

# DEAFNESS IN OSTEOGENESIS IMPERFECTA OR VAN DER HOEVE'S SYNDROME

## OUR EXPERIENCE

**P. CORLIEU**

Cochin & Tenon Hosp.  
Univ. Paris Descartes

*Alexandrie April 2007*

## HEARING IN OSTEOGENESIS IMPERFECTA

Osteogenesis Imperfecta (O.I.) is an autosomal dominant character transmitted syndrome with great variation in genetic expressivity.

It appears in young children with:

- fragile bones
- thinning of bone cortical
- ligament hyperlaxity
- blue sclerae
- belated deafness



## HEARING IN OSTEOGENESIS IMPERFECTA

### Deafness

- generally occurs in adults but are possible in children
- uni or bilateral
- progressive
- concern about half of patients with O.I.

## HEARING IN OSTEOGENESIS IMPERFECTA

### Deafness

- Perceptive deafness : → Prosthesis
- Conductive deafness by ossicular ankylosis : → Surgery

## HEARING IN OSTEOGENESIS IMPERFECTA

In O. I. abnormal collagen constitution induce very thin osseous trabeculations and malformations of the otic capsule

*Milroy CM, Michaels L, J  
Laryngol Otol 1990; 104(2):83-90*



## HEARING IN OSTEOGENESIS IMPERFECTA

- **Morphological and microchemical modifications in otosclerosis and O. I.**

Method: electronic microscopy with analysis of X rays energy dispersion

- **Otosclerosis:** low mineralized lesions with decreasing of Ca/P ratio → spongy lesions with unsteady mineralization (transformation of hydroxylapatite in tricalcium phosphate)
- **O. I.:** presence of magnesium in stapes suggest a stimulation of the osteoclastic activity

*Valléjo-Valdezate LA, Martin-Gil J, Jose-Yacaman M, Martin-Gil FJ, Gil-Carcedo LM. Laryngoscope 2000 Sep;110(9):1505-10*

## HEARING IN OSTEOGENESIS IMPERFECTA

- **Petrous bone locations:**

More or less important lesions in otic capsule development

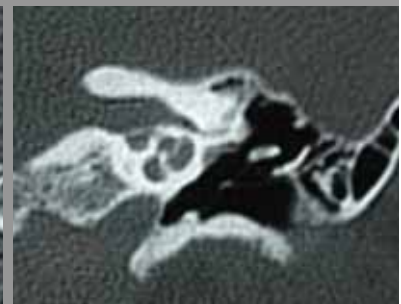
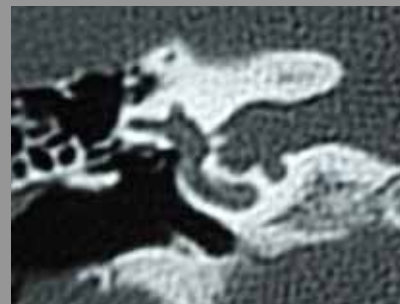
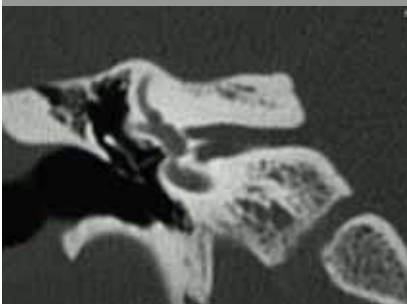
Disturbance of the bony lamellar constitution of the stapes resulting in an excessive fineness, a dehiscence or a lack of union between stapes crura and the footplate.

*Nager GT. Ann Otol Rhinol Laryngol 1988 Nov-Dec; 97(6Pt):585-93*

## HEARING IN OSTEOGENESIS IMPERFECTA

**Normal bone**

**NOB.Emmanuel 38 yrs  
Pathological petrous bone**



## HEARING IN OSTEOGENESIS IMPERFECTA

- **CT scan and high resolution scintigraphy**

High decreasing of bony pericochlear density and increasing of osseous metabolism in cases of O. I. with important mixt deafness

But absence of CT scan and scintigraphic cochlear modifications in some cases with mixt deafness or in cases with normal hearing

*Ross UH, Laszig R, Bornemann H, Ulrich C. Acta Otolaryngol 1993 Sep;113(5):620-4*

## HEARING IN OSTEOGENESIS IMPERFECTA

- **MRI**

Peri-cochlear lesions with soft tissue signal and gadolinium enhancement

*MRI- visible pericochlear lesions in osteogenesis imperfecta type I.*

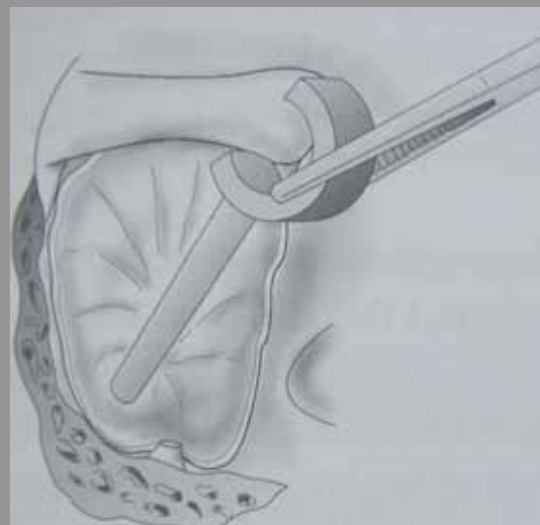
*Ziyeh S, Berger R, Reisner K. Eur Radiol 2000; 10(10):1675-7*

## HEARING IN OSTEOGENESIS IMPERFECTA

- Even the etiopathogeny of conductive deafness of O. I. is lightly different of the pathogeny of otosclerosis, they both result to an ankylosis of the stapes
- The surgical treatment is similar in O.I. and otosclerosis

## HEARING IN OSTEOGENESIS IMPERFECTA

- **Surgical procedure**
  - Stapes prosthesis
  - General or local anesthesia
  - About 30 mn
  - Short hospitalisation



## HEARING IN OSTEOGENESIS IMPERFECTA

- **Our experience**

- 10 patients, 7 men and 3 women
- Average age was 35 ( 16 to 61 )
- 2 patients with sensorineural deafness
- 1 patient operated on both sides by other team with good results
- 1 patient to operate soon on one side
- 6 patients with uni or bilat conductive deafness
- 10 ears operated
- Post-op average delay: 8.9 years ( 17 y – 6 m )

## HEARING IN OSTEOGENESIS IMPERFECTA

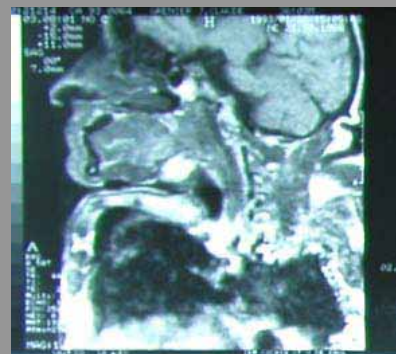
• <b>ROU.</b>	<b>Mehdi</b>	<b>16 y</b>	
		<b>Right</b>	<b>Left</b>
• <b>Post-op follow-up</b>		<b>17 y</b>	<b>16 y</b>
• <b>Pre-op level</b>		<b>51 dB</b>	<b>47 dB</b>
• <b>Pre-op gap</b>		<b>34 dB</b>	<b>22,5 dB</b>
• <b>Constatations</b>		<b>Footplate blocked</b>	<b>Footplate block.</b>
• <b>Technique</b>		<b>Platinotomy</b>	<b>Platinotomy</b>
• <b>Conductive improv.</b>		<b>21,5 dB</b>	<b>9 dB</b>
• <b>Residual gap</b>		<b>12,5 dB</b>	<b>13,5 dB</b>

## HEARING IN OSTEOGENESIS IMPERFECTA

• PER.	Arlette	61 y	
		<b>Right</b>	<b>Left</b>
• Post-op follow-up		11 y	12 y
• Pre-op level		65 dB	66 dB
• Pre-op gap		41 dB	34 dB
• Constatations		Footplate blocked Very thick	Footplate block.
• Technique		Platinotomy	Platinotomy
• Conductive improv.		35 dB	29 dB
• Residual gap		6 dB	5 dB

## HEARING IN OSTEOGENESIS IMPERFECTA

- GRE. J. Cl. 37 y, 80 cm high





## HEARING IN OSTEOGENESIS IMPERFECTA

• <b>GRE. J. Cl.</b>	<b>37 y</b>	
	<b>Right</b>	<b>Left</b>
• <b>Post-op follow-up</b>	<b>9 y</b>	<b>10 y</b>
• <b>Pre-op level</b>	<b>50 dB</b>	<b>61 dB</b>
• <b>Pre-op gap</b>	<b>24 dB</b>	<b>34 dB</b>
• <b>Constatations</b>	<b>Footplate blocked Crura fracture</b>	<b>Footplate block.</b>
• <b>Technique</b>	<b>Platinotomy</b>	<b>Platinotomy</b>
• <b>Conductive improv.</b>	<b>8 dB</b>	<b>30 dB</b>
• <b>Residual gap</b>	<b>16 dB</b>	<b>5.5 dB</b>

## HEARING IN OSTEOGENESIS IMPERFECTA

• <b>NOB. Emmanuel</b>	<b>41 y</b>	
	<b>Right</b>	<b>Left</b>
• <b>Post-op follow-up</b>	<b>6 m</b>	
• <b>Pre-op level</b>	<b>27 dB</b>	<b>0 dB</b>
• <b>Pre-op gap</b>	<b>17 dB</b>	
• <b>Constatations</b>	<b>Footplate blocked</b>	
• <b>Technique</b>	<b>Platinotomy</b>	
• <b>Conductive improv.</b>	<b>4 dB</b>	
• <b>Residual gap</b>	<b>8 dB</b>	



## HEARING IN OSTEOGENESIS IMPERFECTA

- **FAU. Mederic 31 y**

	<b>Right</b>	<b>Left</b>
• <b>Post-op follow-up</b>		<b>1 y</b>
• <b>Pre-op level</b>	<b>9 dB</b>	<b>39 dB</b>
• <b>Pre-op gap</b>		<b>29 dB</b>
• <b>Constatations</b>		<b>Footplate block.</b>
  
- **Technique** **Platinotomy**
- **Conductive improv.** **20 dB**
- **Residual gap** **9 dB**

## HEARING IN OSTEOGENESIS IMPERFECTA

- **MER. Stephane 28 y**

	<b>Right</b>	<b>Left</b>
• <b>Post-op follow-up</b>	<b>5 y</b>	<b>1 y</b>
• <b>Pre-op level</b>	<b>64 dB</b>	<b>41 dB</b>
• <b>Pre-op gap</b>	<b>40 dB</b>	<b>15 dB</b>
• <b>Constatations</b>	<b>Footplate block.</b>	<b>Thick footplate</b>
  
- **Technique** **Platinotomy** **Platinotomy**
- **Conductive improv.** **34 dB** **17 dB**
- **Residual gap** **6 dB** **0 dB**

## HEARING IN OSTEOGENESIS IMPERFECTA

- **Audiological findings**
  - In a group of 56 patients with osteogenesis imperfecta, 31 patients had an hypoacusia
  - Deafness began in the second and third decade: conductive deafness becoming uni or bilateral mixt deafness

*Stewart EJ, O'Reilly BF. Clin Otolaryngol 1989 Dec;14(6):509-14*

## HEARING IN OSTEOGENESIS IMPERFECTA

**Spontaneous evolution of hearing between the age of 10 to 45 years old in a group of 142 non operated patients with O.I.**

- **Annual average worsening of hearing level**  
1dB/y (.5 to 4 kHz) and 1.7 dB/y (8kHz)
- **Annual average worsening of perceptive level**  
0.6 dB/y (.5 à 4 kHz) and 1.3 dB/y (8k
- **Annual average worsening of the gap:**  
.4 dB on each frequency

*Garretsen AJ, Cremers CW, Huygen PL. Ann Otol Rhinol Laryngol 1997 Jul;106(7 Pt):575-82*

## HEARING IN OSTEOGENESIS IMPERFECTA

With regards to 8 ears operated among 6 patients with O.I. (jan 88 to dec 94)

Atrophy and/or stapes fracture associated with a thickening and an ankylosis of the footplate

Résultats: no significant difference with otosclerosis operated during the same time

*Dieler R, Muller J, Helms J. Eur Arch Otorhinolaryngol 1997;254(3):120-7*

## HEARING IN OSTEOGENESIS IMPERFECTA

6 patients with O.I., 5 patients operated by platinotomy (1989 to 2000)

- Good results for all patients
- No complication
- Spontaneous fracture of stapes crura with +/- floating footplates

– *Albahnasawy L, Kishore A, O'Reilly BF. Clin Otolaryngol 2001 Dec;26(6)/473-6*

## HEARING IN OSTEOGENESIS IMPERFECTA

- **Operatory results: Garretsen TJ & Cremers CW**
  - 58 ears operated (47 patients)
  - Good results at 3 months: 85%
  - 68% of 40 ears with a long term control (2 à 24 y.) had preserved their surgical benefit
  - For other operated ears, the decreasing of the surgical benefit by progressive labyrinthisation was the same evolution as non operated ears: spontaneous evolution of the illness

*Garretsen TJ, Cremers CW. Arch Otolaryngol Head Neck Surg 1990 Mar; 116(3)317-23*

## HEARING IN OSTEOGENESIS IMPERFECTA

**Hearing control of patients with osteogenesis imperfecta is important because the surgical procedure give regular good results.**

**We have to inform colleagues and patients of this possibility of functional improvment**