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Case Report

Auditory and speech-language data in a case of facioscapulohumeral muscular dystrophy in a Japanese child

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ABSTRACT

When he was 5 years old, a boy's father noticed he had difficulty throwing a ball, and at age 7 he was diagnosed with facioscapulohumeral muscular dystrophy (FSHD). When he was a newborn, hearing impairment had been detected via routine screening. Herein, we report follow-up hearing disorder data and speech and language acquisition data. His hearing level was equivalent to a pure tone average of 70 dB or more, and he had been using bilateral conventional hearing aids and had undergone training at a university hospital. Throughout a 6-year follow-up period, his hearing remained almost the same.

1. Introduction

Facioscapulohumeral muscular dystrophy (FSHD) is an autosomal dominant disease of unknown pathogenesis, characterized by weakness of the face and shoulder girdle, and a slow progressive course [1]. Early involvement of the facial and scapular stabilizer muscles results in a distinctive clinical presentation. It is associated with sensorineural hearing loss, which may be subclinical. FSHD has been mapped to the distal most portion of 4q35. Previous reports have shown that bilateral sensorineural hearing loss in the high-frequency range was present [2,3]. Nakagawa et al. reported genetic abnormalities of 71 Japanese individuals with FSHD [4]. In these reports the hearing loss was clearly progressive in some patients, and with time, tended to involve lower frequencies. As hearing disorders cause delays in speech and language acquisition, it is important that hearing impairment is detected early in FSHD patients, and that intervention with hearing aids is initiated.

Herein we present the auditory and language acquisition course in a Japanese boy with FSHD. We undertook several pure tone audiometric examinations and speech and language training sessions, commencing at an early stage. This is the first report of audiometric and speech-language follow-up data during the infantile period in a FSHD patient.

2. Case report

Herein we report the clinical course of auditory and language development in a hearing-impaired Japanese boy with FSHD. The clinical protocol was conducted with the informed consent of patient's parent.

The patient was first seen at the university hospital when he was 1 month old, following a referral based on the results of routine newborn hearing screening. At our hospital, based on objective auditory thresholds he was diagnosed with congenital deafness. The hearing level of the subject was a warble-tone average of 70 dB or more, as determined via visual reinforcement audiometry (VRA). He was subsequently fitted with bilateral conventional hearing aids, and trained in our clinic at Kanazawa University Hospital. He was given language training at the clinic, which involved the

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presentation of auditory, manual, and written language. He did not respond well to the auditory and lip reading training sessions, because he did not like wearing the hearing aids.

The patient developed well during the infantile period. However, difficulty throwing a ball was noted at the age of 5 by his father. Neurological examination at age 5 revealed bilateral facial weakness, tongue atrophy, and weakness of the shoulder girdle, upper arms, and thighs. Gait was assessed as waddling. Subsequently, his walking style gradually began to indicate claudication. At the age of 7 years, he exhibited no dysphagia.

2.1. Genetic testing

At the age of 6 years 8 months, Southern blotting analysis of *Eco*RI-digested genomic DNA using p13E-11 probes detected short 14 kb *Eco*RI fragments. He was diagnosed with not inherent but sporadic FSHD.

2.2. Clinical course

2.2.1. Audiometric follow-up data

VRA was performed by measuring the air condition warble-tone thresholds of both ears at frequencies of 0.25, 0.50, 1.00, 2.00, and 4.00 kHz in 5-dB steps at the age of approximately 3 years. Thereafter, auditory testing was performed by measuring the air condition pure tone thresholds of each ear at frequencies of 0.25, 0.50, 1.00, 2.00, and 4.00 kHz in 5-dB steps (Fig. 1). In speech audiometry testing, his maximum speech recognition score was 63% in the left ear, and 50% in the right ear.

2.2.2. Speech and language development

The patient did not perform well in auditory and lip reading training sessions. He used oral language as his primary mode of communication, and attended a regular school where he received special care. At the age of 5 years 8 months, verbal IQ (VIQ) and performance IQ (PIQ) were evaluated via the Wechsler Preschool and Primary Scale of Intelligence (WPPSI) (Fig. 2). His mental score was in the subnormal range. His auditory comprehension level as determined by the Picture Vocabulary Test (PVT) was in the range of age 4 years 3 months when he was aged 6 years. Auditory/oral receptive vocabulary and spontaneous words from 1 year of age to 6 years of age are shown in Fig. 3. His speech intelligibility was suboptimal, but not "poor" (2) with regard to the 5 levels of the test. In his unintelligible speech, "p", "b", "m", and "s" sounds were relatively weak. It is known that weakness of articulating "s" sounds is associated with impairment of hearing high frequencies [5]. However, such hearing impaired patients do not routinely exhibit errors or omission of "p", "m", and "b" sounds. It may be that in the case reported herein, labial stop consonant sounds were unclear due to weakness of the facial muscles.

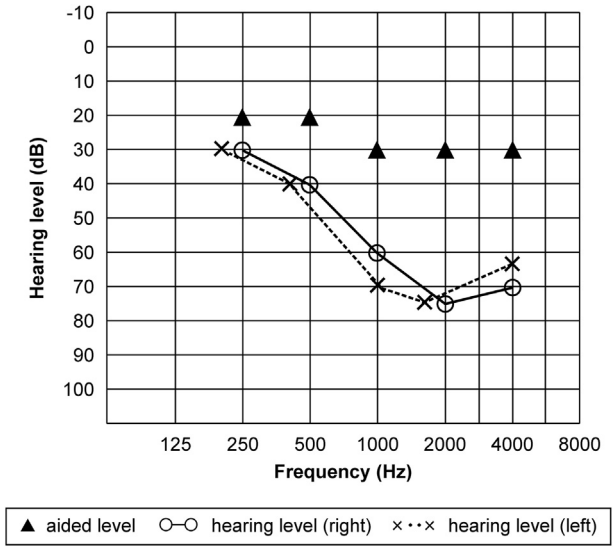

Fig. 1. Audiogram at 3 years old.

3. Discussion

FSHD is an autosomal dominant progressive myopathy, characteristically associated with a 4q35 deletion [2]. In the unusual infantile-onset form of this degenerative disease, sensorineural hearing loss is a frequent clinical manifestation [2]. Herein we have reported the details of a young Japanese boy with sporadic FSHD, attributed to a sporadic 4q35 deletion. In this case study, we recorded his audiometric, speech, and language data from the ages of 0–6 years. Although his hearing impairment was discovered when he was a newborn via standard screening, hearing aids were of limited benefit in this patient as he did not like wearing them, resulting in suboptimal compliance. His language acquisition level was retarded at the age of 6 years, with a VIQ of 53 as determined via the WPPSI. His PIQ as determined via the WPPSI was in the subnormal range. He did not exhibit obvious mental retardation. His hearing level exhibited almost no change by the age of approximately 7 years, which was concordant with the fact that he had been diagnosed with high-frequency hearing impairment at an early age (Fig. 4).

In a study investigating early onset FSHD patients, Nakagawa et al. [2] reported 42 patients (30 familial, 12 sporadic) suspected of having FSHD. Early onset FSHD was detected in 7 patients, tortuosity of retinal arterioles and hearing impairment was detected in 3, progressive respiratory failure was detected in 3, and limb-girdle type muscular weakness was detected in 6. However, hearing follow-up data were not reported in detail. In the case reported herein, retinal arteriole involvement was not detected. Miura et al. [3] reported 2 cases of early onset FSHD with mental retardation and epilepsy. Both cases were sporadic FSHD. One case exhibited infantile spasm at the age of 4 months, muscular atrophy in the face, and shoulder girdle and upper limbs involvement were observed from the age of 4 years. In the other case, lack of facial expression was observed from the age of 1 year, and at the age of 10 years weakness of the lower limbs and moderate hearing loss were apparent. Their sensorineural IQs were 33 and 45 respectively, but hearing-follow up data were not reported. The fact that the case reported herein did not exhibit obvious mental retardation may be associated with the lack of observed infantile spasm. Takeya et al. [6] reported two cases of lesions associated with hearing loss in FSHD patients at the cochlear level. One was a girl aged 5 years, and the other was a boy aged 15 years. In the first case, retarded-language development from the age of 2 years was observed, and pure tone audiograms revealed high tone hearing loss without an A-B gap. However, her maximum speech recognition score was 100%, presumably because her high tone hearing loss was reportedly mild. In the case reported herein, there was moderate hearing loss from an early age and this may account for his compromised maximum speech recognition score.

Trevisan et al. [7] reported that FSHD patients with unusual large 4q35 deletions tended to exhibit atypical features in early childhood. Sensorineural hearing loss was found in 4 patients who exhibited an infantile-onset dystrophic phenotype. Brouwer et al. [8] reported audiometry screening results from 56 patients with autosomal dominant FSHD and 72 healthy family members, and

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