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Case Report
Relato de Caso

Hearing and speech performance after cochlear implantation in children with Waardenburg syndrome

Desempenho de audição e linguagem pós implante coclear em crianças com síndrome de Waardenburg

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Keywords

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Waardenburg Syndrome
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ABSTRACT

Waardenburg syndrome (WS) is a rare autosomal-dominant syndrome that can be presented with sensorineural hearing loss. In this report, we describe the outcomes of three children with WS at zero, three, nine, twelve and sixty months after cochlear implant (CI) fitting. The outcomes were assessed using IT-MAIS (Infant-Toddler Meaningful Auditory Integration Scale – younger than 5 year), MAIS (Meaningful Auditory Integration Scale – older than 5 year), MUSS (Meaningful Use of Speech Scale), and categories of auditory performance and speech intelligibility. The results showed an improvement in auditory and language performance over time, two patients who used CI for 5 years achieved 100% in IT-MAIS and MUSS tests. In addition, both were able to understand sentences in open set and achieve fluent speech. Moreover, both reached fluency on auditory and language performance scale. The third patient with 50 months of follow-up and in the 48 months evaluation, is in category 5 of auditory performance and 3 of speech intelligibility. We concluded that all children who had low levels of hearing and language before cochlear implant have improved hearing and language skills after implantation and rehabilitation.

Descritores

Implante Coclear
Síndrome de Waardenburg
Perda Auditiva
Criança
Reabilitação

RESUMO

A Síndrome de Waardenburg (SW) é uma condição genética rara de padrão autossômico dominante e que pode cursar com perda auditiva neurossensorial. Nesse relato, apresentamos o desempenho da audição e linguagem de três crianças com SW, com zero, três, nove, doze e sessenta meses de seguimento após implante coclear (IC). As crianças foram avaliadas através dos questionários Escala de Integração Auditiva IT-MAIS (até 5 anos), MAIS (maiores de 5 anos), Escala de Utilização da Fala (MUSS) e categorizados quanto à audição e linguagem. Os resultados demonstraram uma melhora no desempenho da audição e linguagem ao longo do tempo, sendo que os dois pacientes que usaram o IC por 5 anos alcançaram 100% no IT-MAIS, MAIS e MUSS. Além disso, ambos foram capazes de compreender sentenças em conjunto aberto e possuem fala fluente, considerando-se as categorias de audição e linguagem. O terceiro paciente, com 50 meses de seguimento, encontra-se, na avaliação aos 48 meses, na categoria 5 de audição e 3 de linguagem. Conclui-se assim que as crianças da pesquisa, que apresentavam níveis baixos de audição e linguagem antes do implante coclear, passaram a apresentar melhores respostas auditivas e linguísticas após a implantação e reabilitação.

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INTRODUCTION

Waardenburg Syndrome (WS) is a genetic condition of autosomal dominant inheritance and estimated prevalence of 1:42,000 live births. It is manifested by a disorder of melanocytes, which can cause hypopigmentation of the iris, retina, skin and accessory structures and, mainly, hearing loss[1]. The syndrome manifests in 4 clinical types: types I and II are characterized by the presence and absence of *dystopia cantorum*, respectively. In types III and IV, which are rare, the patient presents musculoskeletal malformations and, in the latter type, Hirschsprung's disease or congenital aganglionic megacolon[2].

WS causes between 2% and 5% of all cases of congenital deafness[3]. The degree of hearing impairment in this syndrome can vary. However, in cases of severe or profound bilateral loss, rehabilitation with cochlear implant (CI) shows very good outcomes, especially compared to the general population of implanted children[4]. Due to the rarity of this syndrome, publications on post-treatment hearing results are scarce, and the number of individuals involved is not very high.

In this report we present a follow-up of the hearing and language performance of two children in the short (3 and 9 months), medium (12 months) and long term (60 months), and of one child in the short and medium term, all of whom have Waardenburg Syndrome and received Cochlear Implants. The children were assessed using IT-MAIS Auditory Integration Scale questionnaires (up to 5 years), MAIS (older than 5 years) and Speech Utilization Scale (MUSS) questionnaires, and categorized regarding hearing and speech[5].

PRESENTATION OF THE CLINICAL CASE

The study was approved by the institutional ethics committee under number 1.999.667 on April 4th 2017, and the children's guardians signed the Free and Informed Consent Form. Two of the children included were male and one female. All of them had been diagnosed with Waardenburg Syndrome and bilateral profound sensorineural hearing loss, and were rehabilitated with CI.

Table 1 presents data on the CI model, caregiver's education level and involvement in/frequency at rehabilitation.

Child 1, a male with type I WS, underwent CI surgery bilaterally at 22 months of age. Child 2, also male and with type I WS, underwent bilateral cochlear surgery also at 22 months of age and had to redo the surgery 9 months later, in the right ear, due to a displacement of the electrode. Child 3, female with type II WS, had the CI inserted bilaterally at 21 months of age and suffered no complication in the postoperative period. Table 2 shows the evolution of these three children regarding hearing and speech categories and performance in the IT-MAIS, MAIS and MUSS questionnaires. Auditory perception protocols and tests were applied before implantation and three, nine, twelve and sixty months after CI activation, during speech-language therapy sessions. Categorization was done based on results of the tests performed.

Child 3 did not perform the assessment at 60 months, as the data collection period had ended before the child reached this time of CI use. At 48 months, she had reached category 5 of hearing and 3 of speech.

Table 1. Model of cochlear implant, educational level of caregiver and frequency to rehabilitation sessions

Child	Cochlear implant	Educational level of caregiver	Frequency to rehabilitation
1	Contour Advance (Cochlear)	Complete high school	100%
2	Contour Advance (Cochlear)	Complete higher education	100%
3	HiFocus (Advanced Bionics)	Complete higher education	100%

Table 2. Evolution of hearing and speech in implanted WS patients

Individual	WS type	Age at CI surgery		Post-CI evaluations (months)				
				0	3	9	12	60
1	I	22 months	HSC	H0 S1	H1 S1	H2 S2	H3 S2	H6 S5
			IT-MAIS/MAIS	2.50%	22.50%	95%	95%	100%
			MUSS	7.50%	12.50%	80%	85%	100%
2	I	22 months	HSC	H0 S1	H1 S1	H2 S1	H3 S2	H6 S5
			IT-MAIS/MAIS	5%	22.50%	42.50%	77.50%	100%
			MUSS	5%	10%	30%	60%	100%
3	II	21 months	HSC	H0 S1	H1 S1	H2 S1	H2 S2	*
			IT-MAIS/MAIS	5%	10%	52.50%	70%	
			MUSS	20%	22.50%	40%	54%	

Caption: WS = Waardenburg Syndrome; CI = Cochlear Implant; HSC = Hearing and Speech Category; IT-MAIS = *Infant Toddler Meaningful Auditory Integration Scale*; MUSS = *Meaningful Use of Speech Scale*; H = Hearing; S = Speech; *Patient with only 12 months follow-up

DISCUSSION

Regarding audiological repercussion in different types of WS, sensorineural hearing loss is present in 35% to 75% of patients with type I WS and in 55% to 91% of patients with type II WS[6,7]. Two of the children in this study were classified as type I and one as type II, and all of them had total or partial iris heterochromia and congenital sensorineural hearing loss. They were users of other auditory devices, but these resources fell short of facilitating speech perception; as a result, the children were prescribed rehabilitation with CI.

One of the three children (child 2) suffered a displacement of the electrode of the right implant, which needed to be reimplanted. Available data show that this is a common cause of CI reimplantation[8], having no relation to WS itself or any of its particularities. Exchange of implants normally occurs for two reasons: damage or failure of a certain component or update of a model, when implant technology so allows[9]. In this study, in addition to reimplantation by extrusion, the speech processor (child 1) was also bilaterally exchanged due to an upgrade that became available, with no relation to failures or malfunctions.

All patients described in this paper received bilateral implants and were rehabilitated by auditory verbal therapy. Assessment of auditory skill development was done using the IT-MAIS, MAIS and MUSS tests. These questionnaires were applied to parents or caregivers and contained questions regarding the child's auditory behavior in their daily lives (IT-MAIS and MAIS) and oral communication skills or attempts (MUSS). It is noteworthy that, after activation of the CI, auditory and speech skills were developed continuously and progressively over time in all individuals, as expected in implanted patients[5]. Two children (children 1 and 2) reached the maximum scores on IT-MAIS, MAIS and MUSS after five years of using the device, and achieved fluent oral communication. The reimplantation to which child 2 was submitted did not negatively affect his performance. Regarding child 3, who has currently been using the CI for 50 months, it has not yet been possible to carry out a final assessment.

WS patients are not usually cognitively impaired[6]. In addition, evidence shows that hearing loss in this syndrome is mainly related to cochlear histological impairment.[6] These facts are relevant to explain the good performance of these children when implanted, both in the results shown here and in other series in the literature, even when compared with non-syndromic individuals[2,10,11]. There are reports of patients with WS who also present cochlear bone malformations, which could pose greater difficulties to the implantation procedure[10]. The children described here did not suffer from this type of malformation.

Engagement of caregivers and parents is an important factor in the rehabilitation of children who receive cochlear implants. In a study evaluating demographic variables that could influence the school performance of implanted children, the authors observed that age at implantation, socioeconomic status, ethnicity and educational level of parents were significant factors for better performance[12]. In addition, a multivariate analysis of these factors found that the most important correlation was with parents' socio-educational level[12]. As for the educational level of parents and caregivers, the cases reported in this study involved individuals with Bachelor's or equivalent degrees (child 2 and child 3) and upper secondary education (child 1), which may have influenced the good performance observed here. Caregivers' commitment to rehabilitation is another important factor to be highlighted in the post-surgical process. Regular visits to the therapist associated with family engagement are essential points for the best possible performance to be achieved[13]. The caregivers of the children presented here have shown an appropriate engagement, with 100% attendance at rehabilitation sessions.

Recent recommendations on the best age for implantation suggest that the procedure should occur as early as possible, provided that the audiological diagnosis has been established consistently. Implantation between 12-18 months of age greatly increases a child's potential to develop speech at a rate similar to that of normal listeners. Children who receive the implant after 12-18 months, but before 3 years of age, may also enjoy a potential similar to that of normal hearing children; however, the variability of results is broader, with a greater possibility of delay when compared with normal hearing subjects in the same age group[14]. Two children were operated at 22 months and one at 21 months of age, a time considered appropriate, with the possibility of full hearing development.

Adequate engagement of parents, who had a high socio-educational status, associated with the early age of implantation can be identified as important factors in the good outcome shown by the children presented here.

We also verified that the MUSS scale scores evolve more slowly than the IT-MAIS and MAIS scales', showing that auditory perception skills are acquired more quickly than speech skills. Similar results regarding these scales were found in a study with Portuguese children who had profound deafness of unspecified etiology and who were also rehabilitated with CI[15].

FINAL REMARKS

From this study, we concluded that the described children with WS, who showed poor audiological results before CI activation, presented better auditory and speech responses after rehabilitation with this device.

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Author contributions

JFP: registration of the study, data collection and writing of the manuscript; APK: data collection and writing of the manuscript; CAO: registration of the study, data collection and writing of the manuscript.