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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 tissues[1)]. 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 tl auditory and language development clinical course four children with severe hearing impairment wi WS1 (Table1). The patients were diagnosed wi congenital deafness when they were between 1 ye and 2 years 5 months old. All of them showed ev dence of pigmentary disturbance, dystopia canthoru and severe hearing impairment. Their hearing lev was equivalent to a pure tone average of 110 dB more (Fig.1). They have been using bilateral conve tional high power hearing aids and were giv language training at a speech clinic in Kanaza University Hospital. The children were giv language training at our clinic using the Kanaza Method, which involved the presentation of auditor manual, and written language from an ea stage[5) 6)]. None of them showed a very positi attitude toward auditory and lip reading traini because they were severely hearing-impaired and h behavioral difficulties that interfered with eye to e contact with the speech therapist.

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

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

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

Case No.	Age/Sex	1st 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	dystopoa 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	dystopoa canthorum ocular albinism

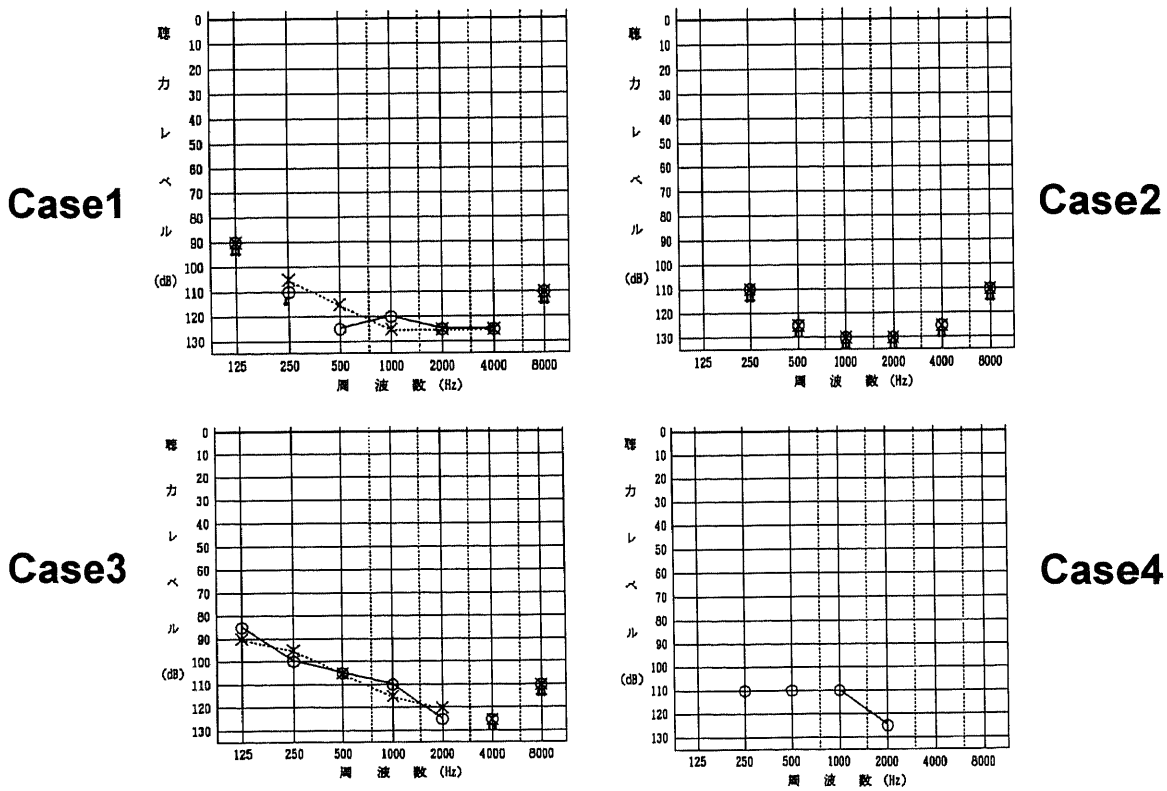

Fig. 1. Pure-tone audiograms of the four subjects

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

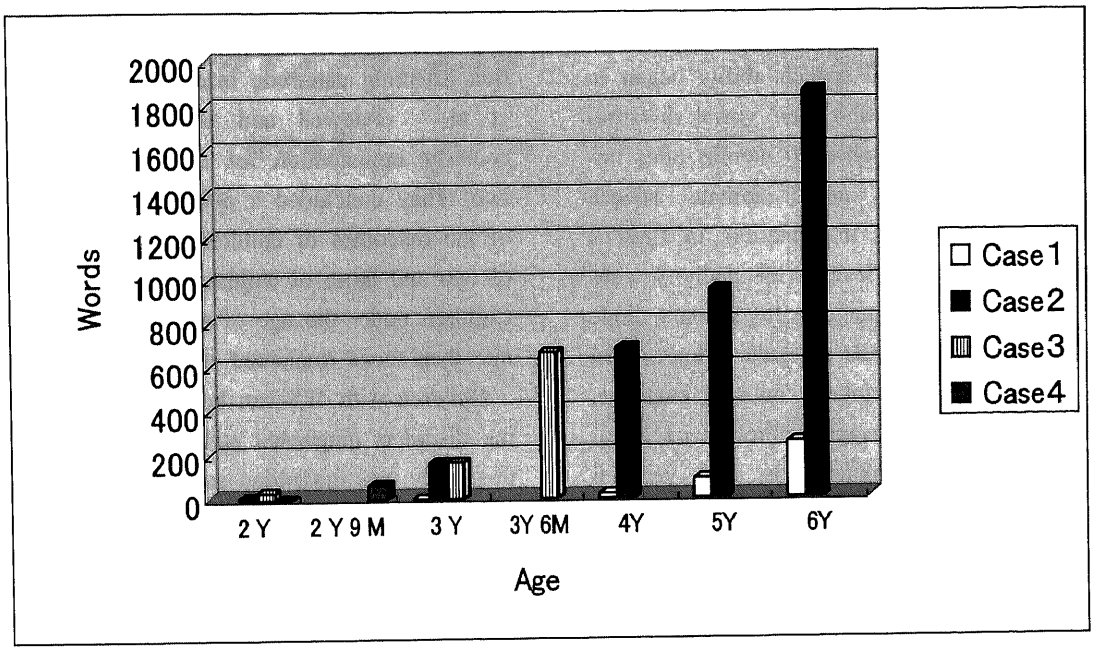

Fig. 2. Accumulation of receptive vocabulary through the oral channel by the four subjects

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,

ıstralia) one at the age of 2 years 11 months (Case and the other when he was 4 years 6 months old 'ase 2). Their hearing and speech ability began to ıprove after implantation, and their vowel discrimiıtion score reached 100% about 6 months after imantation, and Case 3 could imitate speech tonation. Two years after implantation, in Case 2 ıd reached the open-set level with auditory cues ıly and now uses oral language as his primary mode communication. He attends a regular school, even ough he depended predominantly on sign language, ɔ reading, and written language before the implantaɔn. Case 4 also had implantation at 3 years 6 onths, and she has started to recognize many enviınmental sounds and speech sounds.

Discussion

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

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

Lenarz[7] suggests that cochlear implantation should performed within the first two years of life. aditionally, 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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