant Hearing, the optimal age for CI in congenital deafness is between 12 and 24 months [18, 19]. However, our patient was implanted at the age of nine. Her listening skills, communication, and academic achievement at that moment did not make her an ideal candidate for CI. However, she met the following criteria for the procedure: hearing threshold > 90 dB at 0.5, 1, and 2 kHz and no measurable hearing on higher frequencies; no benefit from other types of amplification – BAHA® Softband for three years, BAHA® Intenso Softband for 1.5 years and VSB for two years; open set test performance < 50%.

Conclusion

Although CI is rarely considered as a solution for permanent mixed, predominantly conductive hearing loss, such as in TCS, and the selection criteria for the procedure could

only partially be met, our patient with a cochlear implant achieved good and stable aided threshold, improved listening skills, speech, and communication capacity. Based on this experience, the authors would recommend CI as an ultimate solution for cases of TCS with severe to profound mixed hearing loss when all other bone conduction hearing aids fail to provide sufficient amplification for the development of auditory skills and speech, thus obstructing the achievement of the full communication capacity of a child. Selection criteria should be taken into consideration and adjusted individually in each specific case.

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Conflict of interest

The authors declare no conflict of interest.

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