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Main Articles

Stapedotomy in osteogenesis imperfecta patients

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Abstract
Osteogenesis imperfecta (OI) is a connective tissue disorder characterized by osseous fragility, blue sclerae and hearing loss. In order to assess the impact of stapedotomy on improving hearing on OI, a retrospective, one-group, pre-test-post-test design was used to compare the pre-operative and post-operative audiograms of nine OI patients, treated with stapedotomy for their mixed hearing loss. Operative findings included fixation or thickening of the stapes footplate with normal superstructure configuration and hypervascularization of the promontory mucosa. Immediate post-operative results showed a significant improvement ($p<0.05$) from 250–4000 Hz in air conduction and from 250–2000 Hz in bone conduction. A significant closure of the air-bone gap between 250–2000 Hz was also achieved ($p<0.05$). The long-term results remained satisfactory with a mean threshold shift of 8 dB HL and an almost unchanged air-bone gap. These satisfactory results and the lack of complications make stapedotomy an appealing method for the management of OI-associated hearing loss.

Key words: Osteogenesis imperfecta; Hearing loss; Surgery, operative

Introduction

Osteogenesis imperfecta (OI) is a rare, generalized connective tissue disorder involving bones, ligaments, tendons, fascia, skin, sclerae, blood vessels and hearing.[1] A classification of OI into four types (I–IV) has been proposed by Sillence *et al.*,[2] covering a wide range of phenotypes, varying from intrauterine or perinatally lethal forms to a hardly noticeable increase in the tendency for bone fractures (Table I). When the phenotype allows recognition, OI is mostly inherited as an autosomal dominant trait with variable expression, although autosomal recessive forms also exist.[3] OI results from mutations in two genes, the COL IA1 and the COL IA2 located on chromosomes 17 and 7, respectively, encoding the a1 and a2-chains of collagen type I.[4]

OI may involve virtually any organ system. The hallmark of this disorder is osseous fragility and blue sclerae.[1] There is also a tendency to hernias and in rare cases, heart disease due to valvular insufficiency. In addition, immature connective tissue in teeth, skin, and blood vessels predisposes to faulty carious dentition, inappropriate scarring, and easy bruising.

TABLE I
CLINICAL CLASSIFICATION OF OSTEOGENESIS IMPERFECTA BY SILLENCE[2]

Type IA	Blue sclerae Spontaneous fractures Presenile hearing loss in a large proportion Normal teeth Autosomal dominant inheritance
Type IB	As type IA, with defective dentition
Type II	Extreme fragile skeleton and therefore lethal perinatal OI Autosomal recessive inheritance
Type III	Mostly normal sclerae After birth progressive deformity of limbs and spine with many fractures, small length, kyphoscoliosis, asymmetric skull, triangle facial appearance Sometimes hearing loss Sometimes defective dentition Autosomal recessive or dominant
Type IV A	Normal sclerae Fragile bones Sometimes growth retardation and deformity Sometimes hearing loss Normal teeth Autosomal dominant inheritance
Type IV B	As type IV A, with defective dentition

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Hearing loss usually appears in the late second to early third decades of life[1,5] and is more often associated with OI type I, III and IV.[6] The hearing loss may be conductive, sensorineural or mixed. Until the 1970s, the middle ear changes in OI were considered to be a severe form of otosclerosis and therefore stapes surgery was considered to be more risky. However, histological,[7–9] histochemical[10] and clinical-genetic studies have supported the opinion that otosclerosis and OI are different disease entities, despite having very similar clinical manifestations.[2,11] The first successful stapedectomy in OI was reported by Sooy in 1960.[12] Since then a series of reports on stapes surgery in OI have been published and their results are in general very satisfactory.[1,9,13–19] The present study is the first to attempt an evaluation of the short-term and long-term effect of stapedotomy in improving hearing in patients with OI. Our findings are compared with previously published series on the use of stapedectomy for the same purpose.

Patients and methods

Between 1985 and 1996 nine patients with OI underwent stapes surgery in our department. All patients were followed up for a minimum of three years (range 3–11 years). There were four males and five females, ranging in age from 24 to 45 years (median 35 years). The diagnosis of OI was based on a life-long history of multiple fractures, the presence of skeletal deformities and blue sclerae and a family history compatible with dominant or recessive inheritance.

Audiological evaluation included otoscopic examination, standard aural immittance measurements and pure-tone audiometry. The external ears were examined for otological clinical abnormalities and the tympanic membranes were inspected for evidence of scars, perforation, tympanosclerosis and colour changes. Otoscopic examination was normal in seven patients, while in two cases a translucent, flushed appearance of the tympanic membrane was noticed. Middle-ear function was evaluated through a set of immittance measurements (acoustic admittance at 226 Hz, ipsilateral and contralateral acoustic reflex thresholds at 500 Hz, 1000 Hz, 2000 Hz and 4000 Hz) using the GSI-33 (Grason-Stadler Incorporation, Milford, New Hampshire) middle-ear analyser. Pure-tone audiometric thresholds were measured at octave frequency intervals from 250–8000 Hz using the American Speech and Hearing Association guidelines.[20] All patients presented with a rather symmetrical bilateral mixed hearing impairment (Figure 1). The mean age for the onset of hearing loss was 21 years.

The indications for ear surgery were a normal tympanic membrane, a negative Rinne-test with 512 Hz and 1024 Hz tuning forks and an air-bone gap on pure-tone audiometry >20 dB HL in the main speech frequency range (250–4000 Hz). A small fenestra stapedectomy (stapedotomy) was performed in all patients according to the classic technique of Schuknecht.[21] The oval window was exposed through an endomeatal approach, the anatomical landmarks were inspected and the mobility of the ossicular chain was tested. A small fenestra was created in the middle of the footplate using a small diamond drill. A 0.4 mm thick Teflon wire prosthesis was inserted to the fenestra without interposition and crimped simultaneously on the long process of the incus. Next the stapedial tendon and the posterior crus were sectioned, the incudostapedial joint was separated and the anterior crus was fractured towards the promontory. The hook of the prosthesis was then moved caudally to the desired position on the incus where it was firmly crimped. A blood clot mixed with Gelfoam pledgets was used to seal the fenestrated footplate.

A retrospective, one-group, pre-test-post-test design was used in which each patient's pre-operative and post-operative audiograms were compared. Data are presented as the mean and standard deviation (± SD) of the mean. Differences in air conduction and bone conduction thresholds pre-operatively and post-operatively were compared with a two-tailed paired Students *t*-test. A p value of <0.05 was considered significant.

Results

All patients had type A tympanograms, suggesting the decreased compliance of the middle-ear system, while no acoustic reflex responses were obtained for the speech frequencies tested.

Post-operatively, a marked improvement was observed in all patients. Figure 1 shows the mean pre-operative and post-operative air (250–8000 Hz) and bone conduction thresholds (250–4000 Hz). The pre-operative air conduction thresholds ranged from 40-90 dB HL, with the highest threshold at 8000 Hz. The pre-operative bone conduction thresholds varied between 15–60 dB HL and similar to air conduction thresholds, the higher frequencies were more severely affected.

Post-operatively, air conduction thresholds at 250 Hz, 500 Hz and 1000 Hz improved and were within the normal range ($\leqslant$25 dB HL). However, the thresholds at 2000–8000 Hz were still outside the normal hearing range (from 30 dB HL at 2000 Hz to 95 dB HL at 8000 Hz). Post-operative bone conduc-

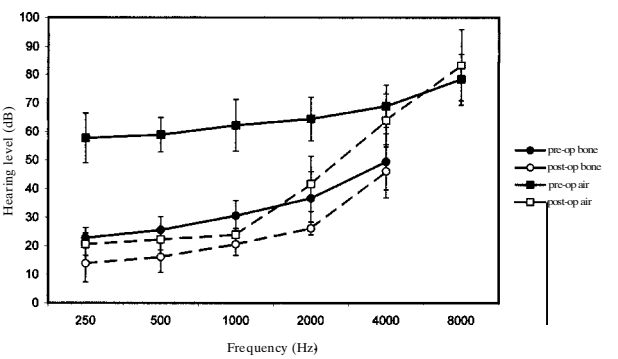

FIG. 1

Pre-operative versus post-operative audiometric results for air and bone conduction thresholds in osteogenesis imperfecta patients. Mean values ± SD are indicated.

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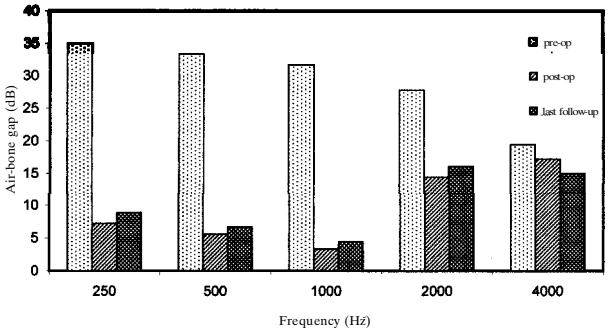

FIG. 2

Pre-operative versus post-operative air-bone gaps in the frequency range 250–4000 Hz.

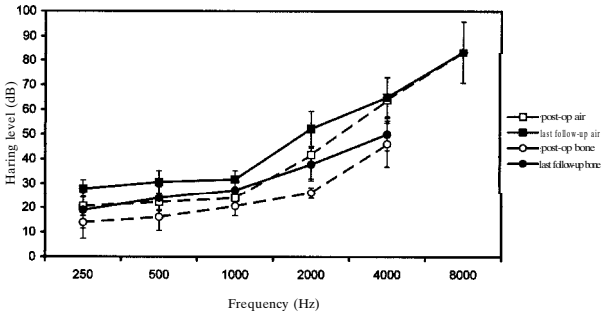

FIG. 3

Long-term results of stapedotomy on mean pure-tone hearing thresholds during the follow-up period. Mean values ± SD are indicated.

tion thresholds were all within the normal range except at 4000 Hz (range 30–55 dB HL). These data suggest that surgery had a greater impact on hearing in the low rather than in the high frequency range.

Surgery resulted in a statistically significant improvement in thresholds for air conduction at 250–4000 Hz ($p<0.05$) and in bone conduction for 250–2000 Hz ($p<0.05$). In contrast, the frequencies >4000 Hz in air and >2000 Hz in bone conduction did not show a significant change post-operatively.

Post-operatively all patients showed a large reduction in their air-bone gap. The air-bone gap was significantly smaller ($p<0.05$) at 250 Hz, 500 Hz, 1000 Hz and 2000 Hz. Therefore, surgery was more successful in closing the air-bone gap in the low and middle rather than in the high frequency range. Figure 2 shows the pre-operative versus post-operative air-bone gaps in the frequency range 250–4000 Hz.

During the follow-up period, a progressive deterioration of the post-operative result was noticed for all patients. However, in none had the hearing acuity deteriorated to pre-operative levels. The mean threshold shift was 7.7 dB HL (ranging from 0–25 dB HL), with a non-statistically significant change ($p>0.05$) in air-bone gap for the whole frequency range (Figure 2). Figure 3 shows the long-term results of stapedotomy in the frequency range 250–8000 Hz. This progressive perception loss could not be related directly to the operation and probably results from the natural course of the disease, since a similar progression was also seen in the non-operated ears of the patients.

Our operative findings are presented in Table II. Inspection of the ossicular chain revealed a mobile malleus and incus with normal configuration. The stapes footplate was thick and firmly fixed in six of the nine ears by an anterior, otosclerosis-like focus in the region of the oval window, while in the remaining three a diffuse thickening of the footplate was noticed, bearing no resemblance to otosclerosis. In no case were the stapes crura abnormally slender, brittle or fractured. In two cases, the hyperplasia and hypervascularization of the promontorial mucosa caused mild bleeding, which was easily controlled by short-term local application of vasconstrictor agents on Gelfoam. A floating footplate following stapes manipulations or a 'gusher' or 'oozer' phenomenon after opening the vestibule was not observed.

Discussion

Hearing loss has been repeatedly reported as a major symptom of OI. The conductive component prevails in the younger patients[22] and is usually the result of fixation of the stapes footplate. However, functional ossicular discontinuity, due to stapes superstructure fracture or fibrous replacement, has also been reported.[9,23] Pure sensorineural hearing loss, especially at high frequencies, is quite common independent of age[24] and probably results from microfractures, haemorrhage and encroachment of reparative vascular and fibrous tissue in and about the cochlea.[7]

Hearing ability declines with advancing age, significantly more than could be expected from presbycusis. In OI, the sensorineural component shows an average annual increase of 0.6 dB/year at

TABLE II

INTRA-OPERATIVE FINDINGS DURING STAPEDOTOMY IN PATIENTS WITH OSTEOGENESIS IMPERFECTA

Patient	Sex/age	Intra-operative findings		
		Stapes crura	Stapes footplate	Middle ear mucosa
1	35/F	Normal	Firmly fixed thick anterior focus	Normal
2	45/F	Normal	Firmly fixed thick anterior focus	Normal
3	26/M	Normal	Diffuse thickening	Hypertrophic – vascular
4	30/F	Normal	Firmly fixed thick anterior focus	Normal
5	36/F	Normal	Diffuse thickening	Normal
6	42/M	Normal	Firmly fixed thick anterior focus	Hypertrophic – vascular
7	29/F	Normal	Firmly fixed thick anterior focus	Normal
8	24/M	Normal	Diffuse thickening	Normal
9	44/M	Normal	Firmly fixed thick anterior focus	Normal

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500 Hz up to 1.3 dB at 8000 Hz, while the average annual increase in the conductive component remains constant at 0.4 dB/year at all frequencies.[19] This progression of hearing loss with age occurs in both operated and nonoperated ears and is probably due to the natural course of the disease.[19] Thus, the average hearing gain achieved after operation will tend to be eliminated only after several decades. Patients at increased risk of complications after stapes surgery are usually the ones who present with early onset of hearing loss, higher pre-operative bone conduction threshold, thin and atrophic crura, thicker than normal middle ear mucosa and massive obliterative disease.[25]

Published series on stapes surgery in OI patients have all utilized stapedectomy.[1,9,13–18,26] To our knowledge, this is the first report on the use of stapedotomy in OI patients. It was preferred over stapedectomy based on reports, that suggest that it preserves better the high frequency hearing range in otosclerosis patients.[27,28]

The majority of previous studies on stapedectomy in OI patients present an excellent short-term success rate (mean hearing gain of 25–30 dB HL) in 85 per cent to 100 per cent of their patients.[9,15–17,23,26] Our results with stapedotomy were also very satisfactory, confirming the well-established success of middle-ear surgery in OI patients, especially for the low frequency range. Regarding the long-term results the three existing studies[17,23,26] show an average reduction in hearing gain of approximately 8 dB HL with an almost unchanged air-bone gap. A similar shift was also observed in our series, although not dramatically affecting the patients hearing acuity during the follow-up period. However, all these previous studies focus on the mean hearing gain and the mean change of the air-bone gap only in the low frequency range. The hearing status in the higher frequencies is not mentioned or is only couched in vague terms, therefore leaving unanswered questions regarding the effect of stapes surgery in the high frequency range ($\geqslant$4000 Hz). In addition, all these studies lack statistical analysis. Our study, despite its small number of patients, is the first one to provide detailed audiometric data for the efficacy of stapedotomy in OI patients, covering the whole frequency range both the short- and long-term. A statistically significant improvement of the air conduction threshold at 4000 Hz was observed, although it remained outside the normal range. However, air conduction at 8000 Hz and bone conduction at 4000 Hz remained nearly unchanged.

The most commonly reported surgical findings include the presence of soft bone in the posterior-superior quadrant of the bony annulus, hyperaemic middle-ear mucosa, delicate ossicles with spontaneous fractures and discontinuities and footplates changes in which the footplate is covered with a mass of soft, white, chalky bone, but with little or no fixation to the margins of the oval window.[1,7,9,16,17,24,29] In some studies, however, a large percentage of the patients had normal stapedial crura or a firmly fixed stapes footplate, similar to our patients,[9,17] reflecting the great heterogeneity in OI.

Conclusions

Stapedotomy in OI patients resulted in a significant improvement in air conduction thresholds at 250–4000 Hz and in bone conduction thresholds at 250–2000 Hz. The air-bone gap was significantly decreased in the low and middle frequencies and remained nearly unchanged in the high frequency range. The high frequency hearing loss (>4000 Hz) that existed in the pre-operative audiograms remained essentially stable after surgery. The long-term results were also encouraging with no significant deterioration over time. These satisfactory results and the lack of complications make stapedotomy an appealing method for the management of OI-associated hearing loss.

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