Congenital Ear Problems

Background

There is a range of congenital ear, nose and throat problems - some occurring alone and others as part of a syndrome. The underlying causes are varied but, as with any congenital disorder, they can be broadly divided into chromosomal abnormalities (mutations and inherited problems), diseases associated with prenatal infection, maternal drug abuse, environmental factors, iatrogenic causes and abnormalities of unknown aetiology. The history will generally point you in the right direction and management will be guided by the degree of functional impairment as well as by any psychological distress associated with the condition (including parental when the child is very young). These are generally uncommon conditions, occurring in <5:10,000 births for each of ear, nose and throat abnormalities.

See separate articles Deafness in Children and Prominent Ears for more information.

Abnormalities of the external ear[1]

Anotia/microtia[2, 3]

Anotia is the total absence of the auricle, most often with narrowing or absence of the external auditory meatus. Strictly speaking, in microtia, there is some degree of malformation of the external ear (± narrowing or absence of the external auditory meatus) in contrast to a 'small ear' which is normally formed, as seen in Down's syndrome. These conditions may be unilateral or bilateral - the latter is less common.

- Anotia is rare but seen in 20% of children with thalidomide-induced abnormalities. Microtia (along with protruding ears) is the most common ear problem encountered in plastic surgery.[5] It is seen in 0.03% of all newborns. It is commonly associated with hemifacial microsomia.
- Between 6% and 16% of cases are associated with chromosomal abnormalities and up to 65% of cases occur in isolation. However, both can also be associated with first arch syndrome and the oculo-auriculo-vertebral spectrum (of which Goldenhar's syndrome is the most severe manifestation). Other associations include Treacher Collins' syndrome, Nager and CHARGE syndrome (Coloboma, Heart defects, Aresia of the choanae, Restriction of growth and developmental delay, Genitourinary abnormalities, Ear anomalies), among others.
- External auditory meatus atresia must be ruled out early (within days in bilateral cases and months in unilateral cases), as delayed speech development can ensue.
- Surgical reconstruction of the auricle is usually carried out between 6-7 years of age.[3] The options will involve either a reconstruction using rib cartilage (which requires several procedures, entails chest wall scarring and the ear does not grow with the child) or prosthetic reconstruction, which can achieve very lifelike results. The procedure can be done under local anaesthetic but general anaesthetic is often preferred, particularly if the patient is a child.
- Postsurgical complications can include infection, haematoma and scarring. Psychological complications include disappointment due to unwarranted expectations - this can be obviated by careful counselling pre-operatively.
- The future promises exciting developments, including intrauterine diagnosis and treatment of severe ear deformities, gene therapy and tissue generation, in vitro cartilage growth and the increasing use of prosthetic implants.

Macrotia[2, 3]

This is a large but normally formed auricle, not usually associated with functional abnormality. It is defined as an ear which is two or more standard deviations from the mean. True macrotia is rare but may be seen in association with vascular malformations, hemihypertrophy, neurofibromatosis and secondary to haemangioma. It is more conspicuous if the ear is prominent too. Surgical correction can be carried out. The Antia-Buch technique, which involves freeing the helical flap and repositioning it, is the most commonly used procedure.[4]

Preauricular accessory auricles

- These are usually found just anterior to the tragus and range from simple skin tags to complex structures containing cartilage.
- They are present in up to 1.5% of the population and, in isolation, usually present with no functional abnormality.
- They can be part of a syndrome (eg, Treacher Collins' syndrome or Goldenhar's syndrome).
- Simple lesions can be easily removed but, when they are more complex (eg, cartilage is involved), surgery is more tricky as all the cartilage needs to be removed and the superficially placed underlying facial nerve can be put at risk.

External auditory meatus atresia[5, 6]

Congenital atresia of the external auditory canal is caused by a failure of canalisation of the epithelial plug portion of the first branchial cleft. This results in the formation of a membranous or bony (or both) plate at the level of the tympanic membrane. There may be associated ossicular malformations. The stenosis may be for part or all of its length.

- Stenosis usually does not result in hearing loss if patency is maintained but atresia does.
- It is a rare condition (of the order of 1-5:200,000 live births) and is more common in boys. There is a positive family history in 14% of cases. Unilateral atresia is 3 to 6 times more likely to occur than bilateral atresia.
- External auditory meatus atresia is often associated with other abnormalities.
- Complications may include recurrent otitis media, cholesteatoma and mastoiditis.
- Management is often multidisciplinary: ear, nose and throat (ENT) surgeons will work alongside audiologists, plastic surgeons, paediatricians and geneticists.

An audiological assessment is carried out in the first instance to rule out hearing impairment in the infant. If it is unilateral, this should take place in the first few months of life but, if it is bilateral, within the first few days.\footnote{8}

Unilateral cases usually manage well, although may have some trouble in localising exact direction of sound. Problems arise if there is repeated otitis media or impacted cerumen in the normal ear.

In cases with bilateral abnormalities, bone conduction hearing aids are the first line of treatment (± followed by bone-anchored hearing aids). This is eventually followed by surgery (after the age of 5 or 6 years), after CT assessment of the extent of the problem and regular audiological monitoring. Surgery is always carried out after any correction of the auricle, as the unscarred skin is essential to the surgeon in fashioning a new auricle.

Abnormalities of the middle ear\footnote{9}

In the absence of other problems, hearing loss associated with these abnormalities is often picked up during the course of routine infant and childhood audiological assessments. More specialist assessment and management is carried out in the ENT department.

**Tympanic membrane abnormalities**

The tympanic membrane may be small (eg, congenital rubella syndrome), distorted (eg, VATER syndrome (Vertebral anomalies, \textit{A}nal atresia, \textit{T}racheo-oesophageal fistula, o\textit{E}sophageal atresia and \textit{R}enal anomalies and radial dysplasia)) or replaced by fibrous tissue or a bony plate.

**Ossicular abnormalities**

- There are a number of different ossicular abnormalities, which may affect one or more of the ossicles.
- There may be absence of part or all of these bones and there can also be varying degrees of fusion.
- The associated intratympanic muscles are often affected and there can be an aberrant course of the facial nerve.
- Surgery can go some way towards correcting this.
Abnormalities of the tympanic cavity

**Congenital cholesteatoma (2-3% of all cholesteatomas)**

It is usually unilateral, may be bilateral, and presents as conductive hearing loss. The tympanic membrane is intact and overlies a white mass (this varies from a small pearl size to filling the entire middle ear) which can act as a source of infection. CT scanning to assess the lesion is advisable as this will dictate the surgical approach.[10]

**Vascular abnormalities**

These include the presence in the middle-ear cavity of internal carotid artery aneurysms, jugular bulb abnormalities and very rare cases of an anomalous internal carotid artery. These vascular abnormalities tend to present with limited functional problems but a pulsatile red, smooth mass may be seen behind the tympanic membrane on examination. Their presence should be confirmed in a specialist unit, as it will have implications in considering any future intervention in the ear or to these structures.

**Congenital perilymph fistula**

This may occur, linking the perilymphatic space of the inner ear to the middle-ear cavity. There are often associated deformities. Children present with fluctuating and progressive sensorineural hearing loss ± tinnitus, vertigo and, occasionally, recurring meningitis. Diagnosis is confirmed on CT scanning and surgical correction can be carried out.

Abnormalities of the inner ear

The inner ear is the collection of structures within the bony labyrinth: the semicircular canals, the vestibule and the cochlea. Congenital abnormalities here are rare and will result in deafness in addition to possible dizziness, and account for up to 20% of children with sensorineural hearing loss.

People with abnormalities of the inner ear are at increased risk of developing recurrent meningitis or a perilymphatic fistula. Middle-ear infections should therefore be treated aggressively. There is also increased risk of developing cerebrospinal fluid leaks after minor head injuries and therefore avoidance of contact sports is advised.

- These deformities are typically classified according to embryonic developmental stages.
- Any of the structures can be involved.
- Cochleosaccular dysplasia is probably the most common form of inner-ear congenital deformity and is characterised by a collapse of the cochlear duct and saccule.

Patients suspected of having these problems will undergo a thorough clinical, audiological and radiological evaluation. A positive family history can suggest a genetic origin, whilst a detailed history of the pregnancy may reveal a teratogenic cause. This may be backed by blood tests. High-resolution CT scanning will determine the nature and extent of the problem and there will be a multidisciplinary approach to rehabilitation.

**Further reading & references**


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