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Cochlear implant rehabilitation outcomes in Waardenburg syndrome children

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ABSTRACT

Objectives: The purpose of this study was to review the outcomes of children with documented Waardenburg syndrome implanted in the ENT Department of Centro Hospitalar de Coimbra, concerning postoperative speech perception and production, in comparison to the rest of non-syndromic implanted children.

Methods: A retrospective chart review was performed for children congenitally deaf who had undergone cochlear implantation with multichannel implants, diagnosed as having Waardenburg syndrome, between 1992 and 2011. Postoperative performance outcomes were assessed and confronted with results obtained by children with non-syndromic congenital deafness also implanted in our department. Open-set auditory perception skills were evaluated by using European Portuguese speech discrimination tests (vowels test, monosyllabic word test, number word test and words in sentence test). Meaningful auditory integration scales (MAIS) and categories of auditory performance (CAP) were also measured. Speech production was further assessed and included results on meaningful use of speech Scale (MUSS) and speech intelligibility rating (SIR).

Results: To date, 6 implanted children were clinically identified as having WS type I, and one met the diagnosis of type II. All WS children received multichannel cochlear implants, with a mean age at implantation of 30.6 ± 9.7 months (ranging from 19 to 42 months). Postoperative outcomes in WS children were similar to other nonsyndromic children. In addition, in number word and vowels discrimination test WS group showed slightly better performances, as well as in MUSS and MAIS assessment.

Conclusions: Our study has shown that cochlear implantation should be considered a rehabilitative option for Waardenburg syndrome children with profound deafness, enabling the development and improvement of speech perception and production abilities in this group of patients, reinforcing their candidacy for this audio-oral rehabilitation method.

1. Introduction

Waardenburg syndrome (WS) is an inherited disorder defined by the association of skin, iris and hair pigmentation abnormalities, and varying degrees of sensorineural hearing loss. This disorder was named after a Dutch ophthalmologist, Petrus Waardenburg, who first described it in 1947 [1]. His investigation of deaf patients with depigmentation and dysphormology features led to the description of Waardenburg syndrome, now known as type I WS. Since then, four clinical types have been identified, depending on the phenotype and presence of additional features [2].

Phenotypic findings in WS type I syndrome (WSI) include broad nose root owing to lateral displacement of eye's inner canthus (dystopia canthorum), depigmented patches of skin and hair (white forelock, white eyelashes, leukoderma), premature graying, vivid blue eyes or heterochromic irides, and confluent eyebrows (synophoris). WS type II (WSII) differs from WSI by the absence of dystopia cantorum and has been divided into 4 subtypes (A, B, C, D) depending on the mutation involved. In type III WS (WSIII), also called Klein-Waardenburg, musculoskeletal abnormalities of the upper limbs are added to WSI phenotypic features. Type IV WS (WSIV) [3], or Shah-Waardenburg syndrome, is characterized by WS type II features and Hirschsprung disease [4].

The estimated prevalence of WS is approximately 1 case *per* 42,000 individuals [2]. WSI is 1.5–2 times more common than WSII; type III and IV are far rarer forms of WS. It is an autosomal disorder with genetic heterogeneity and not all of its forms are dominantly inherited, as previously assumed [5].

Sensorineural hearing loss is quite a frequent feature in Waardenburg syndrome, reported in 60% and 90% of patients with type I and type II, respectively. Bilateral forms of hearing loss are more frequent than unilateral, but not necessarily symmetrical; various audiogram shapes can be found, without a typical audiometric pattern [6]. The extent of hearing impairment is a

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quite variable feature within and between families, ranging from no measurable clinical loss to profound deafness [6–8], the last being observed in 42% of patients with WSI and in 73% of patients with WSII [9].

Waardenburg syndrome is responsible for about 2% of profound congenital hearing loss. Congenitally deaf children with WS, severely or profoundly impaired with limited hearing aids benefit, have been integrating cochlear implant programs with encouraging results comparable to those reported for the general population of implanted children [10].

The aim of this study was to review the outcomes of children with documented Waardenburg syndrome implanted in our Cochlear Implant Department, concerning postoperative speech perception and production, in comparison to the rest of non-syndromic implanted children. Surgical and radiological data were also assessed in this subset of patients. By exposing our experience of cochlear implantation in this particular group of patients, we intended to add more useful data concerning CI rehabilitation in WS children, considering the limited information available to medical community imposed by the rarity of this syndrome.

2. Materials and methods

A retrospective chart review was performed for congenitally deaf children who were implanted in the Cochlear Implant Unit – Department of Otorhinolaryngology at Centro Hospitalar de Coimbra, between 1992 and 2011. Of 379 children who had undergone implantation with multichannel cochlear implants, seven cases were diagnosed as having WS. Data recorded included age at time of surgery, clinical and radiological features, operative and perioperative course.

Postoperative performance outcomes were also assessed and compared to results obtained by children with non-syndromic congenital deafness also implanted in our department. This second group, which acted as control, comprised 261 subjects (56.8% males and 43.2% females), with a mean age at implantation of 36.7 ± 18.6 months. The right ear was the most commonly implanted, in 88.1% children. Unknown etiologies contributed to the deafness observed in most children (73.9%); documented genetic anomalies represented 22.6% of deafness etiology and 3.5% of control group children had suffered gestational infection. Preoperatory imagiologic findings included: unilateral acoustic nerve atrophy (1.92%), enlarged endolymphatic sac fossae (1.53%), enlarged vestibular aqueduct (0.77%), cochlear lumen narrowing or irregularity (3.83%), posterior labyrinth narrowing (0.77%).

Postoperative open-set auditory perception skills were evaluated by using european portuguese word discrimination tests (monosyllabic word test, number word test and words in sentence test) and also included vowels discrimination test; items from age-appropriate lists were presented at an average level of 65 dB SPL, with cochlear implant use, in a soundproof room without visual clues. Results were recorded as percentage of items correctly repeated. In monosyllabic and numbers tests, the percentage of correct phonemic items was also taken into consideration. Meaningful Auditory Integration Scales (MAIS) and Categories of Auditory Performance (CAP) were also measured.

Speech production was further assessed and included results on Meaningful Use of Speech Scale (MUSS) and Speech Intelligibility Rating (SIR).

3. Results

To date, among the pediatric population submitted to cochlear implantation in our institution, 6 children (1.58%) were clinically identified as having Waardenburg syndrome type I, and one met the diagnosis of type II WS (0.26%). Data concerning genetic analysis was available for the child with WSII, which confirmed a MITF gene mutation.

All subjects had documented bilateral profound sensorineural hearing loss with minimal or no benefit from appropriate hearing fitting and limited acquired language skills provided by amplification, prior to cochlear implant surgery.

Results of preoperative radiologic assessment were available in all WS patients, including temporal bone high-resolution computed tomography (CT) and magnetic resonance imaging (MRI) of head and inner ear; none of them revealed any inner ear malformation except for one single case of WSI, whose CT scan images revealed a discrete enlargement of vestibule cavity, all three semicircular canals and internal acoustic canal, not confirmed on MRI.

All WS children received multichannel cochlear implants, with an average age at implantation of 30.6 ± 9.7 months (ranging from 19 to 42 months).

Cochlear implant surgery was performed by cortical mastoidectomy followed by posterior tympanostomy and cochleostomy, with a post-aural approach. No intraoperative complications occurred and electrode insertion in cochlear *scala tympani* was well succeeded without any reported difficulties.

The postoperative course was uneventful for all implanted WS children. Postimplantation data are displayed in Table 1.

After a post-implantation follow up time ranging from 24 to 115 months (mean 57.71 ± 30.34), auditory perception skills achieved by the Waardenburg children group (Table 2) are confronted with non-syndromic implanted children's results in Fig. 1 and Table 3.

Amongst Waardenburg syndrome children, the mean percentage of open-set speech recognition was 45.75%, 91.66% and 60.22%, in words in sentence, numbers and monosyllables tests, respectively. Overall, speech perception skills were slightly weaker in these children when compared to the remaining implanted children group, except in vowels test, in which they attained 100% of correct answers; in numbers test WS children also performed better.

Table 1
Postimplantation general data of WS children.

	Patient no.						
	1	2	3	4	5	6	7
Age at implantation (months)	42	25	39	41	23	20	19
Duration of follow up (months)	115	80	54	44	44	43	24
WS type	I	II	I	I	I	I	I
Sex	Male	Male	Male	Female	Female	Male	Female
Device	CI24R CS Contour	CI24R CA Advance	CI24RE CA	CI24RE CA	CI24RE CA	CI24RE CA	CI24RE CA
Speech processor	CP810 (previous SPRINT)	SPRINT	FREEDOM	FREEDOM	FREEDOM	FREEDOM	FREEDOM
Implanted side	Right	Right	Right	Left	Right	Left	Right
Speech coding strategy	ACE (previous SPEAK)	ACE	ACE	ACE	ACE	ACE	ACE
Stimulation mode	MP 1 + 2	MP 1 + 2	MP 1 + 2	MP 1 + 2	MP 1 + 2	MP 1 + 2	MP 1 + 2

SPEAK, spectral peak coding; ACE, advanced combination encoders; MP, monopolar.