Otosclerosis: a review of aetiology, management and outcomes

Otosclerosis is an autosomal dominant condition affecting the temporal bone. It presents predominantly with deafness in a young population. This review looks at the aetiological theories, present treatment strategies and surgical outcomes of this condition.

Otosclerosis is a term used to describe a primary osseous dysplasia of the human temporal bone derived from the embryonic otic capsule. Its inheritance is autosomal dominant with incomplete inheritance and expressivity.

Controversy exists regarding its exact aetiology and concerning treatment strategies, although surgery is widely considered the optimum management despite its potential complications. This review looks at the aetiological theories, present treatment strategies and surgical outcomes of this condition.

History

From the pathological viewpoint, the term 'otospongiosis' more accurately refers to the active and vascular stage of the process although the term otosclerosis is presently widely adopted to define the pathological changes.

Initially, surgical management consisted of fenestrating the lateral semicircular canal. Lempert (1938) performed this in the 1930s. John Shea performed the first stapedectomy in 1958 by removing the stapes and placing a polyethylene graft between the incus and a vein graft placed over the oval window. Because of the large backlog of patients that existed at this time, a few surgeons such as Shea and Marquet obtained extensive experience.

Prevalence

The prevalence of otosclerosis is thought to be around 0.5–2% (Browning and Gatehouse, 1992). The gender incidence is equal and patients typically present between the ages of 20 and 40 years. It is felt that recently the numbers of patients having surgery is decreasing because:

- Patients now presenting represent the new cases
- Improvements in hearing aid technology have resulted in more patients accepting conservative management
- Fluoridation of water supplies may have affected the incidence.

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In the United States an analysis of trends of surgical management has shown the incidence declining over the past 30 years (Vrabec and Coker, 2004).

Aetiology

Despite intensive research, the precise aetiology of otosclerosis remains unclear. Numerous studies have tried to identify potential aetiological factors and the theories that have found the greatest support include: genetic factors, viral aetiology, immune system disorders and connective tissue disorders.

Genetic factors

Otosclerosis has an autosomal dominant inheritance with incomplete penetrance estimated to be about 40% (Menger and Tange, 2003). Chromosomal linkage studies in multi-generational families have identified genes located on chromosomes 15q25-q26 (Tomek et al, 1998), 7q34 36 (Van Den Bogaert et al, 2001) and COL1A1 that are associated with clinical otosclerosis.

Viral aetiology

The presumed aetiology is retrograde infection of the middle ear mucosa via the eustachian tube and subsequent invasion of the bony labyrinth via either lymphatic or pericapillary spaces. McKenna and Mills (1989) demonstrated measles virus antigens with several different monoclonal antibodies in active otosclerotic foci and Niedermeyer and Arnold (1995) additionally showed immunoglobulin IgG anti-measles virus antibodies in the perilymph of otosclerotic subjects. However, conversely Grayeli et al (2000) did not find evidence of measles virus in stapes samples or bone cell cultures.

Immune system disorders

Elevated antibody levels to type II collagen and to double- and single-stranded DNA in sera have been found in patients with otosclerosis, leading to the hypothesis that an autoimmune reaction to type II collagen could be an aetiological factor (Yoo, 1984).

However, several studies have failed to show a significant association with the HLA A, B or C antigens in subjects with otosclerosis (Chobaut et al, 1982; Pedersen et al, 1983).

Connective tissue disorders

Pedersen et al (1984) found a reduction of dermal thickness and minor degenerative changes of elastic fibres in otosclerotic patients but Thalmann et al (1987) found no difference in urinary glycosaminoglycans or in synthesis and secretion of collagen in skin fibroblast cultures of patients and controls. Similarities have also been proposed with osteogenesis imperfecta which gives otosclerotic-like lesions in the temporal bone. However, osteogenesis imperfecta has an earlier onset, a more severe middle ear involvement and a higher incidence of hearing loss.

Pathology

Pathologically mature lamellar bone is removed by osteoclasts, and replaced by osteoblasts with bone of greater thickness, cellularity and vascularity. This new bone is laid down at specific sites the commonest of which is an area between the cochlea and vestibule, just anterior to the footplate of the stapes called the fissula ante fenestram. Other sites include the fissula post fenestram, semicircular canals, round window, base of styloid process and petrosquamous suture.

Clinical presentation

- Deafness is the primary symptom. It is most often noticed between the ages of 20 and 40 years. Patients may report hearing better in noisy environments because sound is raised above normal conversational levels allowing better speech discrimination – a phenomenon called 'paracusis Willisi'. The disease process may accelerate during pregnancy. Bilateral hearing loss occurs in about 85% of cases
- Tinnitus is a common symptom but is non-specific as it can occur with any ear disease
- Vertigo is unusual.

There is a family history in about 50% of patients.

Clinical examination is usually unremarkable as the tympanic membrane is normal. However, in some cases a pink tinge to the tympanic membrane imparted from dilated blood vessels on the promontory (medial wall of the middle ear) may be seen (Schwartz sign). Tuning fork tests may confirm a conductive loss with Rinnes test being negative (bone conduction greater than air conduction) and Webers test lateralizing to the affected ear.

Cochlea otosclerosis

In some patients there is a progressive sensorineural hearing loss in early adult life. This is often associated with a strong family history and examination may show the Schwartz sign.

Investigations

Pure tone audiometry

This is the single most helpful investigation and must be performed in all cases. It will show a conductive hearing loss initially most marked in the low frequencies as a result of fixation of the stapes. With disease progression the hearing loss becomes more marked in the mid and lower frequencies and increasing cochlear involvement results in progressive sensorineural loss.

A particular feature of the bone conduction curve of the audiogram is a dip (elevation of the threshold of 10 dB or more) at 2000 Hz known as Cahart's notch. It is unclear why this occurs. It is an audiological phenomenon and can sometimes be shown postoperatively by its absence in successful cases. There may also be over closure of the air–bone gap postoperatively where the postoperative air conduction is better than the preoperative bone conduction.

Speech audiometry

Not all clinicians perform this test. It is essentially a monaural test where groups of words are presented in quiet conditions over headphones and scored as the percentage correct at various sound intensities. A maximum speech discrimination of over 70% should be obtained.

Impedance audiometry

This test is not considered helpful in the diagnosis or management of otosclerosis.

Vestibular function tests

These tests are not considered necessary for a first operation. However, in second side surgery some clinicians perform them as they may indicate whether vestibular function is normal on the first operated ear. If it is not, then second side surgery presents a risk of bilateral vestibular hypofunction, which needs to be discussed in the consent process.

Imaging

Although otosclerotic foci can be seen with computed tomography (CT) scanning the clinical advantage is limited and it is rarely performed except in cases where cochlear implantation is being considered.

Treatment

The treatment of otosclerosis depends on the patient's symptoms. The options for treatment are: no treatment, medical treatment, hearing aid or surgery.

No treatment

After appropriate counselling about the condition and assessment of its disability a patient may decide, particularly if the hearing is well preserved, that they do not want further treatment but having been made aware of the options may opt to return at a later date to reconsider treatment.

Medical treatment

Fluorine has been advocated as a potential medical treatment for otosclerosis. It is thought to inhibit proteolytic enzymes involved in the pathological process (Causse et

al, 1980). Research into its efficacy has found that a sodium fluoride intake of 1–3 mg daily cannot prevent the development of otosclerosis in a low-fluoride area (Vartiainen and Vartiainen, 1997). A similar dose does not have a significant effect on hearing impairment after stapedectomy. There is conflicting evidence, however, showing a beneficial effect on non-operated otosclerotic ears (Vartiainen et al, 1994).

Fluorine can also cause abdominal pain as a side effect. Hence owing to the conflicting evidence and absence of any major randomized prospective controlled trials its use remains controversial and it is rarely used. Other medical treatments also being investigated include etidronate (a bisphosphonate), and cytokine inhibitors. Of these, fluorides and bisphosphonates appear to hold the most promise but at present are not considered an option.

Hearing aid

Many surgeons will initially offer all patients a hearing aid for a period of 3–6 months. The reasoning is that because of the rare but potentially serious side risks involved with surgery, if a clinician has tried a conservative management option first, albeit with no success, he/she is in a stronger position to recommend surgery and defend any possible complications.

Some patients will find a hearing aid suitable and elect not to consider surgery. Present day digital and digitally programmed aids offer superior quality and greater flexibility in programming, than their analogue predecessors.

Surgery

Surgery is widely practiced for otosclerosis. The minimum requirements are a mean air–bone gap on the pure tone audiogram of 15 dB and speech discrimination of 70% or better.

Informed consent is mandatory and should cover the following issues:

The success rate

It is best to quote one's own personal series if possible but otherwise the results of major surgical series have reported an 85% chance of obtaining a good hearing improvement and a 10% chance of a slight hearing improvement or no change (Smyth, 1997).

Hearing loss

The patient should be aware that there is a 5% risk of a sensorineural hearing loss and a 1–2% risk of a dead ear (Smyth, 1997).

Tinnitus

This may or may not improve after surgery and indeed may get worse.

Vertigo

A transient unsteadiness is not uncommon but if there is acute vertigo one should suspect a complication.

Facial nerve injury

This is rare and the nerve is at higher risk if it is dehiscent or runs an anomalous course.

Chorda tympani damage

This results in taste disturbance, which usually recovers.

Postoperative infection

In the presence of a footplate fistula this can result in a dead ear and destruction of the labyrinth.

Informed consent should also cover alternative treatment methods as mentioned above and their relative merits.

The operation may be considered contraindicated if there is an active middle ear infection, active otosclerosis with the Schwartz sign, an only hearing ear, Ménière's disease and pregnancy (relative).

Surgical method

The operation is usually carried out under general or local anaesthesia. Hypotensive general anaesthesia (to reduce bleeding) is preferred. The surgical approach can be endomeatal or via an endaural incision.

In essence the operation involves removal of the stapes superstructure (head, neck and crurae), removal of a small part of the footplate (fenestra) and insertion of a prosthesis between the long process of incus and the stapes footplate fenestra with or without an area of intervening tissue (e.g. vein graft). This can be done as a large fenestra procedure where a portion of the footplate is removed or a small fenestra procedure where a small drill, needle or laser is used to create a small opening in the footplate.

Numerous surgical modifications have been described with similar published results seemingly independent of the method used. House et al (2000) compared the effectiveness and long-term stability of hearing results between stapedectomy and small fenestra stapedotomy with a follow up of 11.5 and 6 years respectively and showed in the hands of an experienced surgeon, either technique provides satisfactory and stable long-term results.

Biomaterials for stapes reconstruction are required to be highly stable, non-degradable, have a high mechanical stiffness, be of low weight, be radiologically inert and have easy intraoperative workability (Jahnke et al, 1996). A variety of prosthetic materials have been reported in the literature including gold, Teflon, fluoroplastic platinum and more recently titanium (Tange et al, 1998; de Bruijn et al, 1999a).

Second side surgery

In addition to an acute postoperative sensorineural hearing loss, slow developing progressive late hearing loss can also occur. Although its aetiology is uncertain it may be related to surgery. There is therefore some variability in practice with some surgeons not offering second side surgery and others offering it after a successful first

operation but with an interval of 12–18 months. However, two studies looking at second side surgery from a functional viewpoint using the Glasgow benefit plot, both concluded it increases the chances of achieving at least one 'normal'-hearing ear, and makes symmetrical 'normal' hearing possible in the majority of the cases (Porter et al, 1995; de Bruijn et al, 1999b).

Complications

Peroperative complications

During the procedure the tympanic membrane can tear. Injury to the facial nerve causing facial weakness or chorda tympani damage resulting in taste disturbance can rarely occur. A facial nerve overhanging the footplate or a large persistent stapedial artery may prevent access to the footplate and hence surgery may have to be abandoned. A perilymph gusher rarely occurs requiring packing of the ear. A floating footplate is where the footplate becomes fully mobile during surgery presenting a difficult problem. The best solution is probably to abandon the procedure and allow the footplate to refix. A depressed footplate is one that is pushed into the vestibule and is best left alone (*Table 1*).

Postoperative complications

Immediate or delayed hearing loss can occur. The patient may have a persistent conductive loss as a result of a loose prosthesis becoming detached from the incus or footplate or too short a prosthesis. A perilymphatic leak may result in vertigo and fluctuating hearing loss. A high degree of clinical suspicion is required and a re-operation should attempt to identify and seal the leak. A postoperative infection can have severe consequences and result in a dead ear and/or destruction of the labyrinth.

The laser in stapes surgery

The laser has become the method of choice for many otologists. It is particularly useful in revision stapes surgery; a meta-analysis review of revision stapes surgery using the argon laser confirmed its superiority both in terms of safety and efficacy (Wiet et al, 1997).

There are concerns, however, over possible thermal injury to the inner ear when using the laser to fenestrate the footplate, although the type of laser used does not seem to matter (Yung, 2002).

Delayed facial palsy following conventional stapes surgery is a rare event occurring in 0.2–0.5% of cases. This complication appears to be more common when a laser is used (Ng and Maceri, 1999). It is thought that heating of the facial nerve during laser surgery causes oedema, which in turn leads to compression of the nerve within its bony canal.

Assessment of results

The diversity of operating techniques and the lack of uniformity of reporting make it difficult to compare surgical results. At present no consensus exists on which frequency groups should be used to report surgical results. The American Academy of Otology (1995) recommendations previously used a pure tone threshold average at the frequencies 0.5, 1.0, 2.0 and 4.0 kHz for both air and bone conduction but in their new recommendations 4.0 kHz has been replaced by 3.0 kHz. A successful outcome is one in which the postoperative air–bone gap has been closed to within 10 dB. An audit by the Royal College of Surgeons of England used the frequencies 0.5, 1.0, 2.0 and 4.0 kHz (Harkness et al, 1995).

Smyth and Patterson (1985) attempted to address these problems by introducing the Belfast rule of thumb which states that there is 'significant benefit only if the patient has a post operative threshold better than 30 dB over the four middle frequencies (0.5–4.0 kHz) and less than a 15 dB interaural difference postoperatively.' The rule was further elaborated in 1991 by the Glasgow Benefit plot (Browning et al, 1991).

Others have reported results by evaluating benefit in second side stapes surgery with a more disability-orientated approach using the American Medical Association criteria in the guides to the evaluation of permanent impairment (de Bruijn et al, 1998).

Who should perform stapes surgery?

As there are serious potential complications to this procedure some suggest it should be limited to a small number of surgeons performing it frequently and hence maintaining their expertise. Banerjee et al (2002) published an audit of stapes surgery resulting in a change of practice and recommend managing patients in regional centres with a mandatory central registry to which all surgeons submit their results thus allowing a periodic national comparative audit. Another paper looking at stapes surgery in north-west England reported 22 stapes surgeons performing a maximum of 11 and minimum of 1 stapes procedure per year and also suggested concentrating surgery in specialized centres (Bulman, 2000).

Table 1. Complications of surgery

Peroperative	Tympanic membrane tear
	Facial nerve injury
	Chorda tympani injury
	Perilymph gusher
	Floating footplate
	Depressed footplate
Postoperative	Immediate or late dead ear
	Detachment of prosthesis from incus
	Detachment of prosthesis from footplate
	Footplate granuloma
	Perilymph fistula
	Infection

Surgical outcome

A review of publications reveals that most papers look for a closure of the air–bone gap to within 10 db. Of the larger series Somers et al (1997) reviewed 2521 stapedectomies performed by Professor Marquet of which 10% were revision operations and 20% second ears. The success rate was 81% for primary surgery.

Shea Jr (1998) reported a series of 14 449 stapedectomies over a 40-year period with a 95.1% success rate after 1 year and 62.5% after 30 years. Sedwick et al (1997) reported on 550 patients and found no difference in results between small and large fenestra cases and an overall 78% success rate. Harkness et al (1995) reported an audit of stapedectomy conducted by the Royal College of Surgeons of England. Over a 2-year period 28 surgeons carried out a total of 185 stapedectomies. The results showed that 74% closed the air–bone gap to within 10 dB.

Revision stapedectomy

Revision stapedectomy can be considered in those who have not had closure of the air–bone gap after surgery and those who have redeveloped a conductive loss at a later date the principal causes of which are: loose attachment between the incus and prosthesis, necrosis of the tip of the incus, detachment of the prosthesis from the incus and displacement of the prosthesis from the oval window. Persistent vertigo indicating too long a prosthesis may also warrant revision surgery.

Bone anchored hearing aids

A bone conduction hearing aid is an alternative to surgery. It is reported to give good results and avoids the potential complications mentioned above. It does, however, require a small fitment to a titanium screw placed into the skull behind the pinna that is often visible and may be cosmetically unacceptable to some patients.

Cochlear implantation

At the end stage of otosclerosis, severe sensorineural hearing loss may result. An option in managing such patients is cochlear implantation (Rotteveel, 2004). Cochlear implant surgery in such patients can be challenging, with a relatively high number of partial insertions and misplacements of the electrode array demanding revision surgery. A high proportion of patients experience facial nerve stimulation mainly caused by the distal electrodes. This must be discussed with patients preoperatively.

Conclusions

Otosclerosis is an uncommon condition presenting with deafness. The search for its aetiology has generated much research and hypotheses but no clear agent has been identified. The treatment depends on the patient's symptoms but the majority undergo surgery that has potential serious complications. Digital hearing aids and bone anchored hearing aids offer a good alternative. These treatment strategies have their relative advantages but there remains no single optimum treatment. Further research into biomaterials may improve surgical hearing outcomes but the inherent risks of surgery remain. Hearing aid technology continues to develop and may eventually be considered the treatment of choice. BJHM

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KEY POINTS

- Otosclerosis is an autosomal dominant condition with variable penetrance.
- Otosclerosis only affects the human temporal bone.
- The aetiology is unknown although numerous theories have been postulated.
- The predominant symptom is hearing loss although this may be better in noisy environments.
- The diagnosis is clinical supported by audiological investigation, the most helpful being a pure tone audiogram.
- Treatment options include medical treatment, wearing of a hearing aid and surgery which includes stapedectomy, bone anchored hearing aids and cochlear implantation.
- The results of surgery seem to be comparable irrespective of the type of surgical procedure performed.

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