Chapter 5: Congenital anomalies of the ear

Robert Pracy

Congenital anatomical anomalies of the ear are rare. It is difficult to assess accurately the incidence of anomalies per thousand live births because some are not associated with a gross anatomical deformity of the external ear. Since the majority appear to be unilateral, it is probable that there are people who go through life with a unilateral congenital conductive deafness which is never detected.

Furthermore, as there is a great variation in the incidence between the rarer syndromes and the commoner abnormalities, there would appear to be little value in suggesting an overall incidence. However, it is considered that the incidence over all possible causes is in the region of 1:60,000 live births. Broadly speaking anomalies may arise on their own, or in association with other abnormalities which may result from exposure of the mother to drugs during the first trimester of pregnancy or to genetic irregularity. Where the abnormality occurs in association with other obvious defects the dysplasia may be part of a syndrome. Some of these syndromes will be considered below and the dysplasia may be unilateral or bilateral. For a more comprehensive list of all syndromes which may be associated with congenital conductive deafness the appropriate chapter should be consulted.

The purpose of this chapter is to describe the management of congenital abnormalities of the ear as they present, without confusing the reader with lists of very rare syndromes. However, where the anomaly presents as part of a syndrome it is of cardinal importance to take into account the complete picture before offering elaborate surgical procedures. Indeed, as with congenital abnormalities in other sites, it is very desirable to have the patient examined by a developmental paediatrician for other possible abnormalities before proceeding with investigation. A brief recapitulation of the essential developmental and pathological anatomy will be given here (Bowden, 1977). A fuller account of the normal anatomy will be found in Volume 1.

Developmental anatomy

Anatomically the ear consists of three distinct parts, the external, middle and inner ear. Each develops as a separate entity and will be considered separately.

The external ear

This may be divided into two subdivisions, the pinna and the external auditory meatus.

The pinna develops around the first branchial cleft. By the time the pinna begins to appear, the cleft is represented by a pit and this becomes surrounded anteriorly by three small hillocks derived from the tissue of the first branchial arch and posteriorly by three hillocks derived from the tissue of the second branchial arch. The anterior hillocks form the tragus, the crus of the helix and the helix. The posterior hillocks form the antihelix, the antitragus and the lobule. The concavity of the concha is derived from the wall of the branchial pit. Initially the developing pinna lies ventromedially but as development proceeds it moves in a dorsolateral direction. By the twentieth week of intrauterine life the hillocks have fused
forming the adult appearance, but of course much smaller than the final size. After birth the
pinna continues to enlarge and the development cannot be considered as complete before the
twelfth year.

The pinna consists of fibroelastic cartilage covered on either surface by skin, bound
down firmly laterally with fibrous strands and loosely medially by connective tissue.

The external auditory canal also consists of two parts. The outer one-third is
cartilaginous and the medial two-thirds are bony. By the fourth or fifth week of intrauterine
life the branchial pit is in contact with the first pharyngeal diverticulum and the tympanic
membrane will be formed at this point of contact. However, the stimulus for the formation
of the tympanic membrane comes from the handle of the malleus and if this is abnormal no
membrane will appear. By the eighth week a solid cord of epithelium extends from the
primitive external auditory meatus to the tympanic membrane in its lower portion. This cord
is called the meatal plate and canalization of the plate takes place during the twenty-first week
of intrauterine life. Bone forms in membrane round this tube and eventually becomes the
external osseous meatus. Anteroinferiorly the ossification is incomplete at birth but is
completed by the end of the first year. At birth the tympanic membrane lies postero-superiorly
and is horizontal.

The middle ear

This consists of the tympanic membrane, the tympanic cavity, ossicles, muscles, nerves
and blood vessels communicating with the nasopharynx by the eustachian tube. The middle
ear develops from the first branchial pouch which springs from the epipharynx. It is separated
from the external ear by the tympanic membrane and from the inner ear by the bone of the
otic capsule.

The tympanic membrane consists of two parts, the pars flaccida and the pars tensa
which lies inferiorly. Histologically there are three layers in the pars tensa. A middle fibrous
layer is divided into a lateral portion in which the fibres run in a radial direction and a medial
portion in which the fibres run in a generally circular pattern. The lateral surface is covered
by squamous epithelium which is continuous with the epithelium lining the external auditory
meatus. The medial covering is by mucosa of the middle ear. The pars flaccida consists of
two layers, an outer epithelial layer and an inner mucosal layer. The membrane is lodged in
the tympanic ring which is formed from four centres in the medial end of the bone forming
the tympanic plate. It is incomplete superiorly. The handle of the malleus is embedded in the
layers of the tympanic membrane.

The eustachian tube extends from the epipharynx to the tubotympanic recess and
appears at about the third week of intrauterine life. In the infant it passes more or less
horizontally and laterally from the epipharynx to the middle ear. In childhood the
development of the skull and nasopharynx results in an inclination of about 45% passing
upwards and posteriorly and laterally from the nasopharyngeal orifice. The tube has two
portions, a medial cartilaginous part which tapers to a narrow diameter as it passes laterally;
this represents two-thirds of the length of the tube. The lateral one-third is bony beginning
medially at the isthmus and widening as it passes laterally to the middle ear. The tube is lined
with respiratory epithelium which is continuous with that lining the nasal cavity. It is rich in goblet cells particularly at the medial end.

The middle ear proper consists of three parts, the epitympanum, the mesotympanum and the hypotympanum. The floor and roof of the mesotympanum form the roof and floor of the hypotympanum and epitympanum. This is merely an anatomical boundary and is represented by no structure. Essentially the three parts of the cavity lie in that area bounded above by the tegmen tympani, below by the plate of bone over the jugular bulb, medially by the bone of the otic capsule, laterally by the tympanic membrane, posteriorly by the fallopian canal and the facial recess, and anteriorly it merges with the opening of the eustachian tube.

The middle ear contains the three ossicles, intratympanic muscles, nerves and blood vessels. The ossicles are derived from cartilage from the dorsal ends of the first two branchial cartilages. The cartilage of the first arch is called Meckel's cartilage and that of the second Reichert's cartilage. There has been considerable discussion as to the extent to which each contributes to individual ossicles. The current consensus appears to be that the handle of the malleus, the long process of the incus and the stapes, except for the medial layer of the footplate, are derived from Reichert's cartilage. Development of the cartilage begins at the eighth week of intrauterine life and is complete by 16-18 weeks. Ossification begins at 16 weeks and is complete by 32 weeks. The ossicles are suspended in the middle ear by ligaments and surrounded by four mucosal pouches described by Proctor (1964).

The tympanic antrum appears at the twenty-second week of intrauterine life as a posterolateral extension of the epitympanum. At birth it is about 7 mm in diameter. Pneumatization of the mastoid process begins late in fetal life and continues through childhood until the adult state is attained at between 10 and 12 years. The degree of pneumatization varies from ear to ear but total absence of pneumatization should be looked upon as pathological.

The inner ear

The inner ear comprises a membranous labyrinth housed in a bony capsule (the otic capsule) which is part of the petrous bone. At birth the inner ear has already attained adult size.

The membranous labyrinth appears at the third week of intrauterine life as thickenings on the side of the rhombencephalon known as the otic placodes. During the fourth week each placode invaginates giving rise initially to an otic pit and later an otocyst. Subsequently the otocyst divides into a dorsal and a ventral portion. The dorsal portion forms the utricle, semicircular canals and endolympathic duct and the ventral portion, the saccule and the cochlea. Development is complete by the sixth fetal month.

Pathologic anatomy

The normal anatomy may be distorted in either the external, middle or inner ear or, indeed, in all three in the same patient.
Abnormalities of the external ear

The commonest abnormality is an accessory skin tag which may result from incomplete fusion of the tubercles which coalesce to form the pinna. The whole pinna may be smaller on the affected side. The meatus may be stenosed or totally atretic and occasionally the tympanic membrane may be normal but obscured from view by an otherwise uncanalized meatus. When there is a total atresia, a thick wedge of dense bone lies between the mastoid process and the ascending ramus of the mandible in the position normally taken up by the external meatus. In such cases the epitympanum lies superior to the atresia.

Abnormalities of the middle ear

The tympanic membrane may be absent and replaced by a solid bony plate. The whole conformation of the middle ear cleft may be distorted being narrower in some diameters and wider in others. The angle of presentation of the lateral semicircular canals may be altered. The handle of the malleus may be absent or turned through a right angle to face anteriorly where it is ankylosed with the anterior meatal wall.

The malleus and incus may be fused to form one ossicle of considerable mass in which the contribution from malleus or incus is impossible to analyse. The incus may not articulate with the stapes or the long process may be considerably shorter than normal and the connection is established by a fibrous strand. There are many possible types of stapes abnormality. Broadly they may be divided into two categories:

(1) The superstructure may be missing or the crura may be replaced by a solid column of bone. It is then said to be monopodal. Frequently the single crus bends forward from the oval window region and points towards the eustachian tube opening.

(2) The stapes footplate may be abnormal. The normal footplate is formed from two origins. The lateral portion is derived from the second branchial arch and the medial from the bone of the otic capsule. The inner lamina can remain attached to the otic capsule and give rise to congenital stapes footplate fixation (Steele 1969). On other occasions there may be a hole in the centre of the footplate perhaps covered with a fine membrane.

In addition to the bony deformities there are commonly abnormalities in the course of the facial nerve and the blood vessels. The normal covering of the bone of the fallopian canal may be missing and the nerve may prolapse over the oval window. Frequently the descending portion is found in what appears to be the posterior border of the tympanic membrane in those cases where a membrane is present. The descending portion of the nerve may curve backwards sharply as it begins its descent and it then approaches the lateral sinus. On occasion the descending portion of the nerve may be found in two strands which diverge as the nerve travels caudally. Where there is a substantial atretic plate of bone in place of a meatus, the nerve may pass forwards at the level of the neck of the malleus to disappear from the middle ear in the region of the temporomandibular joint. Access to the middle ear cleft may be made more difficult by overhanging dura with large veins on its inferior surface. The stapedial artery may be larger than normal and interfere with instrumentation in the oval window region.
Abnormalities of the inner ear

Sometimes the development of the cochlea is grossly abnormal. Such a child may well be born severely deaf but may appear to have relatively good bone conduction and may be considered to be suffering from congenital conductive deafness. However, surgery in these cases frequently results in a dead ear and on occasion to a perilymph 'gusher'. When the cochlea is found on radiology to consist of a single large cavity with no sign of the normal cochlear turns this is termed the Mondini deformity. It is not amenable to treatment.

The problem

The surgeon must plan with great care the investigation of the patient, the counselling of the parents and the timing of any surgery proposed. His objective should be to have the surgery completed and any hearing aid which may be required, fitted in time for the explosion in language development which occurs normally soon after the age of 2 years. In this way the really important time for language development can be exploited most advantageously. In addition, the child will have little memory of his period of hospitalization. Anatomically there is no reason why this should not be done. The middle ear is almost of adult size at birth and, unless the cleft is greatly distorted by the abnormality, it should be possible to provide a skin-lined cavity with some form of artificial tympanic membrane linked to the ossicular conducting mechanism.

A great deal has been written about how this may be achieved and the fact that there are many opinions as to how it should be done indicates that no perfect solution to the problem has yet been found. The method described below has been evolved over the past 25 years and in the author's hands has proved to give reasonably reliable results which can be kept in a stable condition throughout the period of childhood upper respiratory tract inflammations. As in the case of the normal child, otitis media may occur but, if it is treated promptly, there is no reason why there should be any sequelae. After the age of 12 years one can expect to have a dry stable cavity which will require the same occasional inspection and cleaning as every mastoid cavity.

Clinical presentation of types of dysplasia

(1) There may be no abnormality of the pinna or the outer part of the external auditory meatus. The tympanic membrane may be present and may appear to be normal or the meatus may be stenosed or not patent in the medial portion.

(2) Absence of one or each external auditory meatus with no other obvious abnormality. This is the commonest presentation. The pinna on the affected side is rarely normal but the degree of abnormality may vary from being slightly smaller than normal through presentation as a crumpled skin tag to complete absence. There will be no palpable space between the rudimentary mastoid process and the ascending ramus of the mandible. Gill (1969) has provided a useful 'rule of the thumb' in stating that the greater the degree of abnormality of the pinna the more complex the abnormality of the middle ear will prove to be.
(3) **Absence of one or each pinna with an associated facial weakness on the homolateral side.** The presentation is thus much the same as described above in (2). However, the facial weakness indicates that the nerve to the second branchial arch is involved and it is therefore possible that exploration of these cases will expose an abnormal stapes or even absence of the stapes. This was the condition found quite commonly in those children whose mothers had taken thalidomide in the first trimester of pregnancy and in some cases was associated with abnormalities of the inner ear. On the whole the hearing results in these children were poor and in the author's series all required the use of a hearing aid.

(4) **Absence of the pinna on one or each side associated with other signs indicating a congenital abnormality of other first branchial arch structures.** The most common form of this presentation is the Treacher Collins' syndrome. In such cases it is likely that there will be dysplasia of the lower jaw, flattening of the zygomatic arch and ' antimongoloid' slant to the palpebral fissure. The pinna will lie at a lower level on the side of the head than in a normal child (Wright, Phelps and Fraser, 1977).

(5) **Dysplasia of the ear may be part of a more obvious syndrome** such as Crouzon's syndrome in which there are other facial and structural abnormalities of the head which will draw attention to the obvious difference between the patient and the normal baby.

(6) **Dysplasia of the ear may be one of multiple abnormalities of the head and other organs presenting no recognizable features of any syndrome.** In such cases the abnormality of the ear, disabling though it undoubtedly is, must be considered to take a second place to the other more serious disabilities. Surgery should not be offered.

**The assessment of the patient and counselling of the parents**

The parents of every child born with an obvious structural abnormality, particularly one which cannot be hidden, are going to be very concerned. The baby is therefore likely to be brought for advice at a very early age. Naturally the worried parent is going to expect to be told how his or her child is going to be affected physiologically as a result of the anatomical abnormality. It cannot be emphasized too strongly that until there is objective evidence of some cochlear function, it is unwise to discuss the prognosis of the particular patient. However, it is equally important that the whole picture of the patient with bilateral atresia should be discussed with the parents if their child presents in this way. If the preliminary examination indicates that the atresia is merely one facet of a syndrome or of multiple disabilities then the first step should be to seek the advice of the developmental paediatrician in order to get some idea of the prognosis for the future pattern of life.

If the condition is unilateral, once the cochlear function on the healthy side has been verified as being normal the parents should be reassured and every effort made to postpone surgery on the atretic side until the child is old enough to make up his own mind about surgery. If the condition is bilateral, a bone conductor hearing aid should be fitted immediately in order to bring ambient sound to the child at the earliest possible moment. It then becomes necessary to carry out a radiological examination to confirm that a cochlea is present on each side. A 'transorbital' view is the best way to obtain this information as only one exposure is required and, if the cochlea is present on both sides, it will be necessary at
a later stage to ask for hypocycloidal polytomography in order to have detailed information about the anatomy of the inner ear on both sides. If the cochlea is present on one side only this represents an absolute contraindication to all attempts to construct an artificial sound pathway because of the possibility of producing a dead ear as the result of surgery.

If the patient is found to have a cochlea on each side it is necessary to wait until about the age of 4 months in order to be able to verify that there is a cochlear reserve. (It is not always easy to verify that it is equal on the two sides.) Once this has been established the whole plan for the future management of the child can be discussed with the parents. Hypocycloidal polytomography is best left until shortly before it is proposed to operate. It is usually possible to obtain better films with less risk to the patient and the anatomic findings can then be discussed with the radiologist (see Chapter 2).

No possible service can be done for either the patient or the parent by offering optimistic prognoses for hearing or for the ultimate 'natural' appearance of the pinna. Experience shows that the hearing results are poor in the majority of cases, stenosis of the new meatus is common and the final appearance of the pinna bears little resemblance to the norm. The first step towards forming a satisfactory relationship with the family is to be quite open about what is 'on offer'. The promise of relatively little will of course be disappointing to the parents, but this is infinitely better for both the surgeon and the family than the inevitable frustrations and misunderstandings which are bound to follow from an unwarranted optimism on the part of the surgeon. Furthermore, the better the rapport with the parents the more chance of their cooperation if the course of management has to be modified.

The practice of offering to create only a meatus without making a definitive link with the ossicular chain has much to commend it. This has been the author's practice for many years, with an offer to combine this procedure with an ossicular link if it can be accomplished relatively easily without danger to important structures such as the facial nerve. The modern hearing aid is small and can be fitted fairly satisfactorily to the new meatus and will almost certainly provide more satisfactory hearing than a surgical linkage. In about 5% of cases it is possible to achieve a good hearing result which will not require the provision of a hearing aid. Experience shows that what the child wants more than anything is to have an 'earhole' like other children. This requirement can be met in a high percentage of cases.

**Operations on both ears**

There can be no justification in operating on both ears in early childhood. The object of the operation is to provide an effective meatus to which, if necessary a hearing aid may be fitted. The possibility of providing stereophonic hearing by operations on both sides is so remote that it cannot be weighed in the balance against the additional trauma to the patient and the family. The difficulty is that a very successful result achieved on one side is very likely to result in extreme pressure from the parents for the other side to be treated immediately. This should be resisted because a good result on both sides proves to be a very rare occurrence.
Surgery for conductive deafness without meatal atresia

The approach of choice for this condition is by the tympanotomy route. The actual reconstructive procedure employed will naturally depend upon the underlying pathology and may vary from stapedectomy procedures to the establishment of a functioning ossicular chain by the use of a prosthesis, wire or a bone graft. Surgery should not be attempted until the meatus is large enough to accommodate the instruments required and, at the same time, to provide an adequate view of the operative field down the microscope. Generally this is not earlier than 4 years of age. Experience shows that it is often helpful to use a small endaural incision to allow better access. Extreme caution should be adopted in turning the tympanomeatal flap forward because of the possibility of damage to an exposed facial nerve which, as has been described above, may lie in the free border of the membrane or in the immediate vicinity. If, in attempting to free a fixed footplate, a perilymph 'gusher' occurs the leak should be controlled with a plug of fat or muscle kept in position with a strut or short piston attached to the incus; the original objective of ossicular reconstruction should be abandoned.

Surgery when there is atresia of the external ear

The complications of procedures designed to correct meatal atresia are infection of the cavity with resulting death of the skin graft, leading to granulation tissue formation and subsequent stenosis or closure. Poor hearing may result either from failure to anchor the new tympanic membrane to the ossicular chain, which then moves laterally, or a dead ear following drilling on an intact and mobile ossicular chain. As in the tympanotomy procedure described above, there is always a risk of damage to the facial nerve. Earlier operations for this condition were of the 'open' type (Ombredanne, 1947; Livingstone, 1959, 1965), in which a bony meatus was shaped by drilling and then lined by a Thiersch graft fashioned in the form of a tube which was then splinted by packing. Once the packing had been removed the site became prone to infection as a result of investigation by the patient's fingers. If the operation was delayed until a more cooperative patient had developed, the objective of attaining a stable meatus in time for the language explosion was lost (Colman, 1974).

For this reason a two-stage operation was developed which could be carried out in the second year of life. The principle behind the operation was to create a closed skin-lined cavity not accessible to the introduction of infection from outside which could be converted into a meatus after an interval of 3 or 4 months, by which time the graft both of the tympanic membrane and of the meatal wall would have had time to 'bed down'. Initially the attachment of the new meatus to the pinna was left until the child was older, but now that the fashion is to wear the hair longer, the second stage can be combined with a Z-plasty of the pinna rudiment, with or without rotation, and this provides a satisfactory result without the need for multiple procedures. The first stage of the operation is carried out once the child is clean and dry at about 14-16 months and the second stage at 18-20 months. Many paediatricians feel that this is early for toilet training to be complete but experience of over 70 cases shows that adequate training can be achieved with full parental cooperation.

A fuller description of the operation will be found elsewhere (Pracy, 1977) but the broad steps will be given here. The incision should be planned with the Z-plasty in mind. It
should be straight and angled at 45° to the vertical. It extends from the lower border of the mastoid process to the lower border of the temporalis muscle.

Once the soft tissues have been retracted and the bone uncovered the mastoid antrum is exposed. It should be appreciated that this will lie apparently more posteriorly because the mastoid process in these cases lies against the ascending ramus of the mandible. Once the antrum has been defined, the opening should be enlarged superiorly as far as the middle fossa dura, posteriorly as far as the lateral sinus and anteriorly just below the tegmen tympani until the upper portion of the ossicular mass is exposed.

It is then possible to remove the atretic plate from above working from the epitympanum without danger to the facial nerve. The drilling should proceed with caution as the region of the neck of the malleus is approached, since it is in this area that the facial nerve can turn forwards horizontally. If the ossicular chain is fixed the malleus and incus should be disarticulated from the stapes in order to avoid damage to the cochlea by noise or vibration. Once an adequate view of the medial wall of the middle ear has been obtained the drilling can be directed to forming a suitable bed for the tympanic membrane graft. When this has been done a decision should be made about the use of the ossicles removed to provide a rounded columella which can be shaped to fit over the stapes.

The middle ear region is now completed by the positioning of a graft made from temporalis fascia, taking care to ensure that it lines at least the medial one-third of the newly defined anterior meatal wall.

The remainder of the cavity is lined with a Thiersch graft taken from the inner side of the homolateral thigh and supported by a sterile non-absorbable plastic sponge. The final piece of skin is turned so that the outer surface faces inwards on to the sponge. The wound is closed in layers without drainage.

In the second stage of the procedure the incision is reopened and extended into a Z-plasty. The cyst formed deliberately by the first operation is opened and will be found to be lined by healthy skin. There will be enough skin to suture that inside the cavity to the external skin and, by the use of the Z-plasty, the opening can be brought out anterior to and above the pinna remnant. The cavity should be lightly packed for a further 14 days and subsequently treated like a mastoid cavity.

**Results**

Results have to be judged from the surgical standpoint and from the degree to which it has been possible to offset the pathological anatomy. A recent review of 20 cases, treated by two surgeons by the method described, and reported to a meeting of the Section of Otology of the Royal Society of Medicine (Pracy, 1987), offers a good illustration of what may be expected. Twenty parents were circulated and only 10 replied. One mother was totally dissatisfied. Her child was not suitable for surgery and therefore did not perform well at school as she had virtually no cochlear reserve and did not find a hearing aid of much use. All of the remaining parents who replied said that the advice that they had been given at counselling was accurate. The children were being educated in normal schools. In some the language development was retarded, but all performed normally from an educational point of
view. Only one child did not need to use a hearing aid. Due to complications discussed above some had required a second meatoplasty but all had manageable cavities.