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Cochlear implantation in a patient with Epstein syndrome

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ABSTRACT

Epstein syndrome is a rare disease which is accompanied by nephritis, sensorineural hearing impairment and macrothrombocytopenia. It has been suggested that this syndrome is a hereditary disease associated with mutations in MYH9, which encodes non-muscle myosin heavy chain IIA. We report a case of a patient with Epstein syndrome in whom bilateral profound hearing impairment developed and who had undergone cochlear implantation 9 years previously. Prior to this, the patient showed progressive sensorineural hearing impairment and had become completely deaf by the age of 25. A cochlear implant was successfully used with a speech discrimination score of 98% (sentence test). However, in the present case, peri- and postoperative complications occurred: tympanic perforation remained after a promontory stimulation test, followed by transitory otitis with purulent discharge. Therefore, tympanoplasty was performed simultaneously with cochlear implantation. These complications were considered to be caused by platelet dysfunction and delayed wound healing. Furthermore, cochlear destruction was observed 8 years postoperatively. In Epstein syndrome, the mechanism of osseous change remains uncertain. To the best of our knowledge, this is the first case report of Epstein syndrome in a patient with long-term use of a CI.

1. Introduction

Epstein syndrome is a rare disease which is accompanied by nephritis, sensorineural hearing impairment and macrothrombocytopenia. It has been suggested that this syndrome is an autosomal hereditary disease associated with *MYH9* mutation, which is commonly seen in several types of macrothrombocytopenia such as May-Hegglin anomaly, Sebastian syndrome and Fechtner syndrome. Among these *MYH9*-related disorders, the presence of clinical symptoms varies according to the location of the mutation. Sensorineural hearing loss is one of the symptoms characteristic of Epstein syndrome. Hearing impairment can progress to complete deafness. We report a case of a patient with Epstein syndrome in whom bilateral profound hearing impairment developed and who had received cochlear implants (CIs) 9 years previously.

2. Case

The patient was first referred to our hospital at the age of 5 with epistaxis during investigation of idiopathic thrombocytopenic purpura. Her platelet count was 45,000/μl at that time, and 2 years later sensorineural hearing impairment was observed, although her hearing loss was mild and her recruitment phenomenon was positive. However, her hearing impairment worsened, and she was given conventional hearing aids. She had become completely deaf in her right ear by the age of 23 and in her left ear by the age of 25 (Fig. 1). During that time, she received a diagnosis of Epstein syndrome on the basis of pathological findings of a renal biopsy specimen, and of clinical symptoms such as hematuria, proteinuria, macrothrombocytopenia and sensorineural hearing loss diagnosed by pediatricians at another hospital at age 15.

Cochlear implantation was then considered owing to the limitations of hearing aids. Her computed tomography (CT) and magnetic resonance imaging findings were normal, and she showed sensitivity to a promontory stimulation test. However, a pinhole-sized perforation remained on the left tympanic membrane after the promontory stimulation test. Her platelet count decreased to 8000/μl and immunoglobulin was administered in an attempt to improve her blood dyscrasia, but it was ineffective. She had become antiplatelet antibody-positive owing to a previous platelet transfusion upon previously undergoing resection for an open cyst with endometriosis. Therefore, a human leukocyte antigen (HLA)-matched platelet transfusion was performed to prevent bleeding, and tranexamic acid was given. Her platelet count then increased to 67,000/μl. A Nucleus 24 (Cochlear Ltd., Lane Cove, Australia) CI was implanted in her left ear and myringoplasty was performed. Although her platelet count decreased to 37,000/μl after 3 days and to 14,000/μl after 11 days, perioperative bleeding was not observed. Furthermore,

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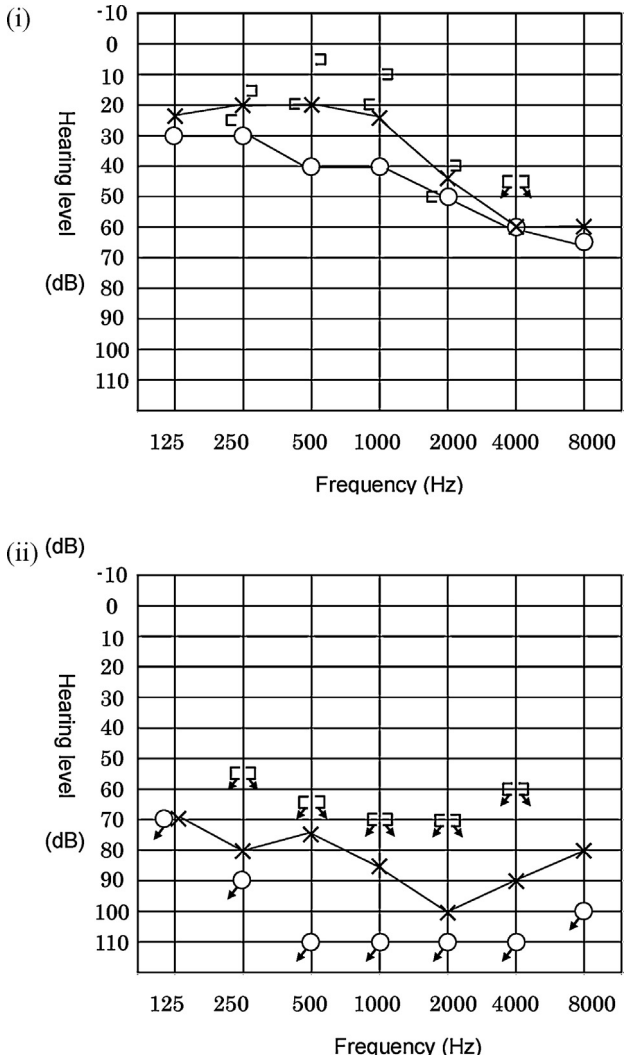

Fig. 1. Hearing levels at (i) 7 years old; (ii) 23 years old.

although the wound had healed by 4 months postoperatively, a perforation remained on the operated tympanic membrane. In addition, methicillin-resistant *Staphylococcus aureus* and a fungal infection developed and continued for 1 year (Fig. 2). However, despite the infection, the CI remained functional and her speech discrimination score showed improvement of up to 98% on a Japanese sentence recognition test. Her hearing threshold was stable between 30 and 35 dB with CIs.

However, 7 years postoperatively, her word recognition score decreased to 76% and her MAP-threshold level (T-level) and maximum comfort level (C-level) were observed to fluctuate. Subsequently, electrode extrusion was confirmed by X-ray and CT imaging (Fig. 3), and the 2 electrodes at the apical end of the cochlea were switched off. Her speech perception was 80% (words) and 98% (sentences) at 9.3 years postoperatively.

In addition to these changes in her left ear, a perforation in the right tympanic membrane was observed 2 months postoperatively, but there was no evidence of infection in the right ear. Furthermore, cerumen accumulations were consistently observed, which were considered to be the cause of her enlarged external ear canal.

3. Discussion

Epstein syndrome was first reported in 1972 [1] and it was subsequently discovered that the disease is associated with mutations in *MYH9*, which encodes non-muscle myosin heavy chain IIA (NMMHC IIA) [2] [3]. NMMHC IIA is a type of non-muscle myosin that is distributed in many types of tissue [4]. These non-muscle myosin molecules contribute to maintaining the cytoskeleton and regulating cell adhesion, cell migration and cell division [5]. There are nearly 40 reported mutations in NMMHC IIA, some of which are considered to be associated with *MYH9*-related diseases. Epstein syndrome is one such *MYH9*-related disease, which is associated with mutations in exon 16. Hearing impairment is considered to be sensorineural because NMMHC IIA is present in the inner ear [4]. In the present case, sensorineural hearing impairment was prominent only in the high frequencies in the early stages, but progressed to bilateral severe hearing loss in all frequencies. The clinical course of the current case was consistent with Epstein syndrome [6]. However, the use of a CI was effective for the deafness due to Epstein syndrome in the present case. To the best of our knowledge, this is the first report on the long-term follow up of a CI in a patient with Epstein syndrome.

It has been reported that Epstein syndrome can be misdiagnosed as chronic autoimmune thrombocytopenia. It can be treated by splenectomy, immunosuppressive therapy and corticosteroid hormone therapy, but these treatments are presently considered to be ineffective in *MYH-9*-related diseases [6]. In the present case, Epstein syndrome was diagnosed on the basis of the clinical symptoms and findings of a renal biopsy specimen, but only after a previous period during which *MYH-9*-related diagnosis was suspected.

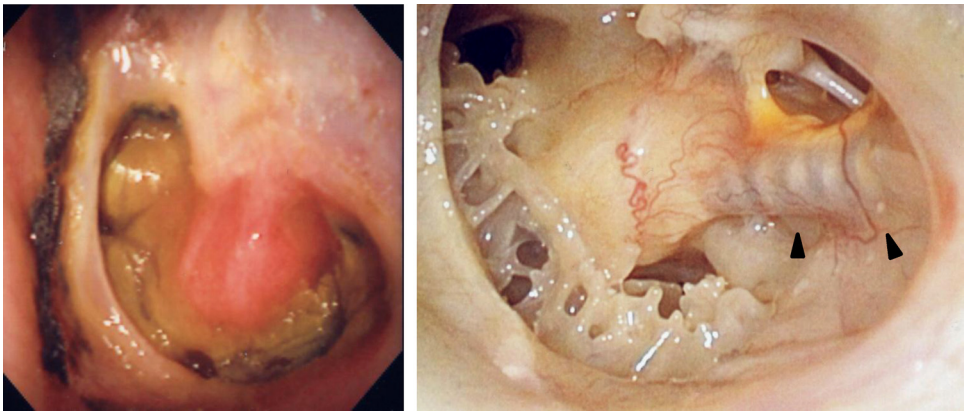

Fig. 2. Postoperative tympanic membrane. Yellowish otorrhoea can be observed in the left middle ear cavity. The electrode array, covered with fibrous tissue (arrowheads) was placed into the cochleostomy site between the round window niche and the stapedial muscle.