Acta Otorrinolaringol Esp. 2009;**60(2):**126-30

Acta Otorrinolaringológica Española

www.elsevier.es/otorrino

BRIEF COMMUNICATION

Surgical options for hearing loss in patients with osteogenesis imperfecta

Manuel Sainz, Juan García-Valdecasas,* and José M. Ballesteros

Servicio de Otorrinolaringología, Hospital Universitario San Cecilio, Granada, Spain

Received September 25, 2008; accepted October 24, 2008

KEY WORDS
Osteogenesis imperfecta;
Hearing loss;
Cochlear implants;
Ossicular prostheses;
Middle ear implants

Abstract
The aim of the study is to present the severe, extensive lesions in the temporal bone appearing in the mildest forms of osteogenesis imperfecta, correlate these with audiometric results, and discuss the possible surgical treatments available. We present 3 patients suffering hearing loss due to osteogenesis imperfecta type 1 to various functional degrees. All patients underwent an audiological, medical, and radiological evaluation and were then treated with different surgical procedures. The patients presented mild, severe, and profound mixed hearing losses and the radiological images showed extensive areas of demineralization affecting the ossicular chain and removal of protection for the vital structures inside the temporal bone. Also, the cochlea showed otic capsule demineralization, dehiscence, distortions, and even destructions. The various surgical treatments, indicated under current international criteria, obtained limited functional results. This study reviews the need to evaluate the current surgical criteria for this specific condition.

© 2008 Elsevier España, S.L. All rights reserved.

PALABRAS CLAVE
Osteogénesis imperfecta;
Hipoacusia;
Implantes cocleares;
Prótesis osiculares;
Implantes de oído medio

Posibilidades de tratamiento quirúrgico de la hipoacusia en pacientes afectados de osteogénesis imperfecta

Abstract
El objetivo del estudio es presentar las extensas y severas lesiones óticas que acontecen en la forma más leve de osteogénesis imperfecta, correlacionarlas con las alteraciones audiométricas y discutir las posibilidades quirúrgicas disponibles. Se presenta a 3 pacientes afectados de osteogénesis imperfecta tipo 1 con hipoacusia en distintos estadios funcionales. Se realizó a cada paciente estudio audiológico, médico y radiológico, y se sometieron a distintos tratamientos

*Corresponding author.
E-mail address: orlvaldecasas@gmail.com (J. García-Valdecasas).

0001-6519/$ - see front matter © 2008 Elsevier España, S.L. All rights reserved.

quirúrgicos. Los pacientes presentaron hipoacusias mixtas de carácter moderado, severo y profundo, imágenes radiológicas con extensas áreas de desmineralización que afectaban la cadena osicular y con desprotección de las estructuras vitales alojadas en el peñasco temporal, desmineralización de la cápsula ótica, dehiscencias, distorsiones y destrucciones cocleares. Los tratamientos quirúrgicos realizados, e indicados según los criterios actualmente aceptados, obtuvieron escasos resultados funcionales. El presente estudio plantea la necesidad de evaluar los criterios quirúrgicos específicos para esta enfermedad.

© 2008 Elsevier España, S.L. Todos los derechos reservados.

Introduction

Osteogenesis imperfecta is a genetic disorder of the connective tissue caused by a mutation in one of the 2 genes encoding the alpha chains of type I collagen.[1] The type I collagen molecule comprises 2 polypeptide chains, alpha 1 and alpha 2, which form a triple helix housing a molecule of glycine, which is essential in the definitive structure and a final target for genetic alterations.[2] Depending on the type and location of the glycine substitution, the phenotype varies from slight ailments to the most severe which include even death in early stages of life. On the other hand, the mildest form of osteogenesis imperfecta, the type I form, with autosomal dominant inheritance,[1-3] leads to greater bone mineral density with less tensional resistance and greater fragility,[2] blue or dark grey sclera, and progressive hearing loss.

Hearing loss occurs in 42%-58% of patients with type I osteogenesis imperfecta and begins in the second or third decade of life. This hearing loss is progressive; it is initially transmissive; then mixed and finally sensorineural.[4-6] Transmissive loss is caused by atrophy and subsequent fracture of stapedial crura, thickening and fixation of the plate of the stapes and increased vascularization and hypertrophy of the promontorial mucosa. This can be treated by stapedial prosthesis[4,5,7] and even implantable devices in the middle ear.[8] Sensorineural hearing loss (25%-60%) results from pericochlear and intracochlear changes promoted by demineralization, microfractures, haemorrhages, and subsequent inclusion of fibrous and regenerative tissue[9,10]; it can be treated in severe and profound stages by cochlear implants.[6,11,12] These lesions of the inner ear progress rapidly to severe demineralization affecting, in early stages, an adequate cochlear implantation.

The aim of this study is to present the extensive and severe lesions of the inner ear and the temporal bone which occur in the milder forms of osteogenesis imperfecta, correlate this condition with the audiometric findings and discuss the surgical options available.

Methods

We present 3 patients, 2 men and 1 woman, suffering from type I osteogenesis imperfecta, who came to our otolaryngology service for audiological advice due to poor perception of sound and reverberations using headphones. All patients reported hypoacusis, vertigo, and tinnitus. The average (standard deviation) for age of onset of hypoacusis was 27 (7.13) years and average evolution until the first otolaryngology interview was 9.5 (9.79) years.

We conducted a medical and radiological evaluation and a computerized tomography (CT) with serial axial slices of 1 mm and audiological test (tonal audiometry).

Results

All patients presented mixed hypoacusis (Table 1), profound in the case of patients 1 and 2 and moderate in patient 3. The CT images produced showed various degrees of demineralization of the temporal bone (Figures 1-3 and Table 1), leaving its vital structures unprotected (dura mater, facial nerve, lateral sinus, jugular bulb, and internal carotid artery). The otic capsule presented rarefaction and even fistulae of the inner ear with middle ear and medial fossa. Demineralization also affected the ossicular chain, located primarily on the incus and the stapes, although lesions were also found in the malleus. Patients 1 and 2 presented demineralizing intracochlear lesions erasing areas of the basal and middle spiral canals (Table 2).

Cochlear implantation was attempted with a Med-El Pulsar Ci100™ device in patient 1, with no problems during the antrotomy, the subsequent tympanostomy, or the cochleostomy. The insertion of the electrode guide presented arrests and deviations which prevented the implant after multiple attempts.

Patient 2 was referred for brainstem implantation due to the large distortion of the basal and middle spires. This possibility was rejected by the patient.

Only 3 patients had been operated on through exploratory tympanostomy and cleaning of the space, with excellent initial results despite widespread demineralization of the stapes, anvil and hammer. After the failure of this procedure, an implantation was carried out of an electromechanical device for the middle ear (Vibrant Soundbridge™) as well as the placement of the corresponding transducer in the round window. One month after surgery, during the start-up of the device, the patient did not show any benefit whatsoever with worsening by 30 dB of the hearing threshold obtained by stimulation of the bone pathway. The subsequent revision surgery confirmed the presence of connective tissue in the middle ear that may have affected the inner ear through the multiple fistulae from the otic capsule.

Discussion

While severe forms of osteogenesis imperfecta cause physical defects and even death at early stages of life, the mild forms (type I) may lead to moderate and severe

Table 1 Hearing thresholds obtained through tonal audiometry

Patient	Ear	Threshold, dB-HL	125 Hz	250 Hz	500 Hz	1 kHz	2 kHz	4 kHz	8 kHz	Max. verbal discrimination
1	Right	Air way	85	105	105	115	120	120	120	45%
		Gap	35	Ind	Ind	Ind	Ind	Ind	Ind	
	Left	Air way	120	120	115	120	120	120	120	40%
		Gap	45	50	Ind	Ind	Ind	Ind	Ind	
2	Right	Air way	115	115	115	120	120	120	120	50%
		Gap	Ind	Ind	Ind	Ind	Ind	Ind	Ind	
	Left	Air way	85	105	115	120	110	115	120	40%
		Gap	Ind	Ind	Ind	Ind	Ind	Ind	Ind	
3	Right	Air way	50	75	75	70	80	80	95	70%
		Gap	10	45	40	15	15	15	Ind	
	Left	Air way	35	40	55	65	60	60	85	75%
		Gap	15	20	30	30	25	0	Ind	

The thresholds expressed are those obtained by acoustic stimulation of the air way and the gap (difference between thresholds obtained by air and bone pathways).
Ind indicates indeterminate gap. This is due to not being able to obtain the bone threshold due to the impossibility of stimulating above 70 dB.

Table 2 Major demineralizing lesions found in the CT scans of patients with osteogenesis imperfecta, classified according to the place of occurrence

Middle ear	Inner ear	Temporal bone
Ossicular demineralization. Anvil and stapes	Rarefactions and fistulas of the otic capsule. Cochlear destruction	Loss of protection: dura mater, facial nerve, lateral sinus, jugular bulb, and internal carotid

hearing loss in the second and third decade of life due to extensive demineralization of the structures of the temporal bone.[3,5] These demineralizing lesions advance as the disease progresses. Initially, the ossicular chain undergoes intense demineralization with atrophy and fractures of the stapedial crura. In addition to this, the thickening and fixation of the stapes plate and hypervascularization and hypertrophy of the promontorial mucosa[4,5,7,14,15] are involved in the initial transmissive hypoacusis which can be treated, with excellent results in the short term, by the placement of stapedial prosthesis.[4,14,15] The excessive tendency to bleeding, caused by the thin and atrophic skin of the external auditory canal and hypervascularization and hypertrophy of the promontorial mucosa, as well as the fragility of the mastoid bone and the involuntary stapedial fractures, is the most common complication during the performance of this procedure.[7,13]

Next, demineralization continues and more severe and extensive lesions appear, affecting the inner ear. Thus, the dehiscences of the otic capsule, microfractures, haemorrhages, and cochlear occupation by connective tissue favour the emergence of mixed hypoacusis and reduce the effectiveness of stapedial prostheses.[13-15] Finally, the lesions progress to the complete distortion of the cochlear structures and the loss of protection for the jugular bulb, lateral sinus, carotid sinus, dura mater, and facial nerve.[10]

Figure 1 Computerized tomography. Patient 1. Severe-profound mixed hypoacusis.

One of the challenges of this disease lies in the counselling of patients dissatisfied with the adaptation of hearing aids. In general, the initial moderate transmissive hearing loss can be treated through stapedial prostheses, which fail in the medium term (mean, 9.5 years; median, 4 years) as the disease advances.[13,14] On the other hand, as the disease progresses and severe sensorineural hypoacusis

Figure 2 Computerized tomography. Patient 2. Severe-profound mixed hypoacusis.

Figure 3 Computerized tomography. Patient 3. Moderate mixed hypoacusis.

sets in, surgical treatment with cochlear implants would be indicated in line with currently accepted criteria.[16,17] The use of auditory brainstem implants would be reserved for cases of extensive cochlear destruction.

On the contrary, as in other bone dysplasias,[18] cochlear distortions and ossifications hinder the insertion of the electrode guide of the cochlear implant, thereby reducing the effectiveness of these devices. Especially in this disease, these lesions occur early in the natural history and influence a rapid decision on cochlear implantation because, once severe-profound hypoacusis is installed, the extensive lesions may contraindicate implantation and compel the implementation of hearing devices in the brainstem as an elective treatment. These have less efficacy and an increased incidence of major complications.[19,20]

The biggest challenge seems to be moderate mixed hypoacusis. When this appears and the stapedial prostheses fail, solutions do not seem to be defined. In such cases, 2 strategies can be adopted: *a)* middle ear implants: the fragility of the ossicular chain conditions the ossicular placement of these devices, thus implantation in the round window is preferred.[21] In addition, as occurred after the intervention of patient 3, post surgical scarring changes could affect the hearing threshold due to the inclusion of connective tissue in the cochlea through dehiscences of the otic capsule. Even if the middle ear implant can be carried out, the results will presumably last a short time, due to the progression of the disease; *b)* cochlear implantation: when moderate hypoacusis is established, cochlear implantation is not indicated in line with current criteria.[16,17] In any case, as the demineralization advances, hearing loss progresses and cochlear implantation becomes the most appropriate treatment. Due to the rapid progression of demineralizing lesions, cochlear implantation will have more possibilities when moderate hypoacusis is established than in cases of severe or profound hearing loss with severe inner ear injuries.

At least, in our experience, cochlear implantation in severe and profound hypoacusis caused by osteogenesis imperfecta was impossible with any minimum expectation of success.

References

1. Rauch F, Glorieux FH. Osteogenesis imperfecta. Lancet. 2004;363:1377-85.
2. Cheung MS, Glorieux FH. Osteogenesis imperfecta: update on presentation and management. Rev Endocr Metab Disord. 2008;9:153-60.
3. Sillence DO, Senn A, Danks DM. Genetic heterogenicity in osteogenesis imperfecta. J Med Genet. 1979;16:101-16.
4. Doi K, Nishimura H, Ohta Y, Kubo T. Stapes surgery in Japanese patients with osteogenesis imperfecta. Adv Otorhinolaryngol. 2007;65:226-30.
5. Hulcrantz M, Sääf M. Stapes surgery in osteogensis imperfecta: a clinical study of 16 patients. Adv Otorhinolaryngol. 2007;65: 222-5.
6. Streubel SO, Lustig LR. Cochlear implantation in patients with osteogenesis imperfecta. Otolaryngol Head Neck Surg. 2005; 132:735-40.
7. Vincent R, Gratacap B, Oates J, Sperling NM. Stapedotomy in osteogenesis imperfecta: a prospective study of 23 consecutive patients. Otol Neurotol. 2005;26:859-65.
8. Backous DD, Duke W. Implantable middle ear hearing devices: current status of technology and market challenges. Curr Opin Otolaryngol Head Neck Surg. 2006;14:314-8.
9. Cohen BJ. Osteogenesis imperfecta and hearing loss. Ear Nose Throat J. 1984;63:283-8.
10. Alkadhi H, Rissmann D, Kollias SS. Osteogenesis imperfecta of the temporal bone: CT and MR imaging in van der Hoeve-de Kleyn syndrome. Am J Neuroradiol. 2004;25:1106-9.
11. Mens HM, Mulder JS. Average electrode voltage in users of the Clarion cochlear implant device. Ann Otol Rhinol Laryngol. 2002;11:370-5.

12. Migirov L, Henkin Y, Hidelsheimer M, Kronenberg J. Cochlear implantation in a child with osteogenesis imperfecta. Int J Pediatr Otorhinolaryngol. 2003;67:677-80.
13. Kuurila K, Kaitila I, Johanson R, Grénman R. Hearing loss in Finnish adults with osteogenesis imperfecta: a nationwide survey. Ann Otol Rhinol Laryngol. 2002;111:939-46.
14. Ferekidis E, Stavroulaki P, Vossinakis I, Yiotakis J, Manolopoulos L, Adamopoulos G. Stapedotomy in osteogenesis imperfecta patients. J Laryngol Otol. 2000;114:424-8.
15. Antoon JM, van der Rijt, Cor WRJ. Stapes surgery in osteogenesis imperfecta: results of a new series. Otol Neurotol. 2003;24: 717-22.
16. NIH Consensus Conference. Cochlear implants in adults and children. J Am Med Assoc. 1995;274:1955-61.
17. Cohen NL. Cochlear implant candidacy and surgical considerations. Audiol Neurotol. 2004;9:197-202.
18. Sainz M, Garcia-Valdecasas J, Garofano M, Ballesteros JM. Otosclerosis: mid-term results of cochlear implantation. Audiol Neurotol. 2007;12:401-6.
19. Schwartz MS, Otto SR, Shannon RV, Hitselberger WE, Brackmann DE. Auditory brainstem implants. Neurotherapeutics. 2008;5: 128-36.
20. Cervera-Paz FJ, Manrique MJ. Auditory brainstem implants: past, present and future prospects. Acta Neurochir Suppl. 2007; 97:437-42.
21. Colletti V, Soli SD, Carner M, Colletti L. Treatment of mixed hearing losses via implantation of a vibratory transducer on the round window. Int J Audiol. 2006;45:600-8.