Chapter 26: Congenital disorders of the larynx, trachea and bronchi

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The true incidence of congenital deformities of the upper airway is not known for certain, but van den Broek and Brinkman (1979) quoted an incidence for congenital laryngeal defects of between 1:10.000 and 1:50.000 births. Of 219 children with stridor who underwent endoscopic examination, Holinger (1980) found 81% to have evidence of congenital abnormalities of the larynx, trachea or bronchi. Of the total, 60% had abnormalities of the larynx, 16% had tracheal abnormalities and 5% had congenital anomalies of the bronchi. Significantly, 45% of the patients had more than one congenital abnormality; it is thus mandatory that a full endoscopic examination be carried out in all children presenting with congenital stridor (Shugar and Healy, 1980).

Larynx

Supraglottis

Laryngomalacia

The term 'laryngomalacia' (malacia is derived from a Greek word, malakia, indicating morbid softening of a part), was introduced by Jackson and Jackson (1942), to describe a form of congenital laryngeal stridor characterized by flaccidity of the supraglottic structures.

Although the exact pathophysiological correlates of the condition are not known for certain, the following features have been noted (Sutherland and Lack, 1897; Kelemen, 1953):

1. softness, flabbiness or lack of consistency of the laryngeal tissues
2. thinning and hypocellularity of the laryngeal cartilages
3. wrinkled, loose or redundant mucosa, especially over the arytenoid cartilages.

Laryngomalacia is by far the most common cause of congenital stridor, accounting for 60-70% of all cases (Fearon and Ellis, 1971; Holinger, 1980). The male to female ratio is approximately equal (McSwiney, Cavanagh and Languth, 1977).

In the vast majority of patients, inspiratory stridor is the only symptom. Characteristically, the stridor is high pitched, crowing and fluttering. It is usually first noticed within a few days of birth, but in some patients it may not become obvious until the child begins to be more active or develops an acute upper respiratory tract infection. The stridor tends to increase in severity during the first 8 months of life, reaching a maximum between 9 and 12 months, thereafter beginning to resolve (Lane et al, 1984). The stridor is often intermittent, appearing only while the child is feeding or crying and may be much more pronounced during sleep, especially if the child lies on its back. Hyperextension of the head sometimes results in significant lessening of the stridor.
The diagnosis can only be established for certain by direct observation of the appearance and movement of the laryngeal structures. Four significant findings are consistently noted, either separately or in combination:

(1) a tall, tubular, in-rolled epiglottis, with a tendency to prolapse backwards and which is often likened to the Greek letter omega; it should be noted, however, that this is a common finding in otherwise normal neonates and, almost certainly, merely represents an exaggeration of the infantile form (Lane et al, 1984).

(2) short, sometimes almost non-existent, flaccid, medially prolapsing aryepiglottic folds.

(3) prominent, elongated arytenoid cartilages, often covered by loose, redundant mucosa and separated by a deep interarytenoid cleft. On inspiration, the arytenoids will be seen to be sucked inwards, often crossing one over the other, obstructing the airway and giving rise to the typical stridor.

(4) the whole supraglottic larynx is deepened and narrowed with the result that the vocal cords are often quite difficult to see.

In order to observe these features properly, it is not only essential that the child is breathing spontaneously, but that the respiratory efforts should be deep and vigorous. These conditions are, perhaps, best achieved during recovery from anaesthesia. It is likewise important to avoid splinting of the laryngeal inlet; the beak of the laryngoscope must be kept in the vallecula with as little disturbance as possible to the supraglottic structures. Insertion of the tip of the laryngoscope between the aryepiglottic folds will usually result in cessation of the stridor.

One important feature of this condition is that the stridor will often become more noticeable during the early stages of anaesthesia, especially the phase of excitation; this might be explained, perhaps, by the supine position of the child under anaesthesia and the increased respiratory effort.

In most cases, laryngomalacia is otherwise asymptomatic. Occasionally, however, there may be associated feeding difficulties, sometimes severe enough to produce failure to thrive. Rarely, the child will have respiratory distress of such a degree as to require active treatment - this may take the form of tracheostomy, excision of redundant mucosa (Lane et al, 1984) or laser division of the aryepiglottic folds (Seid et al, 1985).

Although the stridor will, in most cases, have disappeared spontaneously by 18 months to 2 years, this is not invariably the case (Smith and Cooper, 1981). The author has seen children in whom both the stridor and the characteristic appearance of the larynx persisted into late childhood and adolescence.

**Laryngeal cyst**

A laryngeal (saccular) cyst is a mucus-filled dilatation of the laryngeal saccule which may distort the aryepiglottic fold, the false cord or the laryngeal ventricle (Suhonen et al,
Saccular cysts may sometimes be confused with laryngocoeles. In the former, however, there is no communication with the airway and the contents are fluid rather than gaseous.

Generally, such cysts are asymptomatic and are only noted, incidentally, during endotracheal intubation. Occasionally, however, the cysts enlarge or become infected, resulting in a rather hoarse stridor and rapidly increasing airway obstruction. Endoscopic examination reveals a large bluish swelling, in the region of the aryepiglottic fold, sometimes totally obscuring the vocal cords. Treatment is aimed at endolaryngeal excision of the cyst or, if this proves impossible, wide marsupialization. Very occasionally, tracheostomy may be required if the respiratory distress becomes severe.

Extensive cystic hygromata (lymphangiomata) can involve the posterior part of the tongue, the vallecula and the epiglottis, resulting in airway obstruction. The most satisfactory method of dealing with this problem is wide excision of the cystic masses using the CO₂ gas laser, but recurrence is common and tracheostomy may prove necessary.

**Congenital laryngocoele**

A laryngocoele is an air-filled dilatation of the ventricular sinus of Morgagni. If the sac extends beyond the limits of the thyroid cartilage, piercing the thyroid membrane, the laryngocoele is classified as being external, the internal variety remaining deep to the laryngeal cartilages. Clinically, the condition is recognized by intermittent hoarseness or respiratory distress which increases on crying or straining. Rarely, the external variety may be diagnosed by palpation of a soft, fluctuant swelling in the neck, presenting above the thyroid cartilage. Plain X-rays of the neck may demonstrate the air-filled sac.

**Glottis**

**Laryngeal web**

Formation of a normal laryngeal lumen depends upon complete canalization of the epithelial lamina between the vestibulotracheal canal above and the pharyngotracheal canal below, a process which begins at about the fifth week of intrauterine development. Total failure of this process will produce a complete laryngeal atresia but, more commonly, partial canalization occurs, resulting in the formation of a laryngeal web. Such webs may involve the supraglottis, or subglottis, but by far the greatest number (over 75%) are sited in the glottis (McHugh and Lock, 1942).

Most glottic webs are located anteriorly, involving a variable length of the vocal cords, but occasionally the membrane lies at the posterior commissure. They may be thin and membranous or, rather more frequently, thick and fibrotic, often extending downwards to involve the subglottis. The symptoms produced depend, to a degree, upon the extent and thickness of the web but stridor, which may be biphasic in timing, hoarseness of the voice or cry and respiratory distress, may all occur.

Thin webs can be lysed endoscopically using scissors, knife or laser. Thick, fibrotic webs, however, are much more difficult to treat successfully. Some authors advocate the formation, as an initial stage, of an anteriorly sited, epithelial-lined tract (Lynch and Le Jeune,
1960); others recommend insertion of a keel, either endoscopically or via a laryngofissure approach (van den Broek and Brinkman, 1979). In many cases, however, where the web is small and causing little in the way of symptoms, the best approach is to leave well alone.

**Cri-du-chat syndrome**

The most striking feature of this relatively uncommon condition is the characteristic high-pitched 'mewing' stridor. At endoscopy, the rima glottis is observed to be diamond shaped, the vocal cords are narrow and the supraglottis is curved and elongated. Respiratory distress is uncommon and tracheostomy rarely required (Ward, Engel and Nance, 1968). Chromosomal investigation confirms the diagnosis, showing partial deletion of the short arm of the fifth chromosome in group B (Cotton and Reilly, 1983).

**Vocal cord paralysis**

Congenital paralysis of the vocal cord is the third most common cause of congenital stridor, accounting for between 6% and 13% of all cases (Fearon and Ellis, 1971; Holinger, 1980). It may be unilateral or bilateral, the former occurring four times more frequently than the latter (Emery and Fearon, 1984).

In unilateral paralysis, the cry is often breathy or weak, stridor is uncommon and there is little tendency to airway obstruction. In contrast, bilateral paresis is associated with a normal cry, and stridor and significant respiratory distress are almost invariable, though these may not appear until the child becomes more active.

The diagnosis is confirmed at laryngoscopy with direct observation of vocal cord movement but this can be difficult, particularly in the small child and, undoubtedly, many cases are missed. Problems may arise from incorrectly placing the tip of the laryngoscope posterior to the epiglottis, causing distortion of the glottis or from too deep an anaesthetic casing inhibition of laryngeal reflexes. The best opportunity to observe vocal cord movement is during recovery from anaesthesia. Care must also be taken to distinguish between true paresis of the vocal cord and fixation of the cricoarytenoid joint - this may be achieved by gentle palpation of the arytenoid cartilage.

Many of these infants exhibit other congenital defects involving the central or peripheral nervous systems, the heart and great vessels or the respiratory tract. Thus neonates with hydrocephalus, meningoencephalocele, meningocele, meningomyelocele, or the Arnold-Chiari malformation, who present with stridor or abnormality of the cry, should have their vocal cords inspected. Congenital myasthenia gravis, which is characterized in the child by drooping of the eyelids (ptosis), inability to suck or intermittent facial paralysis, may present with progressive weakness of the cry and stridor. Cardiomegaly or abnormalities of the great vessels may result in stretching of the left recurrent laryngeal nerve. Either recurrent laryngeal nerve may be involved in abnormalities of the tracheobronchial tree or oesophagus.

Benign congenital hypotonia, Werdnig-Hoffman syndrome, leucodystrophy, Charcot-Marie-Tooth disease and other progressive congenital muscular disorders may demonstrate paralysis of the vocal cords (Dedo and Dedo, 1983).
Most cases of unilateral vocal cord paralysis can be treated expectantly. In the author's opinion there is no justification for the use of Teflon paste injection in the management of this condition in childhood. Bilateral paralysis will almost always necessitate tracheostomy to protect the airway, at least initially. Late recovery of one or both vocal cords has been noted - in one case this occurred at the age of 9 years (Emery and Fearon, 1984). Attempts at improving the airway by lateralization of the vocal cord should, therefore, be postponed. Reduction of one cord using the laser, with or without arytenoidectomy, may be an effective alternative.

Subglottis

Subglottic stenosis

It is often difficult to distinguish between the congenital and acquired varieties of this condition; indeed the two frequently coexist. Stenosis may be considered to be present if the diameter of the subglottic space is less than 3.5 mm, as measured by failure to achieve the unforced passage of a bronchoscope of this size (Fearon and Cotton, 1972). The abnormality lies in the cricoid cartilage which is thickened and oval in shape. There is also a concomitant increase in the thickness of the submucosa, giving rise to the characteristic crescentic narrowing of the anterior subglottic space, maximal 2-3 mm below the true vocal cords. Occasionally, this submucosal thickening extends upwards to involve the cords at the anterior commissure.

Congenital subglottic stenosis demonstrates a wide variation, both in severity and in symptomatology. Airways resistance changes as the fourth power of the radius (Poiseuille's law); as a consequence, even minor degrees of subglottic oedema will produce a disproportionately large increase in airways resistance. Thus, in mild cases of subglottic stenosis, there may be no stridor until the child develops an upper respiratory tract infection or the subglottis is traumatized by endotracheal intubation. In such patients, the stridor is usually inspiratory in nature, whereas, in the more severe case, it is often biphasic. Every once in a while, a child will present with such severe stenosis that the subglottic airway is reduced to pinhole dimensions. Remarkably, a few of these children, while noted to be stridorous, do not appear to get into respiratory difficulty until infancy has passed and the ambulatory phase of childhood has begun.

Demonstration of the characteristic endoscopic findings, referred to above, and the absence of a history of previous prolonged endotracheal intubation, provide the mainstays of diagnosis. Plain X-rays of the neck and thoracic inlet are notoriously unreliable, with many instances of both false-positive and false-negative findings being reported. Considerable care and gentleness must be employed during bronchoscopy in these patients, in order to avoid damage to the subglottic area, otherwise the airway may be further compromised, precipitating the need for tracheostomy. It is the author's practice to record the diagnosis clearly on the front of the patient's chart to warn anaesthetic personnel should the child require endotracheal intubation in the future.

There is a tendency towards spontaneous improvement in congenital subglottic stenosis and, in mild cases, it is often sufficient to advise the parents that problems are only likely to arise during upper respiratory tract infections or following intubation for anaesthesia. In more
severe cases tracheostomy may be required and should be performed earlier rather than later. Treatment consists of either serial dilatation or laryngotraceoplasty, although the former method is of much less value in congenital stenosis than in the acquired form of the disease. For further details on management, the reader is referred to Chapter 34.

**Subglottic haemangioma**

Congenital haemangiomas are hamartomata of blood-vessel origin. The association between skin haemangiomas and vascular tumours affecting internal organs is well recognized (Garfinkel and Handler, 1980). The best known of these symptom complexes is the Sturge-Weber syndrome, in which 'port wine stains' in the area of distribution of one trigeminal nerve are coupled with angiomata of the cerebral cortex.

Similarly, laryngeal haemangiomas, almost always affecting the subglottis, may appear in conjunction with capillary naevi of the face or neck. Symptomatically, the affected child presents with inspiratory or biphasic stridor, dyspnoea and a rather harsh cry. On direct laryngoscopy, the distinctive appearance of the subglottis is almost diagnostic. There is a bluish, pear-shaped swelling arising from the lateral wall of the subglottic space often extending upwards to involve the undersurface of the vocal cord. In most cases the subglottis is affected on one side only, but occasionally there are bilateral tumours. Although often associated with facial lesions, subglottic haemangioma may also be present in isolation. The tumours are soft and compressible and can be easily bypassed with the bronchoscope. In the author's experience, the angiomata are always of the capillary type, do not bleed and may, therefore, be safely biopsied to provide a tissue diagnosis.

Like the facial haemangiomas, the subglottic lesions often rapidly increase in size during the first few years, thereafter spontaneously regressing. A tracheostomy is usually necessary, at least initially, though some authors have claimed that this can be avoided by using steroid therapy (Cohen and Wang, 1972; Sadan, Sade and Grunebaum, 1982); others, however, disagree (Leikensohn, Benton and Cotton, 1976). Laser excision, unfortunately, has not proved to be efficacious (Healy et al, 1980).

**Laryngotracheal cleft**

The respiratory primordium is derived from the foregut at about day 20 of embryonic life by the development of the median pharyngeal groove. Subsequently, the groove deepens to form the pharyngotracheal canal, becoming separated from the oesophagus by development of a tracheo-oesophageal septum which starts caudally and grows in a cephalad direction. Failure in the formation of this septum, or arrest of its rostral advancement, will result in open communication between the laryngotracheal airway and the oesophageal lumen (van den Broek and Brinkman, 1979).

Pettersson (1955) has classified these clefts into three types:

1. laryngotracheal cleft
2. partial laryngotracheo-oesophageal cleft
3. total laryngotracheo-oesophageal cleft.
The hallmark of this condition is persistent aspiration, sometimes accompanied by stridor, respiratory distress and a toneless cry. Diagnosis is difficult and is frequently missed; in one series, 42% were diagnosed at autopsy (Burroughs and Leape, 1974). Screening of the swallow using Gastrograffin or dilute barium will demonstrate aspiration, although the precise site of occurrence may be difficult to determine and is, of course, found in other conditions, such as H-type tracheo-oesophageal fistula. Any patient in whom the possibility of a cleft is considered should be examined, using the laryngeal microscope, and the interarytenoid cleft palpated using a blunt hook or similar instrument. The defect between the larynx and oesophagus may be repaired via a lateral pharyngotomy (Kauten, Konrad and Wichterman, 1984).

Laryngotracheal clefts occur in isolation, but may also be associated with other congenital abnormalities as in the ‘G’ or Opitz-Frias syndrome (Opitz et al, 1969). This is an autosomally dominant disease with male predominance, characterized by craniofacial, aerodigestive and urogenital anomalies. The head and neck manifestations are related to midline defects, including cleft lip and palate, laryngotracheal cleft and neuromuscular dysfunction of the pharynx and oesophagus (Kimmelman and Dennen, 1982).

Anterior clefts of the larynx, due to failure of fusion of the laminae of the thyroid cartilage, creating an anterior midline defect, have also been described (Montgomery and Smith, 1976). It should be noted, however, that these are glottic rather than subglottic.

**Trachea and bronchi**

Abnormalities of the tracheobronchial tree account for about 26% of congenital causes of stridor (Holinger, 1980).

**Agenesis**

Complete or partial agenesis of the trachea is incompatible with life. Short-term survival may be possible, however, if there is a fistulous connection between the oesophagus and bronchus, but utilization of the oesophagus as a tracheal replacement has proved unsuccessful (Peison, Levitsky and Sprowls, 1970). Agenesis of one main bronchus and its associated lung is, however, survivable, although most affected infants are weakly and tend to succumb to chest infections. In the majority of cases, there are other severe, associated congenital abnormalities which further diminish the chances of survival of these patients beyond the neonatal period.

**Stenosis**

Congenital narrowing of the tracheal or bronchial lumen may take the form of membranous webs, segmental or whole organ stenosis. Where the obstruction is sited in the lower trachea or main bronchi, treatment is confined to attempts at gentle dilatation using bronchoscopes of increasing diameter. Rather surprisingly, such management is often successful and should certainly be tried. Stenosis of the upper trachea may be managed in the same way or recourse may be had to tracheoplasty where the lumen is increased by making a vertical incision in the anterior tracheal wall and inserting an elliptical wedge of costal cartilage. Where the stenosis is affecting a long segment of the trachea, an attempt may be
made to increase the lumen by separation of one side of the trachea from the oesophagus and reattachment more laterally.

**Tracheomalacia**

Tracheomalacia exists in both generalized and localized forms, of which the latter is much more common. Although there are some pathological correlates between tracheomalacia and laryngomalacia and, occasionally, they may coexist, there is no proven relationship between the two. The characteristic stridor, which is high pitched and expiratory is said to resemble the expiratory wheeze of asthma (Baxter and Dunbar, 1963). In the localized form, cough is a frequently associated symptom and is harsh and barking in quality, rather like that of viral croup. At bronchoscopy, the trachea is seen to be compressed in its anteroposterior diameter due to a flattening of the anterior aspect of the cartilaginous rings. This abnormal appearance is accentuated by expiration and even more so by coughing, when the anterior and posterior walls may come in contact. Care must be taken, however, to distinguish between this anomaly and that produced by laxity of the trachealis muscle with forward ballooning of the posterior wall - an almost universal finding in the neonate.

As with laryngomalacia, complete spontaneous recovery is the rule. In a few cases, however, where approximation of the anterior and posterior walls is marked, severe obstruction to the airway may be present and a tracheostomy found to be necessary. In these circumstances, it is often imperative to use a longer than usual tracheostomy tube, one which will reach to just above the carina, otherwise the infalling anterior wall may block the end of the tube (Cinnamond, 1977). Suspension of the anterior tracheal wall from the inner surface of the sternum has also been recommended.

In a high proportion of cases of the generalized type, the malacic process will be found to be also affecting the main bronchi. In about 10% of cases, additional abnormalities of the trachea will be present, especially tracheo-oesophageal fistulae, which are considered in Chapter 37. In every case, a careful search should be made for the tracheal opening of an H-type tracheo-oesophageal fistula as this condition may otherwise be missed.

The localized form of the disease is almost always due to compression of the affected portion of the anterior tracheal wall from without. The most probable causes are vascular rings or abnormal vessels, congenital mediastinal or cervical tumours or bronchogenic cysts.

**Vascular compression**

The primary cause of anomalies of the great vessels of the neck and thorax is faulty embryonic development and the only adequate way to classify them is embryologically. Desnos et al (1980), however, have produced a simplified but satisfactory classification based on endoscopic findings.

**Vascular ring**

(1) Double aortic arch, in which the ascending aorta divides into two arches, one passing to the right of the trachea and the other to the left, rejoining posterior to the oesophagus to form the descending aorta. The trachea and oesophagus are thus confined
within a compressing ring of vascular structures. There is considerable variation in the morphology of these rings, the arches may be of equal or unequal size and different configurations of the main branches may occur.

(2) Neuhauser's anomaly, where the aorta is single but passes to the right of the trachea. In this case the ring is only partly vascular, the component to the left of the trachea being formed by the ligamentum arteriosum, the remnant of the ductus arteriosus which, in the fetus, connects the pulmonary artery to the descending aorta.

**Vascular sling**

Here, the left pulmonary artery, instead of passing anterior to the trachea, passes between it and the oesophagus, compressing the trachea from behind and the oesophagus from in front.

**Anterior compression**

(1) Compression of the anterior tracheal wall by an anomalous innominate artery, the origin of which from the aortic arch is more posteriorly sited and more to the left than normal. This produces a characteristic sloping compression of the lower trachea 1-2 cm above the carina, more marked on the right anterolateral aspect. Further confirmation is afforded by noting diminution or absence of the patient's right radial or right carotid pulses, using the tip of the bronchoscope to collapse the vessel against the sternum. On rare occasions, the anomalous innominate artery may be associated with an aberrant right subclavian artery, which passes posterior to the oesophagus (Macdonald and Fearon, 1971).

(2) A larger than normal pulmonary artery may compress the trachea and bronchi at or just below the carina.

**Posterior compression**

This is usually due to an aberrant right or, more rarely, left subclavian artery, passing posterior to the oesophagus. The oesophagus, alone, is compressed in this case.

In those instances where both trachea and oesophagus are involved, the patient may present with symptoms that can be referred to either; in practice, however, the tracheal symptoms of stridor, dyspnoea and a harsh, brassy cough tend to dominate the picture. Similar symptoms are found where the trachea alone is affected. In about one-third of those children in whom there is significant airway obstruction due to innominate artery compression, stimulation of the area of compression with the tip of the bronchoscope may initiate reflex apnoea (Moes, Izuakwa and Trusler, 1975). It has been suggested that activation of this reflex may explain some cases of sudden infant death syndrome.

Barium swallow may show indentation of the oesophagus - this will be bilateral when the abnormality is a double aortic arch or Neuhauser's anomaly, anteriorly in vascular sling or posteriorly with an aberrant subclavian artery. High-resolution ultrasound scanning is often helpful and computerized tomography may give additional information, but the conclusive investigation, in all cases is aortography.
In those patients in whom the symptoms are severe and, especially, when the tracheal airway is judged to be inadequate, as demonstrated by an inability to see the carina from a position proximal to the compression, surgical decompression should be undertaken. In almost all cases of vascular ring, surgery will be necessary, the definitive management being division of the lesser component of the ring. Where the compression is due to an anomalous innominate artery, the vessel may be slung anteriorly away from the trachea by suturing the adventitia to the undersurface of the sternum (Gross and Neuhauser, 1948; Mustard et al, 1969). This, however, is seldom necessary and indeed, in the neonate, some degree of innominate artery compression of the trachea is so common as to be the rule rather than the exception.

In all types of vascular compression, tracheostomy should be avoided as bypassing the obstruction may result in intubation of the right main bronchus and, in addition, there is significant, and usually fatal, risk of erosion of the vessel by the tip of the tracheostomy tube. In all cases where surgical correction of the deformity has been undertaken, it should be borne in mind that the localized area of tracheomalacia, which invariably accompanies compression, may remain for many months postoperatively.

**Anomalous bronchial bifurcations**

Origin of the right upper lobe bronchus from the right lateral wall of the trachea above the carina is relatively common. In almost every case this is an incidental finding and is entirely asymptomatic. Bronchography may be required to delineate the exact morphology. Other minor variations in the bronchial tree also occur and are, likewise, symptom free.

**Congenital cysts and tumours**

These cause non-pulsatile compression of the trachea and main bronchi but their presentation and appearance on endoscopy is otherwise similar to vascular compression.

Tracheogenic and bronchogenic cysts are thought to originate from evaginations of the primitive tracheal bud. They are lined with respiratory epithelium and may contain thick, inspissated mucus. In contrast to bronchogenic cysts, those arising from the trachea do not usually communicate with the lumen. Infection may occur, with resulting increased compression of surrounding structures. Thoracotomy and excision of the cyst will often be required.

Cervical and mediastinal cysts or tumours, including thymomata and teratomata may compress the trachea or bronchi from without (Mills and Hussain, 1984). Teratomata affecting the anterior neck present particular difficulties as the tumour may intimately involve the anterior wall of the trachea. Complete excision of the mass is likely to entail removal of part of the tracheal wall with the possibility of long-term airway stenosis.