Cochlear implantation in children with Waardenburg syndrome

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Abstract

Waardenburg syndrome is an autosomal-dominant trait resulting from mutations occurring in different genes. It is often characterized by varying degrees of: congenital hearing loss; dystopia canthorum; synophrys; broad nasal root; depigmentation of hair (white forelock), skin or both; and heterochromic or hypochromic irides.

A retrospective case study was done to assess speech perception, speech production, general intelligence and educational setting in six profoundly hearing-impaired children with Waardenburg syndrome (four with type I, one with type II and one with type III) ranging in age from two years to 14 years, seven months (mean = six years, six months). None of the patients had malformation of the cochlea and were implanted using Nucleus 22/24 and Med-el combi40+. Five out of the six cases were of average intelligence and one had a borderline intelligence quotient. The follow-up period ranged from one year, 10 months to six years, six months (mean = three years, six months) after implantation. The evaluation of auditory perception in patients was accomplished using the Persian Auditory Perception Test for the Hearing-Impaired, a Persian Spondee words test and the Categories of Auditory Performance Index. The Speech Intelligibility Rating test was used to evaluate speech production ability. All the patients' speech perception and speech intelligibility capabilities improved considerably after receiving the implants, and they were able to be placed in regular educational settings. Patients used their cochlear-implant devices whenever awake, implying that they benefitted from the devices. We suggest that any further expansion of cochlear-implantation criteria in children include those with Waardenburg syndrome.

Key words: Waardenburg Syndrome; Cochlear Implantation; Speech Perception

Introduction

Waardenburg syndrome (WS) is an inherited disorder often characterized by varying degrees of hearing loss and changes in skin and hair pigmentation. It was first described in 1948 by a Dutch ophthalmologist.[1] Other characteristics of WS are: dystopia canthorum; synophrys; broad nasal root; depigmentation of hair (white forelock), skin, or both; and heterochromic or hypochromic irides.[2] The highest reported incidence is among Kenyan Africans. Waardenburg syndrome accounts for between 2 and 5 per cent of all cases of congenital deafness.[3] There are four clinical subtypes: type I, characterized by the presence of dystopia canthorum, sensorineural hearing loss, heterochromic irides, white forelock, hypopigmentation and synophrys; type II, being the features of type I without dystopia canthorum; type III, or Klein–Waardenburg syndrome, which has type I features plus hypoplastic muscles and contractures of the upper limbs; and type IV, or Shah–Waardenburg syndrome, characterized by type II features and Hirschsprung's disease.[2,4,5] Although in some studies enlargement of the vestibular aqueduct and the upper vestibule, narrowing of the internal auditory canal porus and hypoplasia of the modiolus are reported as features of WS, abnormality of the bony labyrinth is not a frequent finding in WS with congenital deafness, particularly in WS type I. Therefore, both the otologist and the audiologist must bear in mind that the inner-ear anatomy of most of these cases is suitable for cochlear implantation.[6,7] Some authors have reported motor delay and mental retardation in WS children.[8–10] Children with syndromic deafness show less improvement in receptive language and speech intelligibility after cochlear implantation.[11] One of the cochlear-implanted patients reported in the Waltzman study, who had Waardenburg

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TABLE I
SUMMARY OF CASES

Case	WS type	Age (years, months) At identificaton	At CI	At Final evaluation
1 (RM)	I	1, 0	8, 3	14, 9
2 (HN)	III	0, 6	14, 7	18, 4
3 (ShSh)	I	2, 0	6, 8	11, 5
4 (AKh)	II	0, 2	4, 5	6, 5
5 (AP)	I	0, 7	3, 7	5, 5
6 (BM)	I	0, 7	2, 0	4, 7

WS = Waardenburg syndrome; CI = cochlear implantation

TABLE II
CATEGORIES OF AUDITORY PERFORMANCE SCALE

Category	Description
0	Displays no awareness of environmental sounds
1	Awareness of environmental sounds
2	Responds to speech sounds
3	Recognizes environmental sounds
4	Discriminates at least two speech sounds
5	Understands common phrases without lip-reading
6	Understands conversation without lip-reading with a familiar talker
7	Can use telephone with a familiar talker

syndrome, showed poor performance in the Glendonald Auditory Screening Procedure Word Test two years after cochlear implantation, compared with other multi-handicapped children.[12] Based on our literature review, only one single case study of a cochlear-implanted child with type I WS has been reported so far. This patient achieved 58 per cent of the total score on open-set speech perception tests two years after cochlear implantation.[13]

In this report, we will describe the results of a retrospective study addressing speech perception, speech production, general intelligence, educational setting and other findings in children with WS who had undergone cochlear implantation.

Material and methods

At the time of writing, six profoundly hearing-impaired children with WS (four with type I, one with type II and one with type III), ranging in age from two years to 14 years, seven months (mean = six years, six months), had received cochlear implants (using the Nucleus 22/24 and Med-el combi40+ devices) at the Iran Cochlear Implant Center (Table I). Five of these children received appropriate amplification before evaluation for cochlear implant; nevertheless, these children did not benefit from their hearing aids due to profound hearing loss.

Patients were subjected to a variety of speech tests; the Persian Auditory Perception Test for the Hearing-Impaired (PAPT/HI),[14] the Persian Spondee words test and the Categories of Auditory Performance (CAP) scale[15] were used for auditory perception evaluation. The Speech Intelligibility Rating (SIR) scale[16] was used for evaluation of speech production ability. The PAPT/HI consists of 50 items based on degree of difficulty, in three levels. The first level has 16 items that evaluate auditory awareness, duration, intensity, pitch identification and identification of words and sentences through using suprasegmental information. The second level comprises 22 items for evaluating vowel and consonant perception through segmental information; these items also evaluate identification of phonemes, words and phrases in closed sets using segmental information. The last level of the test has 12 items intended to measure comprehension of closed and open sets. The total score of the test is 100. A male speaker presented the test items at an average presentation level of 70 dB sound pressure level (SPL) in a soundproof room.

The Persian Spondee word test includes 20 closed sets of four spondee pictured words, which are spoken with equal emphasis on each syllable at an average presentation level of 70 dB SPL. The child is required to match the picture with the correct word. The score is the percentage of words correctly identified out of 20.

The CAP scale quantifies the auditory receptive abilities of linguistically compromised profoundly deaf children, in a clinical setting. It has an eight-point scale and ranges from category 0 (no awareness of environmental sounds) through to category 7 (the ability to use a telephone with a known speaker) (Table II).

The SIR scale provides a standardized rating of a child's speech-production skills in five categories, ranging from 'pre-verbal' to 'intelligible to all' (Table III).

Raven's Colored Progressive Matrices (RCPM) were used to measure patients' intelligence. This is a nonverbal intelligence test for children aged five years and over.[17,18]

Results

Case 1 (RM)

RM was eight years, three months old at cochlear implantation. He was diagnosed with type-I WS, with revealed pure-tone averages (PTAs) of 90 dB hearing level (HL) in the better ear. He received no perceptible benefit from hearing aids. A computed

TABLE III
SPEECH INTELLIGIBILITY RATING SCALE

Category	Description
1	Prerecognizable words in spoken language. Child's primary mode of everyday communication may be manual
2	Connected speech is unintelligible. Intelligible speech is developing in single words when context and lip-reading cues are available
3	Connected speech is intelligible to a listener who concentrates and lip-reads within a known context
4	Connected speech is intelligible to a listener who has little experience of a deaf person's speech. The listener does not need to concentrate unduly
5	Connected speech is intelligible to all listeners. The child is understood easily in everyday contexts

TABLE IV
CASE 1 (RM) OUTCOMES

Test	Timing (year, month)						
	Pre-implant	1	2	3	4	5	6, 6
APT/HI	0%	35%	ND	ND	ND	65%	65%
Spondee words test	0%	20%	ND	ND	ND	90%	90%
CAP	1	4	ND	ND	ND	5	5
SIR	0	0	ND	ND	ND	4	4

APT/HI = Auditory Perception Test for the Hearing-Impaired; CAP = Categories of Auditory Performance Index; SIR = Speech Intelligibility Rating; ND = no data

tomography (CT) scan showed a normal cochlea. The parents had noticed no motor delay. RM showed no mental retardation, based on measuring of intelligence by RCPM. In the family, his mother showed dystopia canthorum. The child was implanted in 1997 with the Nucleus spectra device. Twenty inserted electrodes were activated on the map in CG mode. Six years, six months after implantation RM achieved scores of 65 per cent for the PAPT/HI test, 90 per cent for the Spondee words test, and levels 5 and 4 in the CAP and SIR scales, respectively (Table IV). At the time of writing, RM was a ninth-grade student at a regular high school and was using his cochlear implant during all waking hours.

Case 2 (HN)

HN was diagnosed with type-III WS at 6 months of age, showing dystopia canthorum, patchy depigmentation of skin, flexion contractures in the feet, hypoplasia of the musculoskeletal system and hearing loss. He was referred for occupational therapy at 18 months because of developmental delay in motor skills. Nevertheless, he was not able to walk until three years of age. Audiometry at four years revealed a 75-dB bilateral sensory neural hearing loss, and HN was fitted with hearing aids. At this age, he started education in a kindergarten for the deaf. He was placed in a regular primary school from grade six. He showed no signs of mental retardation, based on RCPM. His hearing loss was progressive, and audiometry at 14 years revealed a bilateral profound hearing loss. Therefore, at 14 years, seven months, HN received cochlear implantation with the Nucleus CI24 device. All 22 electrodes were activated on the map with ACE strategy. Three years, nine months after implantation, HN achieved 75 per cent for the PAPT/HI test, 100 per cent for the Persian Spondee words test, and levels 5 and 5 in the CAP and SIR scales, respectively (Table V). After cochlear implantation, he continued education in regular school to grade nine.

Case 3 (ShSh)

ShSh was six years, eight months old at cochlear implantation. She had been diagnosed with type-I WS at two years of age, showing dystopia canthorum, pale blue eyes, broad nasal root, white forelock and bilateral profound deafness. Pure-tone averages in the better ear showed 95 dB HL. ShSh showed borderline intelligence as measured by RCPM. A medical history of the family revealed that her father, uncle and two of her cousins suffered from WS without hearing loss; three of her siblings had also suffered from WS and had died a few days after birth. ShSh received no benefit from hearing aids. A CT scan showed a normal cochlea. ShSh was implanted with a Nucleus CI24 device. Nineteen electrodes were activated on the map with ACE strategy. Four years, nine months after implantation, ShSh achieved scores of 35 per cent for the PAPT/HI test, 55 per cent for the Persian Spondee words test, and levels 4 and 3 in the CAP and SIR scales, respectively (Table VI). At the time of writing, ShSh was a fourth-grade student in a regular school and used her device during all waking hours.

Case 4 (AKh)

AKh was four years, five months of age at cochlear implantation. He was diagnosed with type-II WS at two months, showing white forelock, heterochromia and bilateral profound deafness. A CT scan showed a normal cochlea. AKh was implanted using a Nucleus CI24 device. Twenty-one electrodes were activated in the map with ACE strategy. Two years after implantation, AKh achieved scores of 45 per cent for the PAPT/HI test, 90 per cent for the Persian Spondee words test, and levels 4 and 3 in the CAP

TABLE V
CASE 2 (HN) OUTCOMES

Test	Timing (year, month)			
	Pre-implant	1	2	3, 9
APT/HI	10%	50%	80%	75%
Spondee words test	25%	65%	100%	100%
CAP	1	5	5	5
SIR	5	5	5	5

APT/HI = Auditory Perception Test for the Hearing-Impaired; CAP = Categories of Auditory Performance Index; SIR = Speech Intelligibility Rating

TABLE VI
CASE 3 (SHSH) OUTCOMES

Test	Timing (year, month)				
	Pre-implant	1	2	3	4, 11
APT/HI	0%	15%	20%	ND	35%
Spondee words test	0%	20%	50%	ND	55%
CAP	0	4	4	ND	4
SIR	1	1	2	ND	3

APT/HI = Auditory Perception Test for the Hearing-Impaired; CAP = Categories of Auditory Performance Index; SIR = Speech Intelligibility Rating; ND = no data

TABLE VII
CASE 4 (AKH) OUTCOMES

Test	Timing (year)		
	Pre-implant	1	2
APT/HI	0%	20%	45%
Spondee words test	0%	75%	90%
CAP	0%	4	4
SIR	1	2	3

APT/HI = Auditory Perception Test for the Hearing-Impaired; CAP = Categories of Auditory Performance Index; SIR = Speech Intelligibility Rating

and SIR scales, respectively (Table VII). AKh had no mental retardation, based on RCPM, and at the time of writing attended a regular school, grade 1, and used his device during all waking hours.

- **Waardenburg syndrome is an autosomal-dominant genetic condition characterized by profound congenital hearing loss and abnormalities of pigmentation**
- **This paper reports the result of cochlear implantation in six children with Waardenburg syndrome, ranging from two to 14 years of age**
- **All the subjects' performance in speech perception and intelligibility improved considerably**

Case 5 (AP)

AP was three years, seven months of age at cochlear implantation. He had been diagnosed with type I WS at seven months, showing dystopia canthorum, heterochromia, broad nasal root and bilateral profound deafness. AP also suffered from minor thalassaemia. The parents had noticed some motor delay: AP could hold his head up at eight months, sit without support at 13 months and walk with one hand held at 20 months. A CT scan showed a normal cochlea. AP was implanted using a Nucleus CI24 device. All 22 electrodes were activated on the map with ACE strategy. One year, 10 months after implantation, AP achieved scores of 78 per cent for the PAPT/HI test, 100 per cent for the Persian Spondee words test, and levels 4 and 3 for the CAP and SIR scales, respectively (Table VIII). Measuring of intelligence by RCPM indicated no mental retardation. At the time of writing, AP was attending a regular preschool and using his device during all waking hours.

TABLE VIII
CASE 5 (AP) OUTCOMES

Test	Timing (year, month)		
	Pre-implant	1	1, 10
APT/HI	0%	35%	78%
Spondee words test	0%	65%	100%
CAP	0	3	4
SIR	1	3	3

APT/HI = Auditory Perception Test for the Hearing-Impaired; CAP = Categories of Auditory Performance Index; SIR = Speech Intelligibility Rating

TABLE IX
CASE 6 (BM) OUTCOMES

Test	Timing (year, month)		
	Pre-implant	1	2, 7
APT/HI	0%	43%	72%
Spondee words test	0%	60%	80%
CAP	0	5	5
SIR	1	2	3

APT/HI = Auditory Perception Test for the Hearing-Impaired; CAP = Categories of Auditory Performance Index; SIR = Speech Intelligibility Rating

Case 6 (BM)

BM was diagnosed with type I WS at seven months, showing dystopia canthorum, pale blue eyes, white forelock and bilateral profound deafness. His parents had noticed no motor delay. BM received no intervention prior to cochlear implantation. A CT scan showed a normal cochlea. BM was implanted using a Med-el Combi40+ device at two years of age. All 12 electrodes were activated on the map with CIS strategy. Two years, seven months after implantation, BM achieved scores of 72 per cent for the PAPT/HI test, 80 per cent for the Persian Spondee words test, and levels 5 and 3 on the CAP and SIR scales, respectively (Table IX). Because of his age, measurement of his intelligence using the RCPM test was not possible, but informal assessment of cognition tasks revealed no significant delay. At the time of writing, BM was attending a regular preschool and using his device during all waking hours.

Discussion

Waardenburg syndrome accounts for between 2 and 5 per cent of cases of congenital deafness.[3] We retrospectively assessed the speech perception, speech production, general intelligence and educational setting of six profoundly hearing-impaired children with WS who had undergone cochlear implantation.

Data from the literature showed that children with WS receive significant benefit from cochlear implantation. In addition to improving auditory skills and speech production, all our cases were transferred to regular educational settings, where they had more chance of developing their communication and social interaction skills. They used their cochlear-implant devices during all waking hours; this implies that they benefitted from the devices. The six patients showed increasing levels of speech perception and speech intelligibility (see Tables VI to IX), and one can assume that they will continue to benefit from the auditory input enabled by their cochlear implants.

Although some studies indicated the presence of mental retardation in some WS children,[8–10] five out of the six patients in our study were of average intelligence range; only one had a borderline intelligence quotient. It may be that mental

retardation is not a very frequent phenotype in deaf children with Waardenburg syndrome.

None of our cases had deformity in the inner ear. Other authors have noted that abnormality of the bony labyrinth in WS with congenital deafness is not a frequent finding, particularly in type-I WS.[6,7]

From the above observations, we recommend that any expansion of cochlear implantation criteria in children include those with Waardenburg syndrome.

References

1 Waardenburg PJ. A new syndrome combining developmental anomalies of the eyelids, eyebrows and nose root with pigmentary defects of the iris and head hair and with congenital deafness. *Am J Hum Genet* 1951;**3**:195–253
2 Read AP, Newton VE. Waardenburg syndrome. *J Med Genet* 1997;**34**:656-65
3 Nayak CS, Isaacson G. Worldwide distribution of Waardenburg syndrome. *Ann Otol Rhinol Laryngol* 2003;**112**:817–20
4 Pardono E, van Bever Y, van den Ende J, Havrenne PC, Lughetti P, Maestrelli SR *et al*. Waardenburg syndrome: clinical differentiation between types I and II. *Am J Med Genet* 2003;**117A**:223–35
5 Reynolds JE, Meyer JM, Landa B. Analysis of variability of clinical manifestations in Waardenburg syndrome. *Am J Med Genet* 1995;**57**:540–7
6 Madden C, Halsted MJ, Hopkin RJ, Choo DI, Benton C, Greinwald JH Jr. Temporal bone abnormalities associated with hearing loss in Waardenburg syndrome. *Laryngoscope* 2003;**113**:2035–41
7 Oysu C, Oysu A, Aslan I, Tinaz M. Temporal bone imaging findings in Waardenburg's syndrome. *Int J Pediatr Otorhinolaryngol* 2001;**58**:215–21
8 Cantani A, Bamonte G, Tacconi ML. Mental retardation and EEG abnormalities in Waardenburg's syndrome: two case reports (EEG anomalies in Waardenburg's syndrome). *Paediatr Pathol* 1989;**24**:137–40
9 Kawabata E, Ohba N, Nakamura A, Izumo S, Osame M. Waardenburg syndrome: a variant with neurological involvement. *Ophthalmic Paediatr Genet* 1987;**8**:165–70
10 de Saxe M, Kromberg JG, Jenkins T. Waardenburg syndrome in South Africa. Part I. An evaluation of the clinical findings in 11 families. *S Afr Med J* 1984;**66**:256–61
11 Rajput K, Brown T, Bamiou DE. Aetiology of hearing loss and other related factors versus language outcome after cochlear implantation in children. *Int J Pediatr Otorhinolaryngol* 2003;**67**:497–504
12 Waltzman SB, Scalchunes V, Cohen NL. Performance of multiply handicapped children using cochlear implants. *Am J Otol* 2000;**21**:329–35
13 Sugii A, Iwaki T, Doi K, Takahashi Y, Yamamoto K, Fuse Y *et al*. Cochlear implant in a young child with Waardenburg syndrome. *Adv Otorhinolaryngol* 2000;**57**:215–9
14 Hassanzadeh S. *Adaptation and Standardization of Persian Auditory Perception Test for Hearing Impaired Children (PAPT/HI)*. Tehran: Ries, 2001
15 Allen MC, Nikolopoulos TP, O'Donoghue GM. Speech intelligibility in children after cochlear implantation. *Am J Otol* 1998;**19**:742–6
16 Archbold S, Lutman ME, Nikolopoulos T. Categories of auditory performance: inter-user reliability. *Br J Audiol* 1998;**32**:7–12
17 Allen MC, Nikolopoulos TP, O'Donoghue GM. Speech intelligibility in children after cochlear implantation. *Am J Otol* 1998;**19**:742–6
18 Raven J, Raven JC, Court JH. The Colored Progressive Matrices. In: Raven J, Raven JC, Court JH. *Manual for Raven's Progressive Matrices and Vocabulary Scales*. Oxford: Oxford Psychologists Press, 1999

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