Despite its complicated embryological development, most significant congenital anomalies of the nose are rare. While a precise explanation for this infrequency is lacking, it may well be that, owing to the intimate association between development of the face and brain, severe structural abnormalities of this region are largely incompatible with continued fetal existence.

There is much to be said for entrusting the long-term management of children with congenital nasal defects to a few large centres, where the necessary radiological, maxillofacial, neurosurgical and other skills are readily available. Few otolaryngologists will gain significant experience, during their working lifetime, in the care of such children and, moreover, many of these patients have additional congenital abnormalities, especially of the face and central nervous system.

Development of the nose

In order that the reader may understand more fully the teratogenesis of those anomalies which do occur, a brief summary of the embryology of the nose will be given here. For a fuller description Volume 1, Chapter 5 should be consulted.

At about the third week of fetal life, paired thickenings (the olfactory or nasal placodes) appear in the cranial ectoderm, near the embryonic anterior neuropore.

Invagination of the placodes, consequent on growth of the surrounding mesoderm, results in the formation of the nasal pits which, as they deepen, serve to delineate medial and lateral prominences of the frontonasal process. The medial prominences fuse to form the central portion of the upper lip, the premaxilla and the primitive nasal septum.

The floor of the nose is, at first, formed by medial growth of the maxillary processes of the mandibular arch. As the nasal pits deepen, they become slit-like; progressive thinning of the mesoderm, dorsally and caudally, results in the formation of the bucconasal membrane, separating the lumen of the nose from the buccal cavity.

The bucconasal membrane eventually breaks down, forming the primitive posterior nasal apertures which, thus, lie horizontally in the roof of the buccal cavity or stomatodaeum. The lower, free border of the developing nasal septum, at this stage, lies in contact with the dorsum of the tongue.

Meanwhile, paired lateral palatal processes are formed, one on each side of the tongue. Fusion of these with each other, with an unpaired, ventrally sited, median process and with the caudal border of the septum, gives rise to the definitive palate, finally separating the nasal and nasopharyngeal cavities from the mouth.
Choanal atresia

Choana is derived from a Greek word, Χοανή, meaning a funnel. The choanae are, by definition, the posterior apertures of the nose (Friel, 1974); the use of the term 'posterior choanae' is, therefore, tautology.

Atresia of the choanae was first described by Roederer in 1775 (Devgan and Harkins, 1977). It is one of the more commonly observed congenital abnormalities of the nose, although its true incidence is uncertain. The most consistently quoted figure is one per 8000 live births, but this is probably an underestimate; bilateral choanal atresia is likely to have been, and may still be, a frequently unrecognized cause of death in the neonatal period. Females appear to be affected about twice as often as males and the condition may be unilateral or bilateral in the proportion 3:2 (Kaplan, 1985).

Embryology

Much confusion exists about which embryological structure is actually involved in choanal atresia. Some authors (Sprinkle and Sporck, 1983; Kaplan, 1985) subscribe to the theory that the anomaly may arise as a result of persistence of the buccopharyngeal membrane. It is difficult to reconcile this view with the embryology of the region, however, as the buccopharyngeal membrane, in the developing fetus, lies posterior to the site of Rathke's pouch, which in turn lies posterior to the nasal septum.

It has also been suggested that the aberration is due to a failure of the bucconasal membrane to undergo involution (Sprinkle and Sporck, 1983). The primitive posterior nasal apertures, produced by breakdown of the bucconasal membrane are, however, considerably more anteriorly placed than the definitive choanae. While this mechanism may explain some instances of choanal atresia, it is the author's view that the majority of cases are due to persistence of the epithelial cells which proliferate within the nasal cavities during the sixth to eighth weeks of intrauterine development.

Pathology

Choanal atresia may be bony (90%) or membranous (10%) and is generally sited just in front of the posterior end of the nasal septum. In most instances where the atretic plate is of the bony type, it is thin and easily perforated; occasionally a much more substantial atresia occurs, a considerable proportion of the posterior nasal cavity being obliterated by dense bone. In this latter form, it is frequently observed that the nasal cavity is funnel shaped (Pracy, 1979; Pirsig, 1986).

Asymmetry of the facial skeleton is common, especially in those with unilateral atresia, and most patients will have a high arched palate. Other orthodontic abnormalities, such as transverse maxillary compression, have been described and there may be deformed or absent teeth, especially those of the second dentition. Pirsig (1986) is of the opinion that congenital choanal atresia should not be considered as an isolated plate of bone but, rather, as one component of a skull base anomaly developing between the fourth and twelfth weeks of gestation.
Computerized tomography (CT) has demonstrated thickening of the vomer, bowing of the lateral wall of the nasal cavity and fusion of bony elements in the choanal region (Wetmore and Mahboubi, 1986). Maurizi et al (1985) have observed various changes in the cell morphology of the nasal ciliated columnar mucous membrane. They described unevenness of the mucosal surface, thickening of the basal membrane and the presence of ciliary abnormalities including compound cilia, cilia with excessive cytoplasm and ciliary structures with loss of the peripheral membrane.

Incomplete obstruction of the posterior nasal aperture may also occur, usually taking the form of bony stenosis; if severe, this may mimic true atresia, necessitating similar management. Congenital atresia of the anterior nares has been described but is excessively rare (McNab Jones, 1971).

**Clinical features**

Unilateral choanal atresia, unless specifically sought at birth, may not become apparent until later childhood or even adulthood. Feeding difficulties may occur, especially during breast feeding when the non-affected side of the nose is occluded by contact with the breast. More commonly, however, it presents as a unilateral nasal discharge.

Bilateral atresia, on the other hand, almost always presents as a respiratory emergency, and is, thus, apparent at birth. Newborn infants are, by instinct, nose breathers. The reflexes, which in the older child or adult will result in breathing through the mouth in response to nasal obstruction, do not develop until some weeks or months after birth. If, however, the mouth is held open, either by insertion of an artificial airway or during crying, then mouth breathing will occur. Thus, neonates with bilateral choanal atresia, or other cause of severe nasal obstruction, will sometimes demonstrate a cyclical change in oxygenation, becoming cyanosed during quiet periods, normal colour returning when the child cries.

While choanal atresia may be found as an isolated anomaly, about 60% of cases are associated with one or more supplementary congenital defect (Kaplan, 1985). In addition to random affiliations, however, choanal atresia has, recently been linked with a limited number of specific defects - the so-called CHARGE association (Pagon et al, 1981):

- C  colobomatous blindness
- H  heart disease
- A  atresia of the choanae
- R  retarded growth or development, including the central nervous system
- G  genital hypoplasia in males
- E  ear deformities, including deafness.

**Diagnosis**

The time-honoured method of determining patency of the nose in the newborn infant, is the visualized ability to pass a soft red-rubber catheter through each side of the nose into the oropharynx. Failure to pass the catheter, however, is not conclusive evidence for the presence of atresia of the posterior nares; the tip may become impacted in the adenoid or may be deflected by minor abnormalities of the turbinates, causing the tube to curl up inside the
nasal cavity. In consequence, the author prefers to use a stethoscope, from which the bell has been removed, the open end of the tube being held over each nostril in turn and the presence or absence of an air-blast noted.

There are, however, causes of total nasal obstruction in the neonate other than choanal atresia: these include massive congenital hypertrophy of the turbinates or adenoid. Radiographic demonstration that contrast medium is help up at the choanae (choanography), has been the traditional method of confirming the diagnosis of atresia. Recently, it has been suggested that computerized tomography gives rather more information, especially with regard to whether the obstruction is membranous or bony and, in the latter case, the actual structures involved and their thickness (Brown et al, 1986).

Management

The essential aim of treatment of choanal atresia is the creation of patent nasal airways. In the case of unilateral atresia, there is seldom any urgency in the presentation and surgery can be undertaken as a planned or 'cold' procedure. Bilateral atresia, however, always presents as a respiratory emergency and management must, therefore, be considered under two headings:

(1) primary, or emergency, treatment
(2) secondary, or definitive, surgery.

Emergency management

In most cases, and especially where it is planned to proceed to definitive surgery within 24 hours or so, it will suffice to insert a standard neonatal Waters airway, taping it securely into position. The child may, if necessary, be fed through an indwelling nasogastric tube. In a few cases, where the child's general condition is precarious, perhaps because of additional pathology, it will be found necessary to delay surgery. In these circumstances, it is probably wiser to intubate the larynx or, if this is not possible, to perform a tracheostomy.

Definitive management

This is best carried out as soon as possible. Four approaches to the posterior end of the nasal cavity have been described:

(1) trans-nasal
(2) trans-palatal
(3) trans-septal
(4) trans-antral.

Of these, only the first two are in common use today. In all surgery of this region, use of the operating microscope not only provides much better illumination but makes the operation easier and safer. General anaesthesia with orotracheal intubation is employed.

The transnasal route is valid only in membranous atresia or where the bony plate is thin. The simplest procedure is perforation of the atretic lamina followed by dilatation.
Although Lichtwitz's trocar and cannula is often used to achieve the primary puncture, there is an inherent risk in this method of causing serious damage to the cervical spine and spinal cord. Instead, the author recommends the use of female urethral dilators which, being curved, direct the perforating force safely downwards, into the lumen of the nasopharynx.

The atretic plate is, almost always, thinnest and weakest at the junction of the floor of the nose and posterior end of the septum. It is, therefore, towards this point that the tip of the instrument should be directed. If gentle pressure does not succeed in perforating the atresia, it is probably too thick and some other method should be employed instead. Once puncture has been achieved, the opening may be widened, using progressively larger dilators, up to about 5 mm (14-16 FG).

Many surgeons recommend the insertion of Portex or Silastic tubes to prevent re-stenosis of the choanae, but the author feels that these do not help and prefers, instead, to dilate serially the choanae once a week for 4-6 weeks.

Other transnasal methods which have been advocated are drilling out the bony atresia with diamond paste burrs or vaporizing the obstruction using the CO₂ gas laser. In either case, it is imperative that an aural speculum is used to protect the skin of the nasal vestibule as it is very easy, otherwise, to cause circumferential damage, resulting in stenosis of the anterior naris.

Where the atresia is thick, it is preferable to employ the transpalatal approach. The most comfortable operating approach is similar to that used in cleft palate surgery, the child's head overhanging the end of the table and resting on the surgeon's lap. The Denis Browne mouth gag gives rather better exposure of the hard palate and is to be preferred to the more usual Boyle-Davis instrument.

An incision is made around the summit of the alveolar ridge or at the gingivopalatal margin if teeth are present. The mucous membrane of the hard palate is elevated, using McKenty or Cottle septal elevators and the flap is developed posteriorly, until the edge of the hard palate is reached. Care must be taken to avoid damage to the greater and lesser palatine vessels and nerves as they traverse the bony palate on either side, close to its posterior edge. The nasopharynx is entered by separation of muscle fibres from the posterior edge of the hard palate and incision of the superior mucosal layer of the soft palate.

Using diamond paste or cutting burs of suitable size, the posterior end of the hard palate is removed to expose the bony atresia. Much has been written about preservation of mucosal flaps, intended to re-line the new choanae, but in the author's experience, this is almost always impossible to achieve. Continuity of the nasal cavity can now be restored by drilling away the obstructing bone, the posterior end of the vomer being removed at the same time. Care is needed, at this stage, to avoid damage to the vessels and nerves which run in the lateral nasal wall.

Before replacing and suturing the palatal flap, soft Silastic tubes may be inserted and anchored in position with an anterior septal transfixion suture; the tubes should be removed 4-6 weeks later. As in the transnasal approach, the author does not believe that such tubes are necessary.
Pirsig (1986), in a very comprehensive review of the surgery of choanal atresia, has drawn attention to the possibility of subsequent maldevelopment of the upper dental arch in patients who have undergone the transpalatal operation. There is also risk of palatal perforation if the palatal flap is too short.

**Septal deviation**

Some degree of nasal septal deviation is found in 58% of all newborn babies, and in 4% of births there is also an associated external nasal deformity (Gray, 1985). Two mechanisms have been proposed to explain how such deformities may arise:

1. differences in the rate of growth of the septum as compared to other midfacial structures, resulting in a septum which is too big for the space it has to occupy

2. trauma to the nose, either as the result of prolonged contact with the uterine wall or during parturition, especially when this is protracted.

It should be noted that septal deviation is a frequent concomitant of clefting of the upper lip and palate.

The resulting nasal obstruction, which may affect one or both sides, usually presents, in the neonate, as difficult or slow feeding, often accompanied by colic due to air swallowing. If, as frequently happens, nasal infection supervenes, the child will become snuffly and in some cases the nasal blockage is so severe as to mimic choanal atresia.

Inspection of the nose may reveal displacement of the quadrilateral cartilage, but more posteriorly sited deformities of the perpendicular plate of the ethmoid or vomer, are not always visible. Gray (1985) described specially shaped nasal struts which he used to determine the septal configuration. A good estimate of nasal patency may be made by comparing the air-blast heard over each nostril, as described in the section on choanal atresia.

Both external pyramidal and internal septal deformities may be corrected, within the first few days of life, using specially designed, neonatal nasal septum forceps (Gray, 1985; Alpini et al, 1986). By about 6 years of age, the nose is large enough to allow septoplasty to be performed, but it is recommended that tissue removal be kept to an absolute minimum, otherwise subsequent growth and development of the nose may be jeopardized.

**Congenital nasal masses**

Congenital nasal masses are rare, occurring once in every 20,000-40,000 live births. All intranasal masses in children, and especially if unilateral should be treated with the gravest suspicion and circumspection. Failure to differentiate between a simple nasal polyp and a communicating meningoencephalocele may lead to cerebrospinal fluid rhinorrhea, with resultant risk of meningitis. The differential diagnosis of nasal swellings in a child is given in *Table 15.1.*
Table 15.1 Causes of nasal swelling in childhood

<table>
<thead>
<tr>
<th>Cystic</th>
<th>Solid</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Congenital</strong></td>
<td><strong>Congenital</strong></td>
</tr>
<tr>
<td>meningoencephalocele</td>
<td>glioma</td>
</tr>
<tr>
<td>meningocoele</td>
<td>haemangioma</td>
</tr>
<tr>
<td>dermoid cyst</td>
<td>lymphangioma</td>
</tr>
<tr>
<td>epidermoid cyst</td>
<td>neurofibroma</td>
</tr>
<tr>
<td></td>
<td>neuroblastoma</td>
</tr>
<tr>
<td></td>
<td>rhabdomyosarcoma</td>
</tr>
<tr>
<td></td>
<td>chordoma</td>
</tr>
<tr>
<td></td>
<td>craniopharyngioma</td>
</tr>
<tr>
<td><strong>Acquired</strong></td>
<td><strong>Acquired</strong></td>
</tr>
<tr>
<td>sebaceous cyst</td>
<td>lipoma</td>
</tr>
<tr>
<td>lacrimal duct cyst</td>
<td>papilloma</td>
</tr>
<tr>
<td>mucocoele</td>
<td>lymphoma</td>
</tr>
<tr>
<td></td>
<td>nasopharyngeal carcinoma</td>
</tr>
<tr>
<td></td>
<td>angiofibroma</td>
</tr>
<tr>
<td></td>
<td>ethmoidal polyp</td>
</tr>
<tr>
<td></td>
<td>antrochoanal polyp</td>
</tr>
<tr>
<td></td>
<td>abscess.</td>
</tr>
</tbody>
</table>

**Nasal dermoids**

These are cysts or sinuses occurring anywhere in the midline of the nose from the glabella to the columella, whose walls contain skin adnexae (Bradley, 1981). They are formed as the result of sequestration of epithelial elements during fusion of the median nasal processes. The diagnosis is often delayed until adolescence or adulthood and may only become manifest if infection ensures.

Dermoid sinuses, recognized by a dimple or minute opening sometimes containing a single hair, may extend deeply into the nasal septum, occasionally reaching as far as the cribriform plate. Contrast sinography will assist in determining both the extent and configuration of the sinus tract.

Complete excision is usually difficult to achieve, but may be facilitated by prior cannulation of the external punctum and instillation of methylene blue.

**Nasal gliomata**

These account for approximately 5% of all congenital nasal swellings. They are said to be more common in males and may occur entirely outside the nasal cavity (60%), entirely within the nasal cavity (30%) or in a combination of both sites (10%).

Although the exact embryological details are uncertain, it seems likely that gliomata and meningoencephaloceles share a common origin, resulting from faulty closure of the
anterior neuropore. However, while meningoencephaloceles retain their communication with the subarachnoid space, gliomata become detached from the intracranial cavity by closure of the skull sutures, although in some cases a fibrous tract may remain, connecting the glioma to the skull base.

Macroscopically, gliomata are smooth and rubbery with a grey, yellow or purple surface. On histological examination, there are aggregates of mature glial cells, predominantly astrocytes, interspersed with fibrous tissue (Swift and Singh, 1985).

Most cases are diagnosed at or soon after birth, presenting as either a subcutaneous lump, to one side of the nasal bridge, or as an obstructing intranasal mass. Unlike a meningoencephalocele, a glioma does not increase in size with straining or crying. Tomography or CT scanning of the anterior skull base is usually carried out, although it should be noted that absence of a bony defect does not rule out an intracranial communication. Generally, the diagnosis is not in doubt but should always be confirmed by biopsy, having first checked for the presence of cerebrospinal fluid by needle aspiration.

Nasal gliomata tend to enlarge slowly with age although, occasionally, rapid expansion may occur. Treatment is by excision. Intranasal masses may require a lateral rhinotomy approach although laser excision is also effective. Incomplete removal may lead to recurrence. In those few cases where a dural connection can be demonstrated, anterior craniotomy will be necessary to prevent cerebrospinal fluid leak.

Nasal meningoencephaloceles

Meningoencephaloceles, sometimes incorrectly referred to as encephaloceles, are local herniations of glial tissue and meninges, through a defect in the skull. While these may occur at any site, they are commonly classified into five main groups, depending upon the size and site of herniation (Table 15.2). Cranioschisis, it should be noted, refers to very large bony defects in the skull. Most of the meningoencephaloceles seen by otorhinolaryngologists are of the frontoethmoidal or basal types.

Table 15.2 Classification of meningoencephaloceles

<table>
<thead>
<tr>
<th>Occipital</th>
<th>Cranial vault</th>
<th>Basal</th>
<th>Cranioschisis</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>interfrontal</td>
<td></td>
<td>cranial upper facial cleft</td>
</tr>
<tr>
<td></td>
<td>anterior fontanelle</td>
<td></td>
<td>basal lower facial cleft</td>
</tr>
<tr>
<td></td>
<td>interparietai</td>
<td></td>
<td>occipito-cervical cleft</td>
</tr>
<tr>
<td></td>
<td>posterior fontanelle</td>
<td></td>
<td>acrania and anencephaly.</td>
</tr>
<tr>
<td></td>
<td>temporal</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Fronto-ethmoidal meningo-encephaloceles probably share a common origin with nasal gliomata, as described in the previous section; the origin of other groups will depend on local
developmental abnormalities. The sac-like protrusion of meninges contains brain tissue and the subarachnoid space, which is filled with cerebro-spinal fluid, communicates freely with the cranial cavity. Injury to a meningo-encephalocele is, therefore, likely to cause cerebrospinal fluid rhinorrhoea and meningitis.

A meningo-encephalocele usually presents as either a soft cystic mass overlying the root of the nose (fronto-ethmoidal type) or as a pedunculated intranasal swelling (basal type). Crying or straining is said to increase the size and tension of the mass, although, in the case of intranasal swellings, this may not be easy to detect. All such swellings must be subject to rigorous radiological examination, including plain films, tomography and CT scanning, to determine the exact site and size of the cranial defect.

In those few cases where the skull defect is small and readily accessible, local external excision with careful plugging of the cranial opening, may suffice. Most cases, however, will require the assistance of a neurosurgeon. Craniotomy is performed, with removal of the herniated brain tissue, followed by closure of the bony defect using tantalum mesh and repair of the meninges with fascia or homologous dura.

Other malformations

Haemangioma

Haemangiomata, which most authorities regard as vascular hamartomata, rather than true neoplasms, are common in childhood (Walter and Israel, 1970). Although the majority of these tumours are found in the head and neck region, primary involvement of the nose is relatively rare. Histologically, haemangiomata have been identified as capillary, cavernous, mixed or hypertrophic, although the usefulness of this classification for either prognosis or typing has been questioned (Batsakis, 1974).

Despite being unsightly and disfiguring, almost all of these lesions will regress spontaneously, with little or no residual deformity (Thomson and Lanigan, 1979). Those few tumours which do not involute are said to be associated with increasing numbers of arteriovenous fistulae, the development of which can be monitored by serial Doppler examinations. In the majority of cases, therefore, masterly inactivity is the treatment of choice, active intervention being restricted to those instances where tumour growth continues.

Agenesis of the nose

Total agenesis of the nose is an exceptionally rare abnormality with fewer than a dozen cases having been reported in the literature (Sprinkle and Sporck, 1983). In two cases reported by Gifford and McCollum (1979), there was an associated absence of the nasopharynx and no evidence of any nasal development. The child may learn to mouth breathe, or surgery may be undertaken in a bid to establish a nasal airway. A nasal prosthesis is probably more cosmetically acceptable than the results of surgical reconstruction.

Partial agenesis with failed development of one nasal cavity has also been reported.
Cleft nose

This deformity is also very rare. The actual degree of clefting which occurs varies considerably, from minor notching of the nasal tip to total midline division of the nose into widely separated nasal cavities. There may be associated median clefting of the upper lip and palate or notching of the alar margins. Most cases exhibit hypertelorism and it has been suggested that there are strong associations between separation of the eyes, cephalic anomalies and the probability of mental deficiency (DeMyer, Zeman and Palmer, 1963).

Surgical repair of these anomalies is likely to require the assistance of a maxillofacial surgeon and, perhaps, a neurosurgeon. In the more severe cases, multiple procedures will be necessary and, where bony elements are involved, it may be advantageous to delay repair until growth of the nose and face have ceased.

Proboscis lateralis

This unusual deformity consists of a tube of skin and soft tissue, arising at the inner canthus of the eye. The nasal cavity, on the affected side, may be completely normal or there may be maldevelopment, of varying degree, up to and including total agenesis.

The embryological defect would appear to result from imperfect fusion of the lateral nasal and maxillary processes. As a consequence, there is also failure in development of the nasolacrimal duct.

Where there is maldevelopment of the nasal cavity, repair will be facilitated by incorporation of some of the extraneous tissue of the tube in reconstruction of the nose. Some form of dacryocystorhinostomy will also be required.