

Clinical course of four children with Waardenburg Type1 syndrome

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Clinical course of four children with Waardenburg Type1 syndrome

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KEY WORDS

Waardenburg syndrome, hearing impairment, language training, cochlear implant

Introduction

Waardenburg syndrome (WS) is caused by autosomal dominant mutations, and is characterized by pigmentary anomalies and various defects of neural crest derived tissues¹⁾. Many cases of WS also feature lateral displacement of the lacrimal puncta, heterochromia, irides, and white hair which are all classified as WS type1 (WS1)²⁾. One especially important clinical feature of WS1 is congenital hearing loss, which may constitute a severe handicap for children. Patients with the WS but without canthal deformity are categorized as having WStype2 (WS2)³⁾. Most reports on WS consist of a description of single cases or reports on the genetic mutation in these cases, which provide evidence that WS is not a homogenous disease. However, there are no detailed reports on training for WS patients with hearing impairment, nor has a consensus been established regarding such training. Sugii et al.⁴⁾ reported the cochlear implantation clinical course of a patient with WS type 1.

In this study, we shall present the clinical course of four Japanese children's cases with WS type 1 (WS1), two of whom were treated with cochlear implantation. The study presented here deals with the clinical course of the four Japanese children with WS1.

Materials and Methods

1. Subjects

This study used a retrospective examination of the auditory and language development clinical course of four children with severe hearing impairment with WS1 (Table1). The patients were diagnosed with congenital deafness when they were between 1 year and 2 years 5 months old. All of them showed evidence of pigmentary disturbance, dystopia canthorum and severe hearing impairment. Their hearing level was equivalent to a pure tone average of 110 dB or more (Fig.1). They have been using bilateral conventional high power hearing aids and were given language training at a speech clinic in Kanazawa University Hospital. The children were given language training at our clinic using the Kanazawa Method, which involved the presentation of auditory manual, and written language from an early stage^{5) 6)}. None of them showed a very positive attitude toward auditory and lip reading training because they were severely hearing-impaired and had behavioral difficulties that interfered with eye to eye contact with the speech therapist.

Case 1 : A 14-year-old boy who had been deaf from birth and had congenitally blue irises with vision-related complaints, and displacement of the lacrimal puncta. None of his family members had a history of deafness or albinism.

Case 2 : A 7-year-old boy who had been deaf from

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Table1. Clinical findings for WS type 1 subjects enrolled in this study

Case No.	Age/Sex	1 st visit to our clinic (y,m)	Pure tone average hearing level(better ear) (dB)	Average aided level (dB)	Ophthalmological findings
1	14y/M	2y5m	122.5	62.5	dystopia canthorum ocular albinism
2	7y/M	1y6m	133.8	77.5	dystopia canthorum blue iris
3	3y9m/F	1y0m	112.5	60.0	dystopia canthorum blue iris
4	2y10m/F	1y6m	113.8	70.0	dystopia canthorum ocular albinism

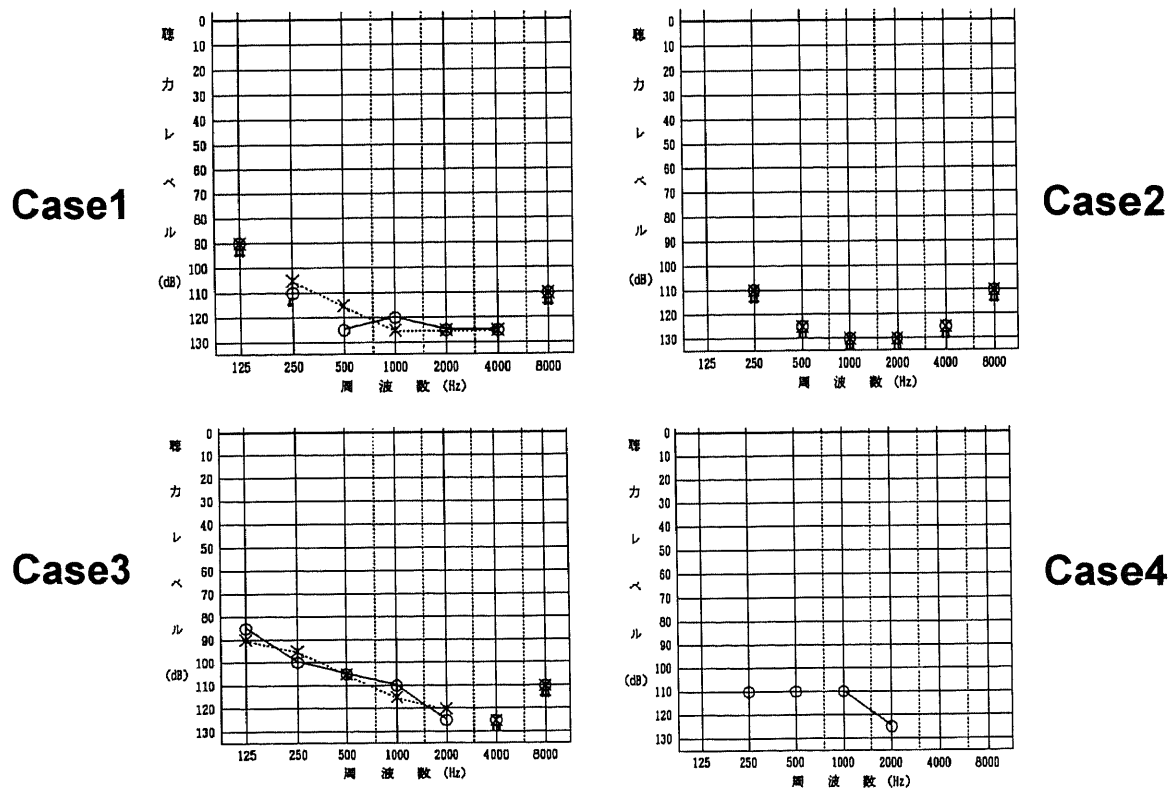


Fig. 1. Pure-tone audiograms of the four subjects

birth and had congenitally blue irises and displaced lacrimal puncta. None of his family members had a history of deafness or albinism. His congenital deafness was accompanied by an abnormally narrow

internal auditory canal.

Case 3 : A 3-year and 9-months old girl who was congenitally deaf and had congenitally blue irises without vision-related complaints, and displaced

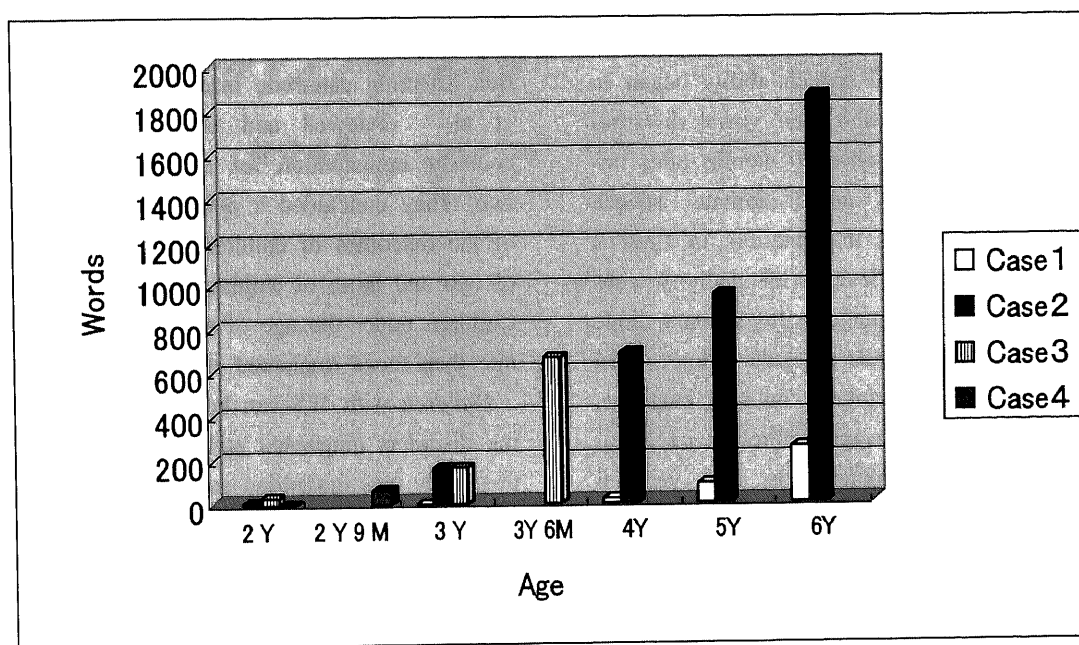


Fig. 2. Accumulation of receptive vocabulary through the oral channel by the four subjects

lacrimal puncta. Her father had a congenitally heterochromia iris and had left suffered from deafness in his left ear as a boy.

Case 4 : A 2-year and 10-month-old congenitally deaf girl. White marks on the bilateral iris were noticed by her parents in early infancy, and the lacrimal puncta was displaced. Her severe congenital deafness was accompanied by a bilateral narrow internal auditory canal. In addition, her motor development was retarded.

2. Language Training (Kanazawa Method)^{5) 6)}

The children were given language training at a clinic in Kanazawa University Hospital, which involved the presentation of auditory, sign, and written language. The children's mothers did all of the teaching at home. Techniques for developing written language, signing, and auditory oral skills were demonstrated for children's mothers once every two weeks. There was also a two-hour group session for the mothers and their children once a week to check on what was being performed at home. The written language training consisted of teaching the children to understand written words and sentences and to express their needs by selecting cards on which a word

was written in Japanese characters (ideogram and /or syllable).

Clinical course

None of the subjects had a very good attitude towards auditory and lip reading training, because they were severely hearing impaired and had behavioral difficulties that interfered with eye-to-eye contact with the speech therapist. Our results demonstrated the influence of severe hearing loss on understanding spoken language through the auditory channel, but the ability to acquire written and sign language through the visual channel. They could learn, both receptively and productively, sign language not only at the word level, but also at the sentence level. As a result of this, they enjoyed a good communication relationship with both their family and speech therapists.

Case 1 had not received a cochlear implant because, when he came to our clinic he was 2 years and 5 months old, so his language training start was comparatively late. Moreover, in his infancy, only a few children in Japan under the school age had been treated with cochlear implantation. However, two of the patients had received cochlear implantation (Nucleus 24; Cochlear Corporation, Lane Cove, NSW,

ustralia) one at the age of 2 years 11 months (Case 1) and the other when he was 4 years 6 months old (Case 2). Their hearing and speech ability began to improve after implantation, and their vowel discrimination score reached 100% about 6 months after implantation, and Case 3 could imitate speech intonation. Two years after implantation, in Case 2 he reached the open-set level with auditory cues orally and now uses oral language as his primary mode of communication. He attends a regular school, even though he depended predominantly on sign language, lip reading, and written language before the implantation. Case 4 also had implantation at 3 years 6 months, and she has started to recognize many environmental sounds and speech sounds.

Discussion

In this report, we described the development course of children with severe hearing impairment with WS. Their hearing level was equivalent to a pure tone average of 110 dB or more, and all of them showed pigmentary disturbance. None of them showed a very positive attitude towards auditory and lip reading training, because they were severely hearing-impaired and had behavioral difficulties that interfered with eye contact with the speech therapist.

Two of our subjects underwent cochlear implantation. Since undergoing implantation, their hearing and speech abilities have begun to progress. Sugii et al. reported a 4-year-old male with WS type 1 who had cochlear implantation. They concluded that a cochlear implant was effective for children with WS who fulfilled the criteria for a cochlear implant. Although Sugii et al. did not describe their language training methods for their patient before implantation in detail, we want to add to their conclusion that the training in sign and written language combined with oral communication has little effect on improvement of the post-operative speech ability of severely hearing-impaired children with WS. Moreover, we believe that cochlear implantation leads to better selection for severely hearing-impaired children with WS.

Lenarz⁷⁾ suggests that cochlear implantation should be performed within the first two years of life. Additionally, only children with an etiology of

meningitis were considered for implantation at such a young age in order to prevent the onset of ossification affecting electrode insertion. Recently, Anderson et al.⁸⁾ reviewed and evaluated the benefits of cochlear implantation for children under the age of two. They concluded a comparative evaluation study of the outcomes of children implanted under the age of two and those of children implanted at a later age. Children under the age of two improved at a quicker rate than those implanted when they were older.

Children with WS can be identified and their hearing disorders diagnosed early in life. One subject in this study was diagnosed as late as 2 years 5 months after birth. We want to emphasize that early diagnosis may have a beneficial effect on severely hearing-impaired subjects with WS.

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ワールデンブルグ症候群タイプ1の4症例の臨床経過

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