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Case report

A case of improved hearing with cochlear implantation in Gaucher disease type 1

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ABSTRACT

Gaucher disease is a lysosomal storage disorder that is caused by congenital defective function of the enzyme glucocerebrosidase. Glucocerebroside that is not hydrolyzed by glucocerebrosidase mainly accumulates in the reticular tissue. We describe a Japanese boy with Gaucher disease type 1 who developed bilateral profound sensorineural hearing loss within approximately 4 years. We performed cochlear implantation initially on his right ear and again on his left ear 5 months later. The cochlear implants were successfully utilized with a speech discrimination score of 95% on a Japanese sentence recognition test. There are many reports of central hearing loss in Gaucher disease type 2 or 3. However, to the best of our knowledge, this is the first report of profound inner ear hearing loss with Gaucher disease. It also appears to be the first record of cochlear implantation for Gaucher disease. Cochlear implants may be useful for sensorineural hearing loss in patients with Gaucher disease without neurological symptoms other than hearing loss.

1. Introduction

It has been estimated that Gaucher disease occurs in approximately one birth out of 57,000–75,000 globally [1,2] and in one out of 330,000 in Japan [3]. Although it is relatively rare, it is considered the most common lysosomal disease. It is an autosomal recessive genetic disease caused by a mutation in the gene encoding glucocerebrosidase located on chromosome 1 [4]. Glucocerebroside that is not hydrolyzed by glucocerebrosidase accumulates in various parts of the body, mainly within the reticular tissue. The symptoms of Gaucher disease, including hepatosplenomegaly, bone fracture, anemia, and thrombocytopenia, are caused by glucocerebrosidase accumulation in the liver, spleen, and bone marrow.

Gaucher disease is classified into three types on the basis of neurological manifestations and age of onset. Type 1, the most common, presents without neurological manifestations. Types 2 and 3 are associated with neurological manifestations such as convulsion, trismus, and strabismus; type 2 is more severe and acute than type 3. Herein, we describe the first case, to the best of our knowledge, of a patient with Gaucher disease type 1 who developed bilateral profound inner ear hearing loss and received bilateral cochlear implantation.

2. Case report

The patient was a Japanese boy aged 15 years and 11 months. After his birth, no delay in growth development was indicated.

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Hepatosplenomegaly, anemia, and thrombocytopenia were noted by a pediatrician when he was hospitalized with respiratory infection (pertussis) at 8 months of age. When he was 14 months old, blood biochemical examination showed an increased angiotensin-converting enzyme level at 70.6 U/L (normal value 0–14.4 U/L) and an increased acid-phosphatase level at 163.2 U/L (normal value 8.3–21.4 U/L). The activity of β-glucosidase from cultured skin fibroblasts was only 6% of the normal activity level, and bone marrow aspiration demonstrated the presence of Gaucher cells.

Since neurological symptoms such as epilepsy, cognitive impairment, and ataxia were not observed, the patient was diagnosed with Gaucher disease type 1. There was no family history of Gaucher disease, but his mother had Parkinson's disease. Enzyme replacement therapy was administered once every 2 weeks from the age of 1 year and 3 months to the present. For enzyme replacement therapy, imiglucerase was initially administered, followed by velaglucerase alfa from the age of 11 years and 2 months. Approximately 1 year after the initiation of enzyme replacement therapy, symptoms such as hepatosplenomegaly, thrombocytopenia, and anemia improved.

The patient was aware of slight hearing loss at the age of 9 years and 2 months, and he visited the otolaryngology department at the age of 9 years and 7 months. His hearing levels at this time were 30.0 dB (right) and 40.0 dB (left) (Fig. 1a), the average in decibels of the thresholds for pure tones at 500, 1000, and 2000 Hz. He began to use bilateral hearing aids from the age of 10 years (between Fig. 1b and c). Subsequently, his sensorineural hearing loss progressed (Fig. 1 d), and he developed profound hearing loss (right 95.0 dB, left 98.3 dB) at the age of 13 years (Fig. 1e).

It became difficult for the patient to attend junior high school because it was impossible to communicate by voice with his friends and teachers. He visited our hospital at the age of 13 years and 1 month. At the first visit, his bilateral tympanic membrane findings were normal. We recorded auditory steady-state response twice, but no response was obtained, even at 105 dB, on both sides. We did not record auditory brainstem responses because it was speculated that response could not be obtained from the results of auditory steady-state response and audiograms. Even with the presentation sounds of 100 dB hearing level, the speech discrimination score was 0% on a Japanese sentence recognition test. There were no inner or middle ear malformations in temporal bone computed tomography. Head magnetic resonance imaging did not indicate any abnormal findings such as brain atrophy or inner ear malformations (Fig. 2).

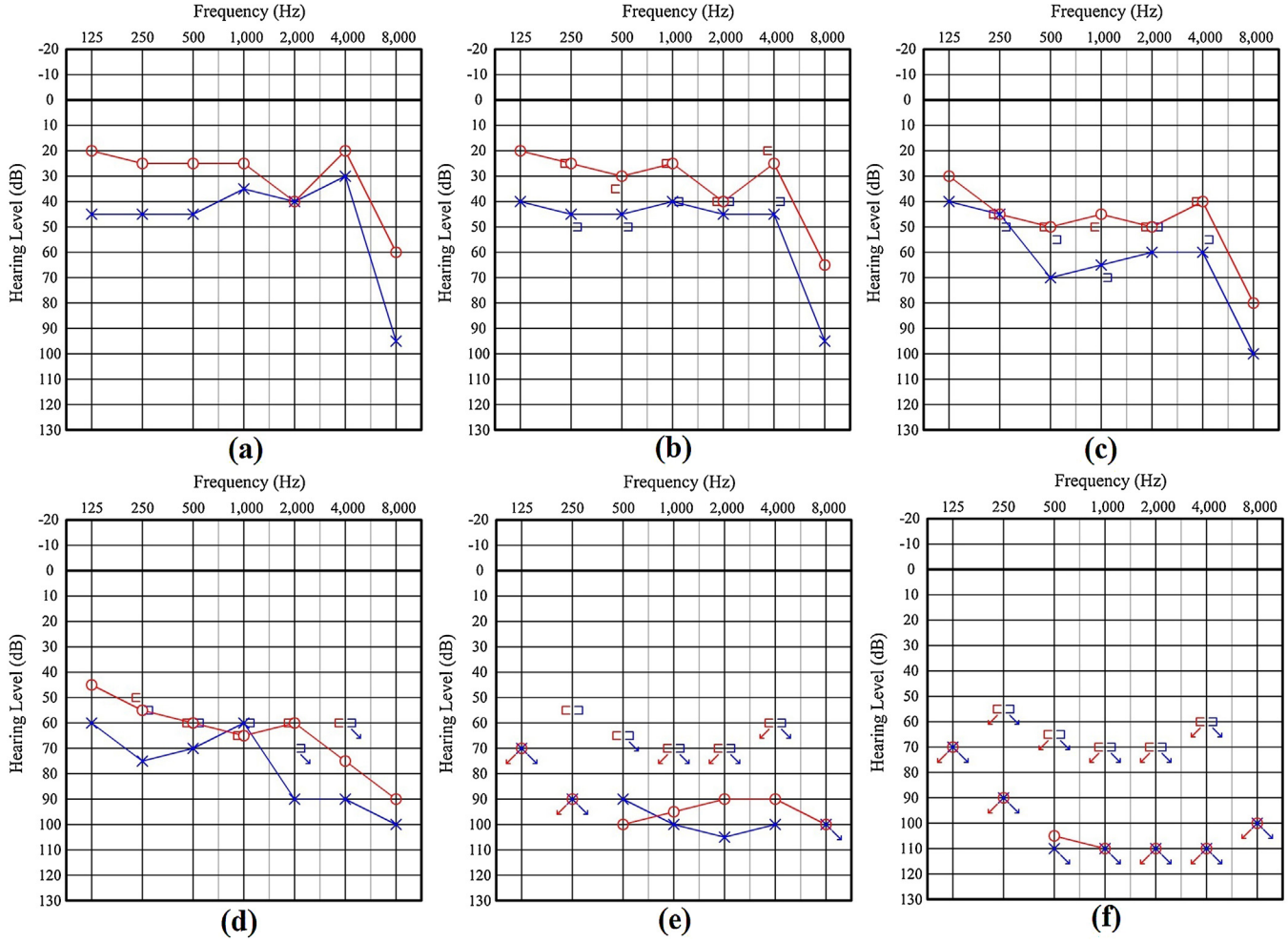

Fig. 1. Progress of patient's hearing level. (a): At 9 years and 7 months of age. (b): At 10 years and 1 month of age. (c): At 11 years of age. (d): At 12 years and 1 month of age. (e): At 13 years of age. (f): At 13 years and 2 months of age.

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