Otosclerosis

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Abstract. Otosclerosis. Otosclerosis is the most common cause of progressive conductive and mixed hearing loss.1-3 Its diagnosis is usually unproblematic and based on the combination of normal otoscopy, a typical audiogram, and absence of stapedial reflexes. In atypical cases, investigation with high-resolution imaging is recommended. In case of conductive hearing loss and depending on the severity of the symptoms, three treatments are available.2 Watchful waiting is advised for patients with very slight hearing losses without social discomfort, and a hearing aid can be provided to patients with hearing problems but unwilling to undergo surgery. Surgery is the treatment of choice for the conductive component and is preferred because of its high success rate and low complication rate. Because surgery is always elective, in-depth counselling of the patient is important.1,2

Definition, Etiology, Epidemiology

Otosclerosis is the most common cause of stapes fixation.1,2 It is primarily a disease of the bone that encases the cochlea and labyrinth. Histologically, otosclerosis is characterized by progressive focal dysplasia with destruction (the so-called “otospongiosis” stage), remodelling, and finally sclerosis of the endochondral bone of the labyrinthine capsule (the so-called otosclerosis stage).2 The disease mostly starts in the anterior part of the oval window, the so-called fissula ante fenestram and extends to the annular ligament and stapes, where it causes bony ankylosis of the stapes, which results in increased stiffness of the ossicular chain and conductive hearing loss. A thick ossification of the entire oval window, also known as obliterator otosclerosis, is accompanied by severe conductive hearing loss. The inner ear structures are often uninvolved, but sometimes, advanced otosclerosis of the labyrinthine capsule occurs without conductive hearing loss. In those cases, progressive, degenerative changes in the inner ear occur, with atrophy of the spiral ligament, and they cause an additional sensorineural hearing loss.

In rare cases, the otosclerotic foci affect wide areas of the petrous bone. Patients with so called “malignant” otosclerosis become progressively deaf.2 The typical natural history of otosclerosis is characterized by a slow progressive conductive hearing loss. When the cochlear otic capsule is also involved, a concomitant sensorineural hearing loss develops. The disorder is usually bilateral, but the progression rate is different for the two ears, and otosclerosis may present as a unilateral conductive or mixed hearing loss.

The onset of the hearing loss mostly occurs after the third decade. Less than 3% of new cases are diagnosed during childhood.2 Females appear to be affected at twice the rate of males. During pregnancy, otosclerosis frequently becomes apparent or aggravates, which suggests that female hormones play an important role in the pathophysiology. Up until now, however, no hard evidence could prove increased incidence of otosclerosis in subjects that take oral contraceptives.2,4

A definite racial predisposition exists: otosclerosis is more common in Caucasians, has a lower prevalence among Asians, and is rare among native Americans. Otosclerosis is a disorder in which both genetic and environmental etiological factors are involved.2 A limited number of large families showing an autosomal dominant inheritance pattern for otosclerosis have been described. Most subjects with otosclerosis, however, occur in small families with unclear inheritance pattern or are sporadic, which suggests a multifactorial aetiology for otosclerosis. Among Caucasians, clinical otosclerosis has a reported prevalence of 0.3% and is the most common cause of conductive hearing impairment. Histological
otosclerosis even has a prevalence of 3.5% among white adults and affects usually both temporal bones symmetrically.

Otosclerosis represents a heterogeneous group of genetic diseases in which genes are involved in regulating bone homeostasis of the otic capsule. It has been hypothesized that bone dysplasia in otosclerosis results from the lack of physiologic inhibition of bone turnover in the otic capsule due to environmental and genetic factors. Many environmental factors have been implicated in the aetiology of otosclerosis, including infectious causes such as measles virus, hormones (related to puberty, pregnancy and menopause), and nutritional factors (fluoride intake).

Large autosomal dominant otosclerosis families have been analysed for gene identification studies, but the first gene responsible for otosclerosis has yet to be cloned. However, five chromosome loci, OTSC1 - OTSC5, have been identified, supporting the hypothesis of genetic heterogeneity. OTSC1 was mapped to chromosome 15q25-q26 in an Indian family in which hearing loss began in childhood. The OTSC2 locus was mapped to a 16 cM region on chromosome 7 (7q34-36) in a large Belgian family detected by our institution (Sint-Augustinus Hospital). More recently, the OTSC3 locus was mapped on chromosome 6 in a large Cypriot family (6p21.3-22.3). The OTSC3 locus covers the HLA region, consistent with reported associations between HLA-A/HLA-B antigens and otosclerosis. OTSC4 was recently found in an Israeli family. A fifth locus for otosclerosis, OTSC5, was mapped to chromosome some 3q22-24 in a large Dutch family. Note that genetic heterogeneity has also been demonstrated for nonsyndromic sensorineural hearing loss.

Diagnosis Management
The diagnosis of otosclerotic hearing loss is usually unproblematic.12,6

A positive family history is found in about 50% to 60% of the cases. Clinical bilateral affection is common (85%-90%). Tinnitus, which is mostly low-pitched (~75%), is also a common symptom of otosclerosis. The exact mechanism underlying tinnitus in otosclerosis, however, is unclear. Tinnitus is most common in those patients with severe hearing loss. It is frequently encountered in the older age group and in those with an early age of onset and cochlear involvement.

Many patients (20%-78%) with otosclerosis have increased understanding of speech during a cocktail party, the so called paracusis of Willis. This phenomenon occurs because the conductive hearing loss attenuates the background noise and renders the dynamic range of the ear at the level of the speaker’s voice, thus effectively increasing the signal to noise ratio. Vestibular disturbance and postural instability are present in more than a quarter of the patients with otosclerosis (25%-55%) and tend to be rather mild, except in co-existing endolymphatic hydrops.4,6

Three types have been described:

1. Periods of unsteadiness and dysequilibrium (20 min – 6 h) with normal caloric response, and without nystagmus.
2. Periods of postural instability.
3. Menieriform attacks with increased tinnitus, fluctuating hearing loss, caloric tests are normal or show hyporeflexia.

Otoscopcy reveals a normal ear-drum with an air filled middle ear.

The sign of Swartze, which refers to a reddish blush on the promontory, is a rare finding and reflects abnormal vascular shunts between otosclerotic foci and the vessels of the promontory.

Tuning fork tests are very helpful for evaluating a patient with otosclerosis. During Weber’s test, sound lateralizes to the ear with the greatest degree of conductive loss and during Rinne’s test, sound will be heard louder when delivered on the mastoid tip compared to delivering via the ear canal.1,2,6

Audiometric testing in otosclerosis reveals a conductive or mixed hearing loss. In its early stages, the conductive loss tends to be confined to lower frequencies (Figure 1A). In advanced stages, conductive loss also occurs at higher frequencies (Figure 1B) and a perceptive component may also appear (Figure 1C). A conductive loss of about 40 dB in the low frequencies with a reduction of the gap towards 2 kHz is typical, because stapes fixation reduces the elasticity of the ossicular chain. An interesting finding in otosclerosis is the deterioration of bone conduction thresholds at middle to high frequencies, which sometimes disappears after successful surgery: the so-called Carhart notch, which can reach up to 25 dB at 2 kHz (Figure 1D). A possible cause is the absence of middle ear resonance, in humans at 2000 Hz, together with reduced perilymph oscillation due to the immobile footplate.2
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Speech audiometry shows a normal increase in speech discrimination when sound intensity increases. Reduced speech discrimination indicates inner ear involvement, and should be further investigated.

Tympanometry is generally normal, but sometimes a diminished compliance (a smaller peak) is observed.2,4,6 The stapedial reflex is absent and this sign is particularly interesting in unilateral forms.

The “on-off” effect, which refers to a reduction of the acoustic impedance at onset and offset of the sound stimulus, is a poorly understood finding at early stage of stapes fixation.

High-resolution computed tomography (HRCT) is currently the radiologic method of choice in assessing the labyrinthine windows and otic capsule. On HRCT images, otosclerotic foci are visualized as hypodense or radiolucent foci. Most common HRCT findings in surgically confirmed otosclerosis are foci anterior to the oval window (in the fissula ante fenestram), pericochlear lucency, and foci in the footplate of the stapes. Absence of foci on HRCT images does not rule out stapes fixation by a sclerotic focus. In severe mixed hearing loss, a peri-cochlear demineralisation can be observed as a characteristic radiolucency giving a “halo” appearance to the otic capsule.2,4,6

Although fenestral otosclerosis remains largely a clinical diagnosis, HRCT is recommended in atypical cases (e.g. children, atypical audiogram, concomitant vertigo) in order to exclude inner ear deformities (e.g. large vestibular aqueduct, inner ear dysplasias...). A large patent cochlear aqueduct or no partition between the inner ear and the fundus of the internal auditory canal should alert the surgeon for a potential perilymph gusher. HRCT is also recommended in revision cases and in far advanced cases with (sub)total deafness especially if cochlear implantation is planned.

MRI is recommended in asymmetric audiograms to exclude concomitant retrocochlear disease (e.g. a vestibular schwannoma). On MR images of ears with otosclerosis, slight-to-marked contrast enhancement can be seen, which are interpreted as inflammatory hypervascularization.2

Some otologic diseases can mimic the clinical picture of otosclerosis, and their presence only becomes apparent during an exploratory tympanotomy.4,6

- Sequelae of otitis media with intact eardrum: ossicular discontinuity, tympanosclerosis
- Post-traumatic stapes fixation
- Malleus head fixation
- Minor malformations of the middle ear: monopodic stapes, aplasia of stapes superstructure, footplate ossification with co-existing aplasia of the annular ligament, ossification of the stapedial tendon, persistence of a stapedial artery
- Abnormal perilymph pressure
- Paget’s disease
- Osteogenesis imperfecta: patients with blue sclerae should be suspected and questioned about bone fractures in the past.

Therapeutic Management

Conservative therapy

- No treatment is advised when air conduction thresholds are lower than 30 dB HL.
- In patients with moderate to severe hearing loss, and unwilling or unable to undergo surgery, hearing aids often yield good results when cochlear function is well preserved.1,4
Osseo-integrated bone-anchored hearing aids have occasionally been used as an alternative for patients with otosclerosis with very large air-bone gaps and who did not benefit sufficiently from traditional hearing aids.\textsuperscript{2}

In cases of cochlear otosclerosis with progressive sensorineural hearing loss one may also consider the use of sodium fluoride, calcium and vitamin D. The beneficial effects of sodium fluoride are best documented. The rationale of its use is based on epidemiological data and randomized trials. An increased prevalence of stapedial fixation in areas where the levels of fluoride are low has been observed. Several double-blind prospective studies have confirmed that sodium fluoride does stabilize hearing in sensorineural hearing loss, while the controls who did not receive treatment continue to experience a deterioration in their hearing. Fluoride probably transforms active otospongiotic lesions into more dense inactive otosclerotic lesions.\textsuperscript{2}

The daily recommended dose of fluoride is 50 mg. When the hearing stabilizes and radiological evidence of recalcification exists, some authors would give a maintenance dose of 25 mg. Others claim that prolonged fluoride administration does not seem to be superior to a shorter treatment period (1-2 years).\textsuperscript{2,4}

**Surgery**

Stapes surgery should be performed by surgeons who have special interest in otology, and have sufficient experience in middle ear surgery. Performing stapes surgery from time to time should be avoided. In approximately 90\% of the cases, surgery results in a dramatic and prolonged hearing gain with closure of the air-bone gap to within 10 dB.\textsuperscript{1,2,4}

Stapes surgery is most appreciated by patients who experience hearing problems in everyday life. This is mostly the case when Rinne’s test is “negative” and air conduction thresholds are at 30 dB HL or worse.\textsuperscript{2,6}

In bilateral cases, the operation should be carried out on the ear with the highest degree of hearing loss.\textsuperscript{6}

If the average bone conduction thresholds are more than 30 dB lower than those at the contralateral better hearing ear, the patient will most probably notice little or no hearing improvement after surgery.\textsuperscript{1,2,6}

When the air-bone gap is small, the potential risk of an intervention should be considered.

Local or general anaesthesia can be used according to the patient’s and the surgeon’s preference.

The small hole technique (stapedotomy) is preferred above the stapedectomy technique which is associated with a slightly higher risk for inner ear damage. Micro-instruments, microdrill or laser are used to make the calibrated hole. Laser stapes surgery has, to a certain extent, reduced technical difficulties. It must be emphasized, however, that the available techniques (different lasers and microdrills) are only as good as the surgeons that handle them.\textsuperscript{1,2,4}

The diameter of the stapedial prostheses varies between 0.4 mm and 0.6 mm. No significant difference in hearing results are found for prostheses as long as the diameter is comprised between 0.4 mm and 0.6 mm.\textsuperscript{1,2}

In profound hearing loss due to cochlear otosclerosis a cochlear implant (with or without cochlear drill out) may be warranted.\textsuperscript{2,6}

**Indications for surgery**

The patient should be in reasonably good health, especially if general anaesthesia is planned.\textsuperscript{1}

The age of the patient is not a factor in the decision on surgery but one should be cautious in children (exclude inner ear malformations) and in elderly patients. In children, it is prudent to fit hearing aids first and not carry out the operation until both the affected adolescent and also his/her parents insist on the operation. A certain degree of caution may also be indicated in elderly patients. Individual studies (not confirmed by others), suggest that elderly patients develop a high frequency hearing loss postoperatively more often than younger patients.\textsuperscript{2}

The worst ear, based on the patient’s statement and not necessarily on the audiogram, should be chosen for surgery.\textsuperscript{1,2,4,6}

Tuning fork tests should always be used to confirm the audiometric findings. If bone conduction is heard louder than air conduction with a 512 or 1024 tuning fork, the individual is a suitable candidate for surgery.\textsuperscript{1,2}
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- The minimal average air conduction loss should be at least 30 dB according to some and at least 40 dB according to others. The minimum air-bone gap should be 15 dB, as averaged in speech frequencies for some, others would not operate before a 20 dB air-bone gap. More importantly the patient should experience his hearing loss as disturbing in everyday life before surgery is offered.

- Indications for surgery are essentially the same whether the hearing loss is unilateral or bilateral. If for binaural disease, the stapedotomy at the first ear yields good hearing gain, the second ear can also be operated after a waiting period of about six months to one year, because useful binaural hearing is always preferred by patients. Useful binaural hearing enables patients to localize sound, and hear a talker better in a noisy environment, and this occurs when average air conduction thresholds of the two ears are no more than 30 dB apart.

- In unilateral otosclerosis, the postoperative hearing thresholds often do not reach the thresholds of the completely healthy ear, so that subjective hearing gain is less spectacular.

- Stapes surgery must in theory not be carried out in patients with only one functional ear because of the small possibility (about 1%) of sensorineural loss. The exception would be in a patient fully informed of the risk and aware that in this case the only alternative would be a cochlear implantation. Few experienced surgeons, however, are willing to perform this intervention because of the medico-legal implications.

- Tinnitus is not a contra-indication for stapedotomy, and low-tone tinnitus may disappear after a well-performed stapedectomy.

- According to a large study at the House institute, stapedectomy does not increase the risk of inner ear barotraumas in scuba-divers and sky-divers. These activities may be pursued with relative safety following stapes surgery, provided adequate Eustachian tube function has been established.

- In high performance pilots, Katzav et al. found that flight status can be reinstated without endangering flight safety 3 months after stapes surgery.

Contra-indications for surgery

- Poor physical health.
- Balance problems, such as active endolymphatic hydrops or a fluctuating hearing loss.
- Pre-existing tympanic membrane perforation.
- Active external or middle ear infection.
- Inadequate air-bone gap confirmed by an audiogram and the 512 Hz tuning fork test.
- Inner ear malformations as visible on HRCT.

Decision algorithm

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Progressive hearing loss with normal otoscopy

Tuning fork tests

Audiogram: moderate to severe conductive or mixed loss

Tymanometry
- Type A curve
- No stapedial reflexes

Typical presentation

Atypical presentation

CT scan

Signs of otosclerosis

Inner ear malformation

Hearing aid

Exploratory tympanotomy

Confirmation by stapes palpation + Stapedotomy
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Counselling of candidates for surgery

Stapedectomy is an elective operation. In-depth counselling of the patient and providing preoperative information is hence of utmost importance. The mechanics of the hearing loss should be explained in detail, preferably with a suitable illustration.

Patients should be assured that it is very unlikely that they will become totally deaf. Patients who are suitable for stapes surgery should be told that they have the option of wearing hearing aids, and if they have any doubt about the decision to have surgery, they should be encouraged to have a trial period with hearing aids unless they are already wearing them.

Hearing improvement is strongly determined by the preoperative bone conduction level, and the patient must be aware of the degree of improvement that can be expected. The expected result as well as all possible risks, such as further or even total hearing loss, taste disturbance, dizziness, the effect on tinnitus, and the very small possibility of a partial or total facial paralysis, should be clearly explained.

The main problem of stapes surgery is the possible development of postoperative deafness, of which the incidence is estimated at 1%. The patient should be clearly informed about this serious complication.

Slight postoperative symptoms of vertigo occur fairly often in the first days after stapedectomy but less after stapedotomy. Tinnitus that is present preoperatively will not necessarily disappear, but often becomes less pronounced or even disappears altogether after surgery, probably because it is masked by the improved hearing. When hearing is worse after stapedectomy, tinnitus will likely increase.

The patient should be also informed that in rare cases a revision operation might be necessary. Persistence of conductive loss for at least 3-4 months after surgery may indicate a dislodged prosthesis, fixation of the malleus or incus, or necrosis of the distal end of the incus to which the prosthesis was attached. Re-exploration may be warranted, though results with revision surgery are less successful than with primary surgery.

Temporary dysfunction of the chorda tympani may occur in 15% to 30% but only in about 1% a slight dysgeusia persists permanently.

Iatrogenic lesions of the facial nerve after stapedectomy have occasionally been reported, mostly in cases with an abnormal anatomy of the nerve in the middle ear. Sometimes secondary, probably inflammatory, facial nerve paralysis occurs after a latent period of a few days, and is always reversible within a few weeks.

Addendum
Information to patients
(Adapted from the information sheet given at the University of Minnesota)

What is otosclerosis?

Otosclerosis is an abnormal, microscopic growth of bone in the walls of the inner ear and causes the stapes bone to become frozen in place or “fixed” in the oval window. Normally the stapes, the smallest bone in the body, vibrates freely to allow the transmission of sound into the inner ear. When it becomes cemented to the surrounding bone it prevents sound waves from reaching the inner ear fluids, and hearing is impaired.

Occasionally the otosclerotic bone involves other structures of the inner ear so that, in addition to preventing sound from entering the ear, it causes a distortion or difficulty in understanding the speech of others, regardless of how loudly they talk. In such cases there is not only the “conductive” deafness already discussed, whereby sound waves are obstructed in reaching the inner ear, but in addition, “sensorineural” or “nerve” deafness, in which the function of the inner ear itself has been impaired.

Otosclerosis affects the ears only and not other parts of the body. When this condition is present, both ears are usually involved. It occurs more frequently in women and usually begins in young adulthood. Although otosclerosis tends to run in families, it does so irregularly; parents with otosclerosis do not necessarily transmit it to their offspring.

How is it treated?

There is no known medicine available for treating the stapes fixation due to otosclerosis. Although a hearing aid can be worn successfully by most patients, they prefer natural hearing if that is possible. Surgery has been found to be the most effective method of managing the stapes fixation due to otosclerosis.

The stapes operation is done using an operating microscope. The surgeon will first fold forward approximately one-half of the ear drum so that he can reach the area where the stapes is located. Part of the stapes is then removed with
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fine instruments or/and the laser. A hole is made in the remaining footplate of the stapes. A “prosthesis” about 4 mm to 5 mm in length and between 0.4 mm and 0.6 mm in diameter then is introduced and secured in place to bridge the gap created by removal of the stapes.

Can I have the operation and what are my chances of its success?

An examination by an ear specialist, including a hearing test, is necessary to determine if you are a candidate for the operation. As you can imagine, there are many different causes of deafness; and in fact, not even all patients with otosclerosis are candidates for stapes surgery.

In some cases further imaging by CT scan can be recommended. The chances of obtaining a good result from this operation are about 90%. This means that about 9 out of 10 patients will get an improvement of hearing up to the level at which their inner ear is capable of functioning. If the inner ear functions normally, then normal hearing can be restored. Approximately 7% of all patients have only partial recovery of hearing and 2% remain at the same level as before surgery. The main risk is a 1% chance of developing inner ear hearing loss following the procedure due to factors as yet not entirely understood. For this reason, only one ear is operated upon at a time, and the worst ear is always done first.

What should I be aware of before the operation?

If you would catch a cold one week or less prior to the date scheduled for your operation, you should report this to your doctor. For this operation a local anaesthetic or a general anaesthesia can be used depending on the patient’s and surgeon’s preference.

Because the stapes is so small, the operation is performed with the aid of a microscope. Under local anaesthesia you may notice improved hearing while still in surgery and notice a decrease later. Do not become alarmed, as this is due to ear packing, swelling, and fluid build-up from surgery. It may be several weeks before the full effect of surgery can be determined, as far as hearing is concerned. You may have occasional periods of dizziness during the first few days.

What can I expect after the operation?

The evening after the operation you should lie quietly on the unoperated ear. Do not be alarmed if you have some dizziness for the first few days after the operation. The surgery usually takes about one hour.

Please do not:

- Blow your nose
- Remove any packing from your ear

Discharge instructions:

1. Do not get water in the ear
2. No strenuous exercise
3. Do not remove any packing
4. Notify your doctor of fever greater than 37°C, excessive pain, excessive drainage, or drainage that has an odour

Some possible side effects

Stapedectomy is a well established and proven operative procedure with a 90% or greater success rate. Potential but unusual side effects include:

1. Change in sense of taste on the same side of the tongue
2. Vertigo: usually resolves spontaneously
3. Lack of hearing improvement
4. Perforation of the tympanic membrane
5. No change in tinnitus
6. Intolerance of very loud noises

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References


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CME questions

1. Epidemiology: find the false statement.
   A – Otosclerosis is usually characterized by a slow progressive conductive hearing loss
   B – Females are affected more often
   C – Oral contraceptives causes a higher incidence of otosclerosis
   D – The prevalence is lower among Asians
   E – Has a histological prevalence which is ten fold the clinical prevalence

2. Diagnosis: find the false statement.
   A – A positive family history is found in about half of the patients
   B – Vestibular disorders do not occur with otosclerosis
   C – During Weber’s test sound lateralizes to the ear with the greatest air-bone gap
   D – The on-off effect as recorded during tympanometry with stapedial reflex testing is an interesting finding of early stage otosclerosis
   E – Ct scanning cannot always detect an otosclerotic focus confirmed by surgery

3. Surgery: which condition can also be operated by a stapedotomy?
   A – Abnormal high perilymph pressure
   B – Ossicular discontinuity
   C – Malleus head fixation
   D – Congenital absence of oval window
   E – Osteogenesis imperfecta

4. Which treatment modality is never used today for otosclerosis?
   A – Cochlear implantation
   B – Stapes surgery
   C – Sodium fluoride
   D – Hearing aid
   E – Middle fossa approach to decompress the internal auditory canal

5. Surgery should not be recommended in patients when:
   A – The hearing loss becomes difficult in everyday life
   B – Audiometric and tuning fork tests are inconsistent
   C – The worse hearing ear is considered
   D – Average bone conduction is better than 30 dB
   E – CT scanning does not show an otosclerotic focus

6. Which is not a clear contra-indication?
   A – Patient older than 70 years
   B – Poor physical health
   C – Active middle ear inflammation
   D – Inner ear malformation as visible on high resolution CT imaging
   E – Active Ménière’s disease

Answers: 1C; 2B; 3E; 4E; 5B; 6A