Clinical Aspects of Congenital Anomalies of the Trachea and Bronchi

PAUL H. HOLINGER, M.D., F.C.C.P.* and KENNETH C. JOHNSTON, M.D., F.C.C.P.*

Chicago, Illinois

Congenital malformations of the trachea, bronchi and lungs may produce bronchopulmonary problems of unusual and challenging clinical interest. In the first hours, days or weeks of life, severe, acute respiratory emergencies occur as the result of congenital deformities. Some are correctable if promptly recognized, others are incompatible with life, while probably the majority are symptomless, appearing as incidental findings on routine medical, surgical or post-mortem examination. A resume of this problem is presented because of the increasing frequency with which it is being encountered and to emphasize the newer diagnostic and therapeutic aspects, both bronchoscopic and surgical, of the many corrective procedures.

Classification

A simplified, practical classification of lower respiratory tract malformations serves as a basis for this discussion.1 It has been developed for clinical purposes and omits anomalies of monsters and malformations incompatible with life. The modified classification is as follows:

I. Anomalies of the Trachea
   A. Constrictions and Enlargements
      1. cartilage deformity
      2. anomalous vascular compression
      3. webs.
   B. Evaginations and Outgrowths
      1. tracheoceles, diverticuli, cysts
      2. fistulae
      3. tracheal lung
   C. Abnormal bifurcation or deviation

*From the Department of Otolaryngology, University of Illinois, College of Medicine, St. Luke’s Hospital and the Department of Bronchoesophagology, Children’s Memorial Hospital, Chicago.
Presented before the Fourth International Congress on Diseases of the Chest, Cologne, Germany, August 19-23, 1956.

Copyright, 1957, by the American College of Chest Physicians
II. Anomalies of Bronchi and Lungs
   A. Agenesis and atresia
   B. Constrictions and enlargements
      1. Webs
      2. Compression from cardiovascular anomalies
   C. Evaginations or outgrowths.
      1. Bronchoceles, diverticuli
      2. Cysts and emphysematous lobes
      3. Fistulae
   D. Subnumerary bronchi, lobes, fissures
   E. Supernumerary bronchi, lobes, fissures
   F. Anomalous bronchial and lung tissue attached to respiratory system.
   G. Anomalous bronchial and lung tissue attached to tissue other than respiratory system.
   H. Other gross anomalies
      1. Situs Inversus.

   1. Anomalies of the Trachea

   The most common anomalies of the trachea are the stenoses or webs which involve elements of the tracheal wall. Fibrous strictures appear as webs (Fig. 1) or as fusiform segments of stenosis with deformity.
(Fig. 2). Absence of one or more of the tracheal cartilages results in varying degrees of dyspnea with severe inspiratory stridor but a clear cry. Lateral x-ray films of the neck and chest may give the tentative diagnosis, while bronchoscopic inspection of the dyspneic newborn infant will confirm the diagnosis. Tracheotomy may be necessary to alleviate the symptoms. Heavy cartilaginous overgrowth or other deformity of the cricoid cartilage or first tracheal ring is the most typical site of anomalous cartilage enlargement.

Stenosis of the trachea may accompany congenital esophageal atresia and tracheoesophageal fistula. In these cases the area of constriction is found at the level of the fistula between the trachea and the lower esophageal segment.

Of recent interest is the frequency in which stenosis associated with cardiovascular anomalies is recognized. In this important group of cases, the heart or an anomalous great vessel constricts the trachea or a bronchus during development. Many anomalous vessels may contribute to the obstruction to cause a constricting vascular ring around the trachea. A double aortic arch is the simplest of these. In others a patent ductus becomes a part of the ring. Pressure from an abnormally originating right subclavian artery may compress the trachea to cause obstruction. These conditions have long been recognized; they are frequently revealed by bronchoscopy and bronchography, during surgical exploration or at autopsy. Of greatest importance, however, from a therapeutic standpoint, is the finding of tracheal or bronchial cartilage absence or deformity at the site of vascular constriction. Removal of the obstructing vessel, or ligation and severance of a portion of the vascular ring relieves the pressure on the airway, but abnormal narrowing persists. Tracheotomy and temporary advance of a polyethylene tube through the narrow trachea or bronchus serves to give necessary support to the lumen until the wall becomes rigid or firm enough to maintain itself as an airway. (Fig. 3)

Several types of tracheal evaginations are recognized in this study; tracheoceles are demonstrated as the cause of severe respiratory and esophageal obstruction both by means of roentgen study and endoscopy. Resection is the only therapeutic procedure to be considered, giving complete relief of symptoms. It would appear that such cysts are pinched-off aberrant bronchial or tracheal buds in view of the character of their mucosal lining. They are of early embryonic origin.

The common form of tracheal evaginations are the fistulae associated with congenital esophageal atresia. A congenital fistula without atresia is relatively rare but falls into this category. Other tracheal evaginations consist of accessory bronchi with their pulmonary lobes or segments originating directly from the trachea; an accessory right upper lobe bronchus is most frequent. This may appear as a complete duplication of the right upper lobe or as a tracheal origin of only the apical segment and its bronchus. Of five such cases in this series, the anomaly was an incidental finding in four while in the fifth, a congenital lung cyst in the anomalous lobe led to bronchoscopic and bronchographic studies that revealed the nature of the anomaly.
Abnormal bifurcation of the trachea has been observed in two cases. In one, a four month old infant, the bronchoscopic examination showed the right main bronchus to originate from the medial wall of the left bronchus opposite the orifice of the left upper lobe. This observation was confirmed on surgical exploration, done because of tracheal compression. An enlarged thymus was found compressing the trachea. In addition, there was an anomalous subclavian artery lying anterior to the thymus. In the second case of abnormal bifurcation of the trachea, an infant with multiple congenital anomalies, both a right and left upper lobe originated directly from the trachea with three lobes in each lung.

II. Anomalies of the Bronchi and Lungs

Anomalies of the bronchi and lungs consist of various forms of agenesis, duplication, constriction and evaginations. Complete agenesis of one lung is fairly common and has been observed in five cases;\(^5\) in two, the absence of the right lung and in one, the left, was found shortly after birth when bronchoscopic examinations were requested because of dyspnea, absence of breath sounds, density on chest x-ray film and marked shift of the heart and mediastinum. An atelectasis of an entire lung, possibly due to a mucus plug had been suspected. In the fourth case, the absence of the left lung was found on examination of a seven year old boy suspected of having aspirated foreign body. In the fifth case, the absence of the left lung was found at post-mortem examination associated with an absence of the left half of the diaphragm, the abdominal contents filling the left hemithorax. The autopsy also confirmed the diagnosis in two of the first three cases, while the third is still living; multiple congenital anomalies

**FIGURE 2:** Deformed, flattened tracheal and bronchial cartilages causing fatal obstruction.
were associated with the bronchopulmonary anomaly in three that died. Early embryonic disturbances were evidently the cause.

Constrictions or enlargements of the bronchi follow the pattern of similar anomalies of the trachea. Congenital bronchial webs, bronchomalacia and compression or constriction due to cardiovascular anomalies may be responsible for severe respiratory distress. The bronchial deformity which arises during embryonic development through a constricting anomalous vessel was found to be of particular importance. Actual deformity or permanent absence of cartilage is noted in these cases. This is of clinical significance as it explains why surgical removal of the obstructing vessel may reduce the degree of obstruction, but does not necessarily eliminate it entirely. Dilatation and temporary support of the bronchus by the placement of a polyethylene tube beyond the site of the obstruction should be reiterated. (Fig. 3)

Bronchial evaginations or outgrowths consist primarily of fistulae, bronchogenic cysts, diverticuli, bronchoceles, and the numerous congenital cystic malformations of the lungs. The latter constitute a large variety of lesions which present many symptoms and unusual gross and microscopic findings. They may be associated with bronchial as well as vascular anomalies. Large solitary cysts, for example, may be found in an accessory lobe or segment, attached to the bronchial system by an anomalous bronchus. Rapidly expanding cysts may be responsible for acute dyspnea in infants necessitating emergency thoracotomy and lobectomy because the expanding cyst compresses the remaining thoracic contents. The thoracotomy should be preceded by bronchoscopic inspection in order to rule out the possibility of a congenital bronchial stenosis or web.

An extremely interesting clinical entity, somewhat similar in its action to the congenital tension cysts has recently been recognized. The condition consists of an emphysema of one lobe which gradually increases the size of the lobe to cause compression of the remaining pulmonary tissue. Generally observed in the newborn period, the clinical picture is one of dyspnea and cyanosis with x-ray films suggesting a severe obstructive emphysema of the affected lobe. (Fig. 4) The absence of bronchoscopic evidence of bronchial obstruction, either by extra-bronchial compression or intra-bronchial web formation is an indication for a thoracotomy and removal of the emphysematous lobe. Histologic study of such surgically-removed lobes has demonstrated an alveolar fibrosis which may represent an important etiologic factor. The condition cannot be clinically differentiated from obstructive lobar emphysema due to vascular compression of a bronchus or to bronchial cartilage abnormality other than by bronchoscopic inspection or surgical exploration.

Supernumerary bronchi, lobes and fissures are fairly commonly encountered abnormalities, although description of these anomalies obtained from a review of the literature, as well as an analysis of our own records, show that aberrant lobation seldom coincides with bronchial variations. Pulmonary hypoplasia, in the form of subnumerary lobes, is more frequently associated with other serious developmental defects than were other anomalies herein described.
 Supernumerary lobes, bronchi and fissures were most frequently observed as accessory lower lobes. Supernumerary fissures without additional lobes are common surgical observations and of little clinical significance, the superior and medial basal segments being most frequently established as separate lobes. An accurate statistical evaluation of the frequency of these anomalies is difficult to obtain, since the finding is merely incidental and not apparent prior to surgery or post-mortem examination. Anomalous lung tissue attached to respiratory organs is relatively rare. An anomalous lobe with no bronchial communication was recently observed, receiving its aeration through direct alveolar continuity with adjacent pulmonary tissue. Because of increasing dyspnea, the emphysema was recognized and the lobe resected.

Anomalous bronchial and lung tissue attached to other than respiratory organs is occasionally observed. An example of this type of anomaly, reported elsewhere, consisted of a bronchus and lung tissue originating from the esophagus. The fistulous bronchial tract was observed esophagoscopically, and the diagnosis confirmed on thoracotomy and lobectomy. Embryologically, such a case could indicate a strange and rare anomaly. It could be explained on the assumption that the primitive foregut had retained or acquired multiple potentialities of forming tracheobronchial evaginations. Most likely, the aberrant primordium had arisen, in very early embryonic life, close to the normal laryngotracheal groove but,

**FIGURE 3**

*Figure 3:* Polyethylene tube inserted through tracheotomy into right lower lobe bronchus of six weeks old infant maintaining patency of bronchus following resection of compressing vascular ring. Absence of cartilage at site of vascular compression necessitated temporary bronchial support.—**FIGURE 4:** Emphysematous left upper lobe causing extreme cardiac and mediastinal shift in an infant two weeks of age. The resected lobe showed severe emphysema and an alveolar fibrosis.
secondarily, had become separated from it by the growth in length of the esophagus.

Discussion

The diagnosis of congenital malformations of the lower respiratory tract during life is made with the help of various services and the team work of pediatrician, roentgenologist, bronchologist and thoracic surgeon. The history of rubella during the first trimester is important. From the physical findings one may differentiate a laryngeal anomaly from other obstructions to respiration by the character of the cry and by direct laryngoscopy. In evaluating a pulmonary lesion suspected of being a congenital anomaly, physical findings of the chest must be correlated with the interpretation of the x-ray film. Specifically, an obstructive emphysema of one lung with compression atelectasis of the opposite lung, requires such correlation to be differentiated from an obstructive atelectasis of one lung with compensatory emphysema of the opposite lung. An erroneous interpretation will be obtained if one relies on a single chest film, the report stressing the atelectasis of a relatively uninvolved side rather than the much more significant obstructive emphysema of the affected side. Fluoroscopic study, x-ray films in inspiration and expiration, a lateral view with the arms down and back to reveal the larynx, a barium swallow, bronchography, planography, and angiocardiography will add significant information in special instances. Bronchoscopy without anesthesia is now an accepted diagnostic procedure in unusual respiratory conditions in infants. Limiting the time of the procedure, the use of small instruments and the administration of oxygen through the scope during the examination increase the margin of safety. Finally, exploratory thoracotomy in infants, now a feasible, well established procedure, increases the scope of diagnosis and adds therapeutic possibilities which were unavailable until recent years.

As in most studies of congenital anomalies, the review of anomalies of the lower respiratory tract has shown frequent associations with anomalies of other organs and systems, as well as a frequent multiplicity of anomalies within the respiratory tract itself. The formative genesis of many of these anomalies can be explained on the basis of an understanding of the normal embryology of the lungs. The causative factors, in most instances, remain obscure. It is emphasized that truly congenital anomalies arise early in embryonic life, most likely before the end of the second month of intra-uterine development. Abnormal conditions arising during the later fetal life are pathologic rather than teratogenic in nature.

SUMMARY

Congenital anomalies of the lower respiratory tract are described and classified and the clinical aspects are then discussed. Anomalies of the trachea include constrictions and enlargements, evaginations and outgrowths and abnormal bifurcation or deviation. Anomalies of the bronchi and lungs which are described include agenesis, constrictions, evaginations, subnumerary and supernumerary bronchi, lobes and fissures and anom-
lous bronchial and lung tissue attached to the respiratory or other systems. The clinical significance of the anomalies is found to be greatest in the newborn period when the acute respiratory symptoms they may cause require prompt diagnostic studies, including bronchoscopy and exploratory thoracotomy. Particular stress is placed on the clinical picture caused by varying degrees of bronchial obstruction due to cardiac and vascular malformations. The discussion emphasizes the importance of teamwork between pediatrician, roentgenologist, bronchologist and thoracic surgeon in the management of the interesting problem presented, and the fact that when one gross anomaly is found, there are frequently other serious malformations of other systems that should also be recognized.

RESUMEN

Se describen y clasifican las anomalías congénitas de las vías respiratorias bajas, así como se discuten sus aspectos clínicos.

Las anomalías de la tráquea incluyen constricciones y ensanchamientos, evaginaciones e hiperтроfias, y bifurcación anormal o desviación.

Las anomalías de los bronquios y de los pulmones que se describen son: agenesia, constricciones, evaginaciones, bronquios subnumerarios y supernumerarios, así como lóbulos y fisuras, y tejido bronquial y pulmonar anómalos adheridos a los aparatos respiratorio u otros. La significación clínica de las anomalías, se encuentra que es mayor en el período del recién nacido cuando los síntomas respiratorios agudos que ellas causan pueden requerir un diagnóstico pronto mediante broncoscopia y toracotomía exploradora. Se destaca principalmente el cuadro clínico que causan grados diversos de obstrucción bronquial debida a malformaciones cardiacas y vasculares.

La discusión enfatiza la importancia del trabajo en equipo incluyendo el pediatra, radiólogo, el broncólogo, y el cirujano de tórax para hacer frente a los interesantes problemas que se presentan y el hecho de que cuando se presenta una anomalía grande hay frecuentemente otras serias malformaciones en otros aparatos o sistemas, debe tenerse presente.

RESUME

Les auteurs décrivent les anomalies congénitales de l’arbre respiratoire inférieur, les classent, et en discutent les aspects cliniques. Les anomalies de la trachée comprennent les sténoses et les dilatations, les évaginations, les excroissances et les bifurcations anormales ou déviations. Les anomalies des bronches et des poumons qui sont décrites comprennent l’agenésie, les étranglements, les évaginations, les bronches absentes et surnumeraires; les lobes et scissures, les anomalies des tissus bronchiques et pulmonaires liées au système respiratoire et aux autres systèmes. Les auteurs trouvent que la conséquence clinique des anomalies est plus grande dans la période qui suit immédiatement la naissance, lorsque les symptômes aigus respiratoires qu’elles peuvent causer nécessitent des études diagnostiques rapides, comprenant la bronchoscopie et la thoracotomie exploratrice. Ils insistent particulièrement sur le tableau.
clinique que réalisent les divers degrés d’obstruction bronchique causée par les malformations cardiaques et vasculaires. Ils mettent en évidence l’importance du travail d’équipe entre pédiatre, radiologue, bronchologiste et chirurgien thoracique dans la conduite du problème intéressant qui est discuté ici, et le fait que, lorsqu’une anomalie majeure est découverte, il y a fréquemment d’autres malformations graves des autres systèmes qui doivent aussi être reconnues.

ZUSAMMENFASSUNG


REFERENCES