

Short Communication**Otosclerosis among first and second degree Relatives of Otosclerotic patients**

S. Soheilipour MD\*, S. Nemati MD\*\*

**ABSTRACT**

**Background:** Otosclerosis (OS) is the most common cause of conductive hearing loss in young population, and we determined incidence of OS among 1st and 2nd degree relatives of surgically proved otosclerotic patients.

**Methods:** In a cross sectional study among 43 surgically proved otosclerotic patients, positive family history of OS was determined. In suspicious cases, physical exam, tuning fork tests, and audiometry were performed. We also reviewed medical sheaths of admitted OS patients from 1995 to 2003 in Isfahan university hospitals for family history of OS.

**Results:** Among 15 otosclerotic patients (34.88%) with positive family history of OS, 14 individuals of their first and second degree relatives had clinical and audiometric otosclerosis. Among 85 medical record sheaths of otosclerotic patients, 12 patients (27.9 %) had positive family history for OS.

**Conclusions:** The incidence of clinical and audiometric otosclerosis among first and second degree relatives of OS patients is nearly similar to the rate of positive family history of our patients, which is relatively lower than other studies.

**Key words:** Otosclerosis, family history, first and second degree relatives, hearing loss

JRMS 2006; 11(2); 116-118

Otosclerosis (OS) "hardening of ear" is an autosomal dominant disease from endochondrial layer of temporal bone<sup>1,2,3</sup>. It may cause a conductive, mixed, or occasionally a purely sensorineural hearing loss<sup>2, 3, 4</sup>. Among white adults, OS is the most common cause of hereditary hearing loss<sup>1, 5</sup> and there is a considerable racial predisposition in the prevalence of OS<sup>2, 6</sup>. Family history is positive in 49-58 % of the cases<sup>1,4</sup>, but in Iranian patients, its incidence is unknown. Our aim was to determine family history of OS in operated patients for OS, and show its relation to clinical and audiometric OS in suspicious individuals.

(hearing loss or use of hearing aids, tinnitus) or diagnosis of OS in the first and second degree relatives (parents, brothers, sisters, children, uncle and aunts). They were operated in ENT department of Al-zahra and Kashani hospitals, Isfahan, Iran. Suspicious individuals (15 cases) invited for additional studies, including physical exams, Tuning fork tests and audiometries.

We also performed a retrospective study on medical record sheaths of 85 patients underwent OS surgery in these hospitals from 1995 to 2003, and reviewed the family history of its symptoms.

**Subjects and Methods**

In a cross sectional study, 43 otosclerotic patients were asked about otosclerotic symptoms

**Results**

Among 1st and 2nd degree relatives of 43 surgically proved otosclerotic patients (22 female and 21 male with mean age of 32.8 years), 15

Supported by: Research Department, Isfahan University of Medical Sciences, Grant No: 80065

\* Assistant Professor, Department of Otolaryngology, Isfahan University of Medical Sciences, Isfahan, Iran

\*\* Resident of Otolaryngology, Isfahan University of Medical Sciences, Isfahan, Iran

Correspondence to: Dr. Saeed Soheilipour, E-mail: soheilipour@med.mui.ac.ir

individuals from 15 separate families (34.88% of our patients) were suspicious for OS; chiefly suffering from hearing loss and tinnitus, that 14 of them had clinical and audiometrical otosclerosis (Table 1).

From these 14 new patients, 5 individuals underwent middle ear exploration, and otosclerosis was surgically proved in them.

We reviewed medical record sheaths of 85 OS patients (37 females, 48 males, mean age at the time of surgery; 33.4 years) and 42 records

lacked any information about family history of the patients. From 43 remaining records, 31 cases had negative family histories, and 12 patients (27.9%) had positive family histories for symptoms of OS (5 in mothers, 4 in sisters, 4 in fathers).

The chief complaints of these individuals were mainly hearing loss (88%), tinnitus (72%), and occasionally vertigo (25%). The rate of positive history in two parts of our study was approximately different (34.88% vs. 27.9%) ( $P > 0.1$ ).

**Table 1.** Suspicious otosclerosis among first and second degree relatives of otosclerotic patients

Relatives	Mother	Father	Sister	Brother	Aunts, Uncles, others	Total
Suspicious individuals	3 (21.42%)	1 (7.1%)	4 (28.57%)	2 (14.28%)	4 (28.57%)	14 (100%)

## Discussion

Otosclerosis is a disease of otic capsule of temporal bone<sup>1,2</sup>, and is the most common cause of conductive hearing loss (HL) among the ages of 15 to 50 years<sup>4</sup>, and may cause mixed or occasionally, a pure sensorineural HL<sup>2,3,4</sup>.

In some areas such as India, it is the third most common cause of conductive HL after chronic otitis media and after otitis media with effusion<sup>8</sup>. The peak age of onset of hearing loss is between 20 and 30 years, which 90% of the patients present by age 50<sup>2,5</sup>, with the mean age of 41-52 years at the time of surgery<sup>7,10</sup>. Disease process has a female: male predominance of 2 to 1<sup>1,2,4</sup>, however newer studies on the incidence of histological OS on temporal bones have not shown significant differences between male and female subjects<sup>6</sup>.

Racial predisposition in the prevalence of OS is considerable: 8-12% of white population and 7.3 - 10.3% of temporal bones of white males

and females have had histological OS<sup>2,4</sup>. In white population, the prevalence of clinical OS is 0.5-2%. In Caucasians, the prevalence of HL caused by OS is 0.2 - 1%<sup>5</sup>. The prevalence of OS varies with race and its expression<sup>1,2,6</sup>. It is a hereditary disease and is the most common cause of hereditary HL among white adults<sup>1,4,5</sup>. Indeed, it is an autosomal dominant disease with variable penetrance (from 25 to 40 percent) and expression<sup>4,5</sup>. Family history is positive in 49-58% of OS patients and in 70% of selective populations<sup>1-4</sup>.

The etiology of OS has not determined yet, but genetic factors, autoimmunity, measles virus, and fluoride content of drinking water sources have been proposed as etiological factors<sup>1,3,8,9</sup>.

The prevalence of clinical or histopathological OS has not been determined in Iran. However, we frequently encounter with otosclerotic patients in our daily practice, and some familial and geographical assembling patterns exist.

In our study, 27.9 and 32.5% of the patients had positive familial history, and nearly all of suspicious individuals (according to family history taking from otosclerotic patients) proved clinically and audiometrically OS. This emphasizes importance of precise family history taking from otosclerotic patients.

It is obvious that some individuals of these families have sub clinical or mild diseases, and true rate of positive family history will be much greater when we search actively all members of first and second degree relatives of otosclerotic patients for these individuals by physical examination and audiometrical evaluation.

Such active screening program will reveal (when we consider that in our country) many of involved patients do not seek an ENT specialist, because of cultural and economical reasons.

The incidence of clinical and audiometrical OS among the first and second degree relatives of our otosclerotic patients is approximately similar to the rate of positive family of the patients, but relatively lower than studies. Family history taking from otosclerotic patients and screening for suspicious individuals are important, and we suggest an active screening study in the relatives of otosclerotic patients.

## References

1. Glasscock III M.E, Gufya A.J, "Glasscock – Shambaugh Surgery Of The Ear . 5 th ed. BC. Decker 2003 PP: 517-21.
2. John W. House, Calhoun D. Cunningham, III; *Otosclerosis*. in: Cumming C.W. et al. *Otolaryngology , Head & Neck Surgery* 2. 4 th Edition Elsevier Mosby 2005 pp: 3562-72.
3. Snow JB, Ballenger JJ. *Ballenger otorhinolaryngology. Head and Neck Surgery*. 16 th ed. BC Decker 2003; PP: 317-8.
4. Lee k. J, "Essential otolaryngology Head and Neck Surgery "8 th ed., MC Graw Hill, 2003 PP: 518- 519 and 1023.
5. Spiros Manolidis Alford RL, Smith RJ, Ball C, Manolidis L. "Do the Genes that cause otosclerosis reduce susceptibility to Otitis Media?" *Otology & Neurotology* "2003; 24(6): 868-71.
6. Iwao Ohtani, Baba Y, Suzuki T, Suzuki C, Kano M. "Why is Otosclerosis of low prevalence in Japanese; " *Otology & Neurotology* "2003; 24(3): 377-81.
7. Denis Ayache, Corre A, Van Proyen S, Elbaz P. "Surgical treatment of otosclerosis in Elderly patients "; *Otolaryngology – Head and Neck surgery* 2003; 124(6): 674 – 7. 87.
8. Ramesh C. Deka, *Invited comment on. Iwao Ohtani, Baba Y, Suzuki T, Suzuki C, Kano M. "Why is Otosclerosis of low prevalence in Japanese; "Otology & Neurotology* "2003; 24(3): 377-81.
9. Kevin T Kavanagh , 2004 ; [www.entusa.com / Stapedectomy. htm-](http://www.entusa.com/Stapedectomy.htm); *Stapedectomy* 2004.
10. Esquivel CR, Mamikogla B, Weit R.. *Long Term results of small Fenestra Stapedectomy. Laryngoscope* 2002; 112: 1338-41 .