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CLINICAL RECORD

Successful bone-anchored hearing aid implantation in a patient with osteogenesis imperfecta

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Abstract

Objective: To report a case of successful bone-anchored hearing aid implantation in an adult patient with type III osteogenesis imperfecta, which is commonly regarded as a contraindication to this procedure.

Case report: A 45-year-old man with type III osteogenesis imperfecta presented with mixed hearing loss. There was a mild sensorineural component in both ears, with an air–bone gap between 45 and 50 dB HL. He was implanted with a bone-anchored hearing aid. The audiological outcome was good, with no complications and good implant stability (as measured by resonance frequency analysis).

Conclusion: To our knowledge, this is the first recorded case of bone-anchored hearing aid implantation in a patient with osteogenesis imperfecta.

Key words: Osteogenesis Imperfecta; Hearing Loss; Osseointegration; Hearing Aids

Introduction

Osteogenesis imperfecta is a genetic disorder of the connective tissue. It is most often caused by mutations in the *COL1A1* and *COL1A2* genes, which are responsible for the synthesis of type I collagen, the major structural protein of the body.[1–4]

The most used classification of osteogenesis imperfecta, based on clinical and genetic features, distinguishes four major types: mild (type I), lethal (type II), severe (type III) and moderate (type IV).[5] Up to 50 per cent of osteogenesis imperfecta patients present with hearing loss, regardless of the osteogenesis imperfecta type. The auditory impairment usually presents as conductive hearing loss in the second to fourth decade of life, and often evolves to mixed or even sensorineural hearing loss later in life.[2,3,6] Nowadays, the main treatment options for the hearing loss in osteogenesis imperfecta patients are hearing aids, stapes surgery and cochlear implants.[7,8]

Case report

This report concerns a 45-year-old man with type III osteogenesis imperfecta. He presented with a history of multiple progressive deforming fractures in childhood, deformed limbs, scoliosis, blue sclera, dentinogenesis imperfecta and hearing loss. He first contacted our otolaryngological department for audiological advice.

He had experienced hearing loss since late in the third decade of life and was fitted with a conventional skin-drive bone-conduction device connected to spectacle frames, which he had been using for five years. He complained of discomfort using this device and suffered frequent skin irritation with the pressure needed for sound conduction. The patient denied any vertigo or tinnitus.

We conducted medical and radiological evaluations with a computed tomography (CT) scan of the ears and skull and audiological testing. Clinical examination revealed normal ears. Pure tone audiometry (Figure 1) revealed mixed hearing loss and a mild sensorineural component in both ears, with an air–bone gap between 45 and 50 dB HL. The CT scan showed varying degrees of demineralisation of the temporal bone, with extensive demineralisation of the posterior region of the inner ear and the surroundings of the cochlea and internal auditory canal. Fainting of the edge definition of the various structures of the inner ear was also observed, producing an image resembling the fourth turn of the cochlea. Obliteration of the oval window was also evident, with hypodensities in the fissula ante fenestram and demineralisation of the ossicles (Figure 2).

The patient refused exploratory tympanotomy. He underwent a test period with a bone-anchored hearing aid (BAHA) softband. Despite knowing that osteogenesis imperfecta is a relative contraindication for BAHA surgery, he decided to go ahead with this surgery.

After a period of three years, during which time the patient's hearing remained stable, the surgery was performed in a single-stage procedure and he was fitted with the Cochlear™ BAHA BI300 implant.

Immediately after surgery, an implant stability measurement was made using an Osstell osseointegration monitoring

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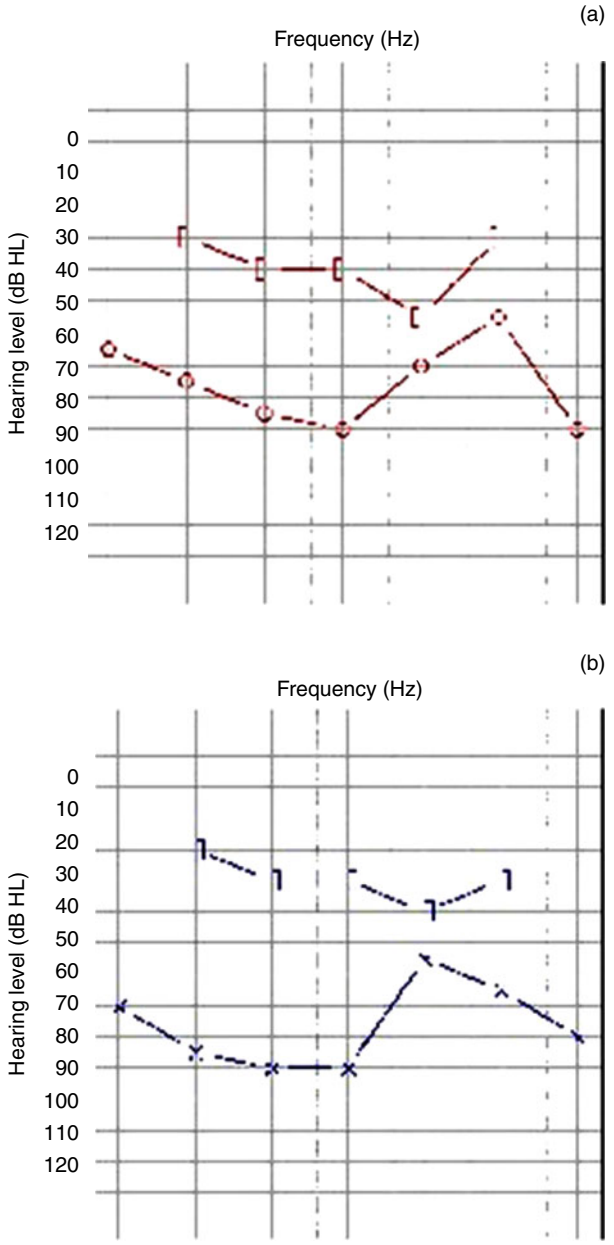

FIG. 1

Pure tone audiogram of the (a) right and (b) left ears, showing bilateral mixed hearing loss. [= Bone conduction (masked), right ear; ○ = air conduction (unmasked), right ear;] = bone conduction (masked), left ear; × = air conduction (unmasked), left ear

device (Integration Diagnostic, Göteborg, Sweden). The implant stability quotient was measured regularly for up to 14 months post-implantation, as shown in Figure 3.

The sound processor was fitted five months after surgery. Since that time, the sound processor has been used on a daily basis, without complications and with very good audiological results (Figure 4).

Discussion

Hearing loss is present in about half of the patients suffering from osteogenesis imperfecta, irrespective of the underlying genotype or phenotype.[3,8] Most osteogenesis imperfecta patients initially develop bilateral conductive hearing loss during the second to fourth decade of life. This often

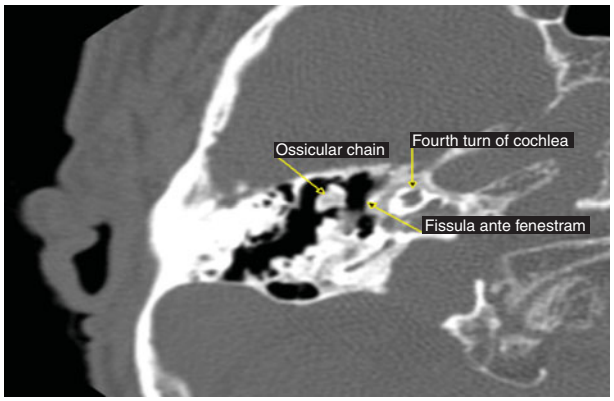

FIG. 2

Axial computed tomography image of the right ear showing a focus of hypodense bone in the fissula ante fenestram, a thickened staples footplate, hypodense ossicular chain and retrofenestral hypodensities resembling the fourth turn of the cochlea.

evolves to a mixed hearing loss because of the gradual onset of a sensorineural component. In a few cases, the loss is only sensorineural. Conductive hearing loss is usually related to stapes footplate fixation, although fractures or atrophy of the stapes superstructures, ossicular discontinuity, or hypervascularised mucosa have been reported.[9] Because the hearing loss is often attributed to an otosclerosis-like pathology, stapedectomy is the treatment of choice in most patients, but the results of this surgery are significantly worse than those described in non-osteogenesis imperfecta series reports.[8,10–13]

In most series of stapedectomies in osteogenesis imperfecta patients, cases of type I disease are dominant. In 2007, Hultcrantz and Sääf published a study of 16 adult patients with osteogenesis imperfecta, in which 81 per cent had osteogenesis imperfecta type I and 19 per cent had osteogenesis imperfecta type IV.[10] In 2014, Vincent *et al*. reported 32 cases, all of which presented with type I osteogenesis imperfecta.[11]

This article reports a case of type III osteogenesis imperfecta, the rarest form of the disease except for type II (lethal), being less frequent than type I and type IV. In a study by Swinnen *et al*., of 184 patients, 83.7 per cent were diagnosed with type I osteogenesis imperfecta, 14.1 per cent had type IV disease and only 2.2 per cent had type III osteogenesis imperfecta.[3] Several other studies found similar results, including Paterson *et al*.,[14] Kuurila *et al*.[6] and Pillion *et al*.[8] As such, there are very few reports of hearing loss management in patients with type III osteogenesis imperfecta, and, as seen in previous studies, stapes surgery is almost always performed in patients with type I osteogenesis imperfecta.[10,11]

Management of hearing loss in osteogenesis imperfecta cases must be determined based on categories according to hearing loss type (conductive, mixed or sensorineural) and its severity.[7,8] Our patient had mixed hearing loss, with a mild sensorineural component in both ears and an air–bone gap of 45–50 dB HL. The treatment options in this case included a hearing aid, stapes surgery or a BAHA. The patient refused stapes surgery and adapted poorly to the hearing aid. He instead opted to undergo BAHA surgery, despite knowing the risks of poor osseointegration associated with his disease. During the five years of hearing aid use and

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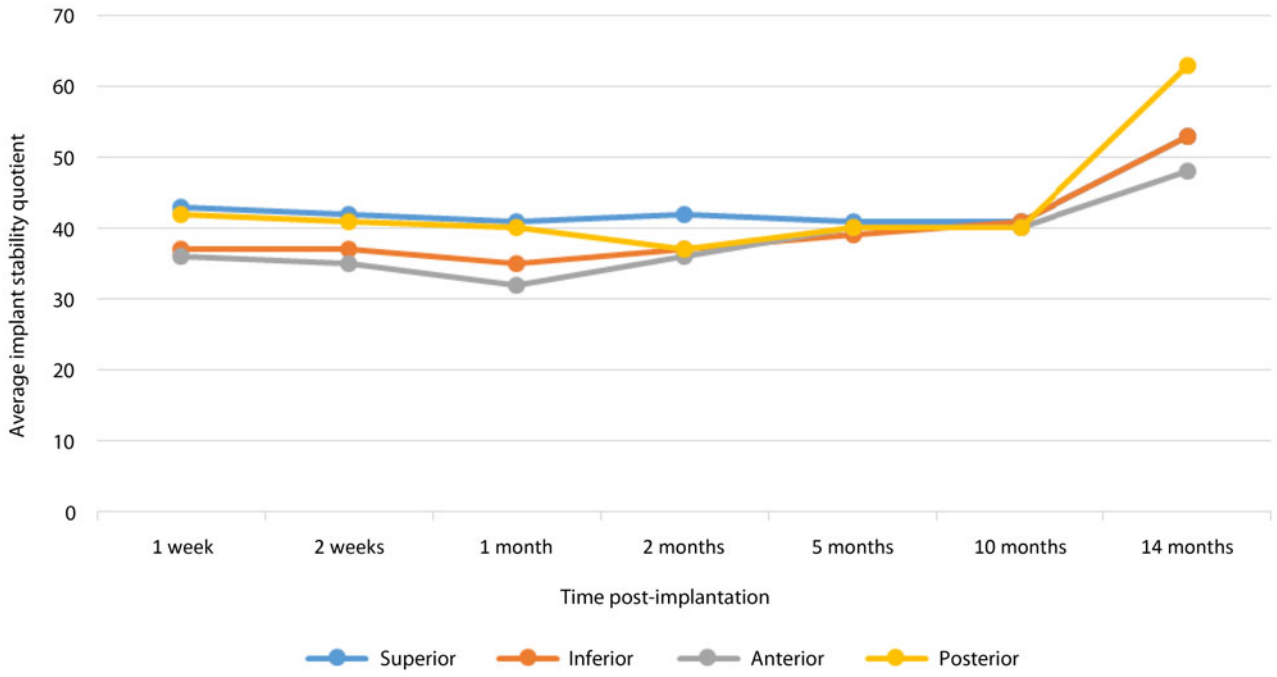

FIG. 3

Average implant stability quotient values for each visit, as measured by resonance frequency analysis in four cardinal points of the abutment.

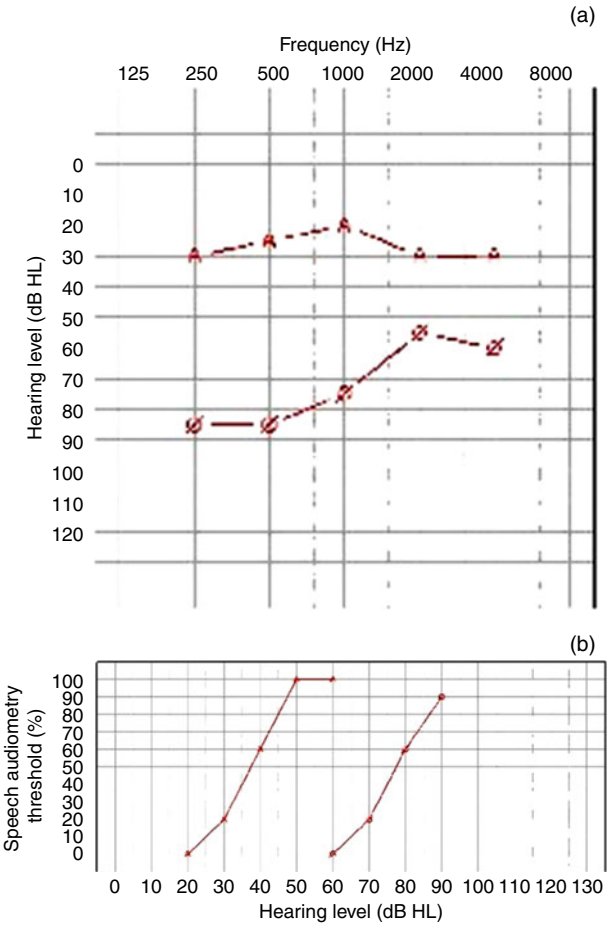

FIG. 4

Free-field audiometry (a) and speech audiometry (b) at 14 months, showing unaided ('Ø') and aided ('A') (with a bone-anchored hearing aid) thresholds.

the three years before surgery, his hearing thresholds remained stable, with no deterioration of the sensorineural component. It is nevertheless postulated that hearing loss in osteogenesis imperfecta (type I) cases is age-related, with an estimated annual increase of 1–1.7 dB.[15]

- **Osteogenesis imperfecta is a genetic disorder of connective tissue, most often caused by *COL1A1* and *COL1A2* gene mutations**
- **Up to 50 per cent of osteogenesis imperfecta patients present with hearing loss regardless of disease type**
- **This paper reports a type III osteogenesis imperfecta case with mixed hearing loss, with a mild sensorineural component bilaterally and a 45–50 dB HL air–bone gap**
- **The patient was fitted with a bone-anchored hearing aid (BAHA), with good results and no complications**
- **This appears to be the first report of an osteogenesis imperfecta patient receiving a BAHA implant**
- **In selected osteogenesis imperfecta cases, osseointegrated implants should be considered for hearing loss**

Osteogenesis imperfecta is a relative contraindication for BAHA surgery because of poor bone mineral density. However, in a thorough systematic review on dental implants, Marquezan *et al*. reported evidence to suggest that the relationship between bone density and implant primary stability is very weak.[16] Again in 2011, Yoon *et al*. reported that bone quality and surgical technique have an influence on implant primary stability, and

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resonance frequency measured with an Osstell osseointegration monitoring device has a positive relation with the density of bone surrounding the implant fixture.[17]

In our case, the implant stability quotient was measured at each visit after surgery, at the same four points of the abutment (Figure 3). This revealed steady results for the first 10 months, with an overall increase in stability at 14 months after surgery, although the absolute values were still slightly below those reported in other studies or in children or adults with normal bone density.[18,19]

There are some features in this case that encouraged us to opt for rehabilitation with a BAHA. Specifically, these included: the audiological thresholds of the patient within the fitting range of this device, the stability of these thresholds over the years, the patient's refusal of stapes surgery, poor adaptation to conventional hearing aids and the patient's strong desire to undergo BAHA implantation. The paucity of reports concerning the management of hearing loss in type III osteogenesis imperfecta cases was also relevant to this patient's motivation for this innovative approach.

To our knowledge, this is the first case report of a patient with osteogenesis imperfecta receiving a BAHA implant. In selected osteogenesis imperfecta cases, osseointegrated implants should be considered in the management of hearing loss.

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