CHAPTER 8.

INHERITED CONDITIONS

I. EMBRYOLOGY

All three compartments of the ear have complex origins, arising from combinations of the primitive ectoderm, mesoderm and endoderm.

1.1. EXTERNAL EAR

The pinna appears as small hillocks derived from the first (maxilla-mandibular) and second (hyoid) branchial arches. These fuse to form a recognisable shape by the end of the second month. The external meatus is derived from the first pharyngeal ectodermal groove, developing medially as a core of ectodermal cells that then hollows out to form the EAC.

![Figure 1: Embryology of the ear. The EAC develops as ectodermal ingrowth, between pinna hillocks. The ossicles are mesodermal, covered with tubo-tympanic recess endoderm.](image)

ear arises as ectoderm of the neural crest, forming the otic vesicle which then sinks into the mesoderm. p: pinna hillocks; i: incus; m: malleus; ov: otic vesicle; ttr: tubo-tympanic recess.

1.2. MIDDLE EAR

The Eustachian tube develops from the dorsal part of the first pharyngeal endodermal groove as the tubo-tympanic recess. The endoderm outpockets to come to line the middle ear and mastoid cells.

The tympanic membrane derives its three layers from the ectoderm of the canal, the mesoderm (the fibrous layer) and the endoderm from the expanding Eustachian tube.

The malleus and the incus (down to the lenticular process) are derived from the mesoderm of the first arch (Meckel’s) cartilage. The tip of the incus and the stapes originate from the second (Reichert’s) cartilage. The ossicles are later enveloped in endoderm from the tubo-tympanic recess.
1.3. INNER EAR

The membranous labyrinth develops as the otic placode, an ectodermal thickening near the pinna. This sinks into the mesoderm to form the otic vesicle, which then forms the cochlear and vestibular pouches, which go on to form the membranous labyrinth. The otic capsule forms from chondrification, then ossification of the surrounding mesoderm.

The auditory nerve is derived from the acoustic-facial portion of the neural crest and is related to the otic vesicle from an early stage.

2. EAR DEFORMITIES

Patterns

a) Protruding auricles
b) Minor:
   i. Pre-auricular sinus/fistula
   ii. Pre-auricular rudiments
c) Major
   i. External Ear:
      Microtia
      EAC Atresia
   ii. Middle Ear
      • Middle ear dysplasia
      • Aberrant facial nerve
   iii. Inner Ear
      • Cochleovestibular
      • Dysplasia
      • Dilated ventricular
      • Aqueducts
   iv. VIII Nerve
      • Dysplasia
      • Dyssynchrony
      • Protruding


Protrusions follow four main patterns. The external ear is comprised of typical skin and cartilage contours that vary only slightly in the great majority of individuals (Figure 1).

Four aberration patterns are seen:

i. The most common is excessive depth of the cartilage that forms the conchal bowl, which causes the ear to stand out from the scalp.

ii. The second, often accompanying a deep bowl, is an undeveloped superior crus cartilage. This results in the upper ear folding forwards and outwards, exacerbating the prominence of an accompanying deep bowl.
iii. The third is a less common fold-over of the helix, forming a “Lop ear”.

iv. Lastly, the upper cartilage of the ear may be over-developed, causing an “elephant ear” appearance.

Surgical correction

Surgery addresses the individual pattern. Correction of the first requires excision of part of the deep bowl, via a postaural incision. The reduced bowl then settles closer to the skull, minimizing the protrusion.

The second procedure follows similar lines, and may be preceded by bowl reduction. The contour of the superior crus cartilage is corrected by weakening the cartilage and then suturing it with stay sutures to achieve the appropriate contour.

The last two problems are more complex to correct. The “lop” variety may require reinforcing
grafts. The enlarged pinna, a more complex flap reduction technique tailored to the individual case.

After surgery, a head bandage may be left in place to allow the tissue to be unmolested for several days.

1.2. MINOR DEFORMITIES

a) Preauricular sinus/fistula

Small pits are commonly seen on the superior root of the helix of the pinna. These are normally shallow and innocuous, but in other instances may ramify widely into the parotid or the external canal, filling with debris and becoming chronically infected. As such these require excision, but this requires a meticulous technique to avoid leaving residual epithelium buried, which will cause recurrent problems. If the infected tract in obscured in inflamed scar tissue within the parotid, facial nerve monitoring during surgery may be advisable.

b) Preauricular rudiments

Also very common, these small skin tags are found just anterior to the tragus and may enclose a cartilaginous core. For cosmetic purposes, small lesions may be ligated, larger examples excised.

Figure 7: Abscess resulting from a pre-auricular sinus infection. Small pits are usually innocuous, larger sinuses may be extensive.

Figure 8: Preauricular rudiment. A small tag is present, with an associated minor earlobe deformity. A small pre-auricular pit is present.

1.3. MAJOR DEFORMITIES

a) Microtia

Underdevelopment/deformity of the auricle varies from subtle deformities and small pre-auricular rudiments to gross developmental failure, distortion, or malpositioning. The external deformity is commonly associated with external canal atresia, middle ear deformity and...
disorganisation, inner ear malformations and aberrations of the course of the facial nerve.

Figure 10: Gross microtia, associated small rudiment and EAC atresia. Common in syndromal cases.

The misshapen pinna may present as a syndromal component, or may be bilateral or unilateral. There may be an associated unilateral facial deformity (hemifacial microsomia).

The condition is a complex mix of cosmetic and functional difficulties.

b) EAC atresia

Failure of canal development occurs when the column of ectoderm that develops from between the pinna hillocks and grows into the mesoderm fails to hollow out. The result is canal aberrations that vary from mild deep canal blunting over the drum, to tiny narrow canals, or total canal osseous occlusion.

Figure 11: Canal atresia. Total obstruction due to congenital fibro-osseous occlusion.

c) Other aberrations

Singularly, or in combination with microtia and/or atresia, the drum, ossicular chain, inner ear, and the VIIth and VIIIth nerve may also display a range of dysplasia aberrations, covering a wide range of gross or subtle abnormalities.

Inner ear deformities are covered in Chapter 12.

1.4. ORIGINS OF MAJOR DEFORMITIES

a) Aetiology

The cause of these problems may be either genetic or developmental, but generally not a repetitive genetic pattern unless part of a syndromal pattern. Genetic causes are generally the more advanced patterns. Lesser examples, especially unilateral, are developmental – failure of the normal tissue combinations that are complex in the ear. The cause of this may be an ill-defined otherwise innocuous event (e.g. viral), or a clearly recognisable noxious agent e.g. thalidomide.
b) Pathogenesis

The auricle develops from six “hillocks” of tissue derived from two folds of the developing head and neck: the first and second brachial arches. Failure of the first often produces lesser problems such as pre-auricular rudiments or relatively slight auricular malformations. Canal atresia may not be present in these cases. Failure of both arches is accompanied by advanced malformations, perhaps with associated syndromal attributes.

![Figure 12: Embryology of the pinna, showing the contributions of the 1st and 2nd arches to the structure of the external ear.](image)

1.5. PRESENTATION

Commonly, external ear deformity is mild; these lesser deformities may cause little concern, or alternatively, may result in major hearing loss in the event of associated canal atresia or middle ear deformities.

Advanced pinna malformations are usually more problematic, as atresia and other cosmetic problems are common in this group. Both difficult cosmetic correction and advanced hearing restoration techniques will be needed for resolution of the combined difficulties. Also, the commonly associated syndromal changes in these children often necessitate other general medical or specific managements (renal, airway, cardiac, psychological).

The degree of hearing loss is unpredictable from the appearance of the auricle, as only minor anatomical derangement may result in substantial conductive deafness. This may be due to a narrow external canal with ossicles that are fused to the canal wall or malformed and dysfunctional. A narrow canal may thwart accurate visual evaluation of the drum and chain with debris in the deeper reaches.

Likewise, the facial nerve may be dysplastic, or malpositioned such that Ossicular repair is impossible.

1.6. INVESTIGATION

a) Audiolog

Conductive deafness will be maximal in severe microtia with canal atresia, but variable in lesser cases. Syndromal cases such as CHARGE association are notorious for inner ear malformations, and possibly VIIIth nerve dysplasia. Profound losses are likely in these cases.

Importantly, in unilateral microtia cases, the hearing in the evidently better ear is checked for normal function on that side; this will ensure normal speech development.

Objective testing (ABR, OAEs) is used to clarify this aspect at birth or shortly after.

b) Radiology

High resolution CT scans and MRI techniques are used to assess the temporal bone anatomy as a guide to the extent of deeper abnormalities and
The scans identify the degree of atresia, and the status of the middle ear dimensions and Ossicular contents. The course of the facial nerve is ascertained, as this may be aberrant. Also, aeration of the ear is noted; failed tubal function will prevent adequate hearing surgical restoration.

The cochlear status is assessed; this is important if cochlear implantation is considered, as a dysplastic cochlea may prevent implantation. The structure of the VIIIth nerve as also checked, importantly if CI may be needed.

Figure 13: 3D reconstruction of an atresia case showing subluxation of the TMJ into the EAC, effectively occluding surgical access to the middle ear.

Importantly, the scans also show the status of the temporomandibular joint. In severe microtia and atresia cases, the joint may be displaced into a position filling the site of the external canal. This invalidates the possibility of canalplasty restoring an external canal; the surgeon would then need to resort to implant surgery to regain hearing.

CT scans also demonstrate skull thickness if a bone conduction implant is considered.

Figure 14: MedPor reconstruction of the pinna, 2 years after surgery.

1.7. MANAGEMENT

Treatment of the deformed ear may focus on the appearance, the function, or a combination of both aspects.

a) Cosmetic

Several methods address the cosmetic aspects. The least surgery removes rudiments or skin tags to “tidy up” the site. This may be supplemented by auricular prostheses; adhesive, or attached via osseointegrated mounts attached through the skin.

Reconstruction of the pinna is much more complex and requires specific skills. Rib cartilage grafts may be used, but require tissue removal via an abdominal incision, then shaping to form a scaffold, implantation, plus secondary skin grafting. Residual external ear elements may need to be included in the repair, especially the ear lobe, if present (this is difficult to create with grafts). Scarring and other tissue dynamics may prove unsatisfactory.

Alternatively, a framework of MedPor (polyethylene) may be utilised to avoid abdominal trauma and to have a ready-made shape available albeit with some risk of biomaterial reactions.
b) Functional

i. Canalplasty and Tympanoplasty

From an early age, a bone conduction aid is fitted, to ensure good development of the auditory pathways, particularly to ensure normal maturity of the auditory cortex, which requires this stimulus for normal anatomical and functional development.

Hearing restoration may be achieved via a canal reconstruction and tympanoplasty techniques to rebuild the drum and chain. However, split skin grafting is needed to re-line the external canal. These grafts are taken from the medial upper arm, for cosmetic reasons. Split skin is not self-cleaning: periodic removal of external canal keratin is needed on a 6-12 month basis in the long term. Canal and chain surgery may be combined with auricular reconstruction in one or two stages.

In less deformed cases, and in expert hands, 80% of cases may achieve good hearing levels from canalplasty and chain reconstruction. Surgical success is not guaranteed, and delayed canal stenosis may occur. Periodic debris accumulation may cause temporary occlusion.

ii. Implantable Hearing Technology

If the ear is anatomically unsuited to surgery because of the extent of deformity, technological aiding is used. From diagnosis, an infant will be fitted with a bone conduction headband aid to stimulate the auditory pathways of the ear. Traditional bone conduction aids are, however, cosmetically evident and may cause uncomfortable pressure effects.

Figure 15: Bonebridge active bone conduction implant, sited under the skin with a small external processor, stabilised by magnetic attraction.

Later (5 years+), electronic implants may restore hearing. Current options include the Med EL Vibrant Soundbridge and Bonebridge devices, or Cochlear BAHA models.

Soundbridge devices are attached to the Ossicular chain to vibrate this in order to create hearing. Middle ear disorganisation invalidates their use in more advanced pathology.

The Bonebridge is an active bone conduction implant, similar to a cochlear implant, which vibrates the skull. Bonebridge devices can be used only when the skull has matured to sufficient thickness.

BAHA devices are also bone conductors, functioning via a titanium screw fixed in the skull through the skin, or held in position by magnetic force. Percutaneous BAHA devices (mounted on screws placed through the skin) commonly incur local tissue reactions require local clearing, and may entangle hair. The magnet-stabilised transcutaneous (intact skin) models avoid these problems.

Cochlear implantation is necessary where severe sensorineural deafness has occurred, but others with concurrent absence of the auditory nerve may necessitate the use of an auditory brainstem implant.
CONCLUSIONS

Microtia and the attendant canal atresia vary from limited aberrations to complex difficulties. For the surgeon, the variability of pathology encountered can be a major difficulty. For the otologist, hearing restoration is a prime concern, especially in the bilaterally afflicted. From the family aspect however, the importance of cosmesis should not be underestimated.

Above all, the family needs realistic advice on the origins of the condition, the management options, the expected outcomes, and the difficulties faced to achieve these gains.