CONGENITAL AND DEVELOPMENTAL ANOMALIES OF THE LUNG

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Seminar outline

- Introduction
- Embryology of lung development
- Classification
- Individual anomalies

Introduction

- Developmental anomalies of the lung are usually detected in the neonatal period and in early childhood.
- Some are not encountered until later childhood or adulthood.
- Some can be confused with more sinister abnormalities.
- An understanding of their imaging features & presentation important for the physician.

Relevant Embryology

- Intrauterine development: Embryonic, pseudoglandular, canalicular & saccular (alveolar).
- 26th day gestation - Ventral diverticulum of the foregut
- Next 2 days, the right and left lung buds arise from this outpouching
- Respiratory portion of the gut becomes separated from the esophageal portion by tracheoesophageal septum.
- Lung buds elongate into primary lung sacs & the 5 lobar bronchi appear.(Upto 5th week – Embryonic phase)

Relevant Embryology

- 5 lobar bronchi branch in a dichotomous fashion
- By 16th week, virtually all of the conducting airways are present.
- Airways are blind tubules lined by columnar or cuboidal epithelium – Pseudoglandular (5 -16 th week)
- Canalicular period (17th to 25th–28th weeks)
- Saccular (alveolar) period – Alveoli demonstrated as early as 30 weeks gestation.
- Final period of normal intrauterine lung development from 36 weeks to term - Prolific development of alveoli.
- Postnatal period - Alveolar development continues

CLASSIFICATION

BRONCHOPULMONARY (LUNG BUD) ANOMALIES	COMBINED LUNG & VASCULAR ANOMALIES	VASCULAR ANOMALIES
A. Congenital bronchial atresia	A. Hypogenetic lung (scimitar) syndrome	A. Absence of a main pulmonary artery
B. Congenital lobar emphysema	B. Bronchopulmonary sequestration, both intralobar and extralobar	B. Anomalous origin of the left pulmonary artery from the right
C. Congenital cystic adenomatoid malformation		C. Anomalous pulmonary venous drainage (partial or complete)
D. Pulmonary bronchogenic cysts (BPFM)		D. Pulmonary arteriovenous malformation
E. Tracheal bronchus		
F. Accessory cardiac bronchus		
G. Tracheomalacia		
H. Tracheal stenosis		
I. Pulmonary underdevelopment		

Bronchial atresia

- Focal obliteration of a proximal segmental or subsegmental bronchus.
- Lacks communication with the central airways
- Development of distal structures is normal.
- Most often affects segmental bronchi at or near their origin.
- Bronchi distal to the stenosis become filled with mucus → bronchocele.

Bronchial atresia

- Alveoli → ventilated by collateral pathways and show features of air-trapping→ region of hyperinflation around the dilated bronchi.
- May be acquired postnatally - Traumatic/ postinflammatory insult .
- Upper-lobe bronchi are more frequently affected.
- Usually asymptomatic incidental finding in approximately 50% of cases, mostly in young men
- Dyspnea, pneumonia, and bronchial asthma have been reported

Bronchial atresia

- Characteristic chest radiographic finding

  Bronchocele → Rounded, branching opacities radiating from the hilum.

- CT - Sensitive modality for demonstrating the typical features → Round opacity at the site of the atresia, medial to the air trapping.

- Clearly depicted by performing expiratory CT.

- Newborns – Fluid filled mass → Later characteristic features when fluid disappears.

CT - Excellent modality for excluding the presence of a hilar mass.

Basal – Contrast enhanced spiral CT - exclude a vascular component- PS

MR imaging – Can delineate bronchocele but not air trapping

Congenital lobar emphysema

- Progressive overdistention of a lobe, sometimes two lobes.
- Check-valve mechanism at the bronchial level - Progressive hyperinflation of the lung.
- May be associated with anomalies of the cardiovascular system in 12%–14%
- More common among males, not familial and occurs predominantly in Caucasians
- Most patients become symptomatic during the neonatal period, most before 6 months of age

Congenital lobar emphysema

- Chest radiograph , CT - Primary imaging tool.
- Predilection for the upper lobes and right middle lobe.
  Lower lobes involved <1%
- The appearance on the chest radiograph depends on timing.
- Symptomatic in infancy (type I), older children (type II), or incidental finding in asymptomatic patients (type III). Types II and III are rare (Myers)
- Accounts for about 50% of all congenital lung malformations
- Pathologic variant of CLE - Polyalveolar lobe.

Congenital lobar emphysema

- D/d Pneumothorax - Attenuated lung markings are seen in the overinflated lobe.
- Compression of adjacent lobes pushes them cephalad or caudad. / Pneumothorax - Lung collapses around the hilum.
- Symptomatic patients undergo lobectomy.
- Asymptomatic children/minor symptoms – conservative management

Congenital cystic adenomatoid malformation (CCAM)

- Uncommon cause of respiratory distress in neonates and infants.
- Overgrowth of bronchioles, with almost complete suppression of alveolar development.
- Communication between the individual cysts within the CCAM and also with the tracheo-bronchial tree.
- CCAMs - Equal frequency in the upper & lower lobes
- Typically, they are unilobar
- Less frequent in the right middle lobe.

Stocker's classification

CCAM I (70 %)	**1 or more cysts > 2 cm in diameter, surrounded by multiple smaller cysts. Lined by Ciliated columnar epithelium**
CCAM II (15-20 %)	Cysts measuring up to 2 cm in diameter. Lined with cuboidal or columnar epithelium
CCAM III	Usually contain cysts less than 0.5 cm in diameter Lined by cuboidal epithelium.

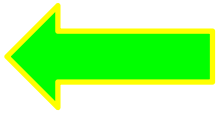
Congenital cystic adenomatoid malformation (CCAM)

- Type II CCAMs – Associated congenital anomalies, including renal agenesis and dysgenesis, cardiac malformations, and pulmonary sequestrations.
- Type III CCAMs - Rarely seen postnatally , Poor prognosis.
- Variation in prenatal presentation - Incidental finding at routine prenatal ultrasonography to severe hydrops with mass effect and mediastinal shift.
- Postnatally - One-half to two-thirds will have some form of respiratory distress or compromise.
- Older patients - Recurrent pulmonary infections.

Management

- Areas of controversy.
- Symptomatic neonates with respiratory distress

  Older children and adults with recurrent pneumonia

  SURGERY 
- Radiologic CCAM, Asymptomatic - CONTROVERSY

  Risk of infection developing in a CCAM

  Several case reports of malignancy arising in CCAM.

  Bronchoalveolar carcinoma, pleuropulmonary blastoma,

  rhabdomyosarcoma, and bronchogenic carcinoma reported

BPFM
(Bronchopulmonary foregut malformations)

- Developmental abnormalities derived from the embryonic foregut.
- Duplication cysts arising anywhere from the pharynx to the duodenum

  Neurenteric cysts

  Bronchogenic cysts

  Pulmonary sequestrations, and occasionally CCAMs and CLE.

- May or may not communicate with the lumen of the GI tract or the airway.
- No complete unifying embryologic theory to explain their origin.

Bronchogenic cyst

- Proposed theory of origin

  Arise secondary to abnormal budding of the primitive ventral foregut, early in fetal life.

- Location - Mediastinum (Commonest – 70 %)

  Pulmonary parenchyma.

  Rarely - Neck, pericardium, or abdominal cavity

- Uncomplicated cysts - Do not usually communicate with the tracheobronchial tree.

- Mediastinal type - Subcarinal, hilar, or right paratracheal locations most commonly.

- Intrapulmonary cysts - More likely to be right-sided.

Bronchogenic cyst

- Symptoms
  - Incidental finding
  - Symptomatic infants – Respiratory distress
  - Older children - Infected cysts
  - Spontaneous pneumothorax – Rarely

- Complications

  Infection / hemorrhage /erosion into adjacent structures.

  Rarely - Malignancy within the walls of the cyst

  Rhabdomyosarcoma,

  pulmonary blastoma, anaplastic carcinoma, leiomyosarcoma, and adenocarcinoma

  All have been reported

Imaging

- In infants and children - Chest radiograph diagnostic in 75% cases

  Water-density mass lesions in chest radiographs

- CT - Locating an intrathoracic cyst

  Defining its extent and relation to key structures

  Characterizing the intrinsic density

  Do not enhance following intravenous contrast administration.

- MR imaging - High signal on T2-weighted

  T1-weighted signal is variable

Propensity of bronchogenic cysts to develop complications

Surgical resection usually recommended, regardless of the age at presentation

Esophageal dulication cysts (EDC)
Neurenteric cysts

- EDC – 2 nd most frequent type of enteric duplication.
  - 60% located in the distal third of the esophagus
  - 17% in the middle third
  - 23% are at the cervical level
- More distal the location, the more likely to be asymptomatic.
- Cervical EDCs may present with symptoms of upper airway obstruction.
- Middle third - More likely to cause airway obstruction. leading to cough, wheeze, shortness of breath, and recurrent infection
- Rare locations- Pleural space, tongue, and subcutaneous tissues.
- Vertebral segmentation anomalies, esophageal atresia, and other types of BPFM (mixed lesions)

- EDCs should be surgically
- resected .
- High incidence of complications
- arising in EDCs, including malignant
- degeneration

Tracheal bronchus

- Bronchial anomalies arising in the trachea or main bronchus - upper-lobe territory.
- Usually in the right lateral wall of the trachea < 2 cm above the carina.
- Entire upper lobe or its apical segment.
- (Sandifort ,1785) - Right upper bronchus originating in the trachea.
- Tracheal bronchus – Displaced (More frequent) or supernumerary

Tracheal bronchus

- Tracheal diverticula - Supernumerary bronchi ending blindly.
- Apical accessory lungs or tracheal lobes- Ending in aerated or bronchiectatic lung tissue.
  - Prevalence – Right tracheal bronchus - 0.1%–2%.
    Left tracheal bronchus - 0.3%–1%.
- Symptoms – Asymptomatic usually.
- Consider diagnosis in persistent/recurrent upper-lobe pneumonia or atelectasis or air trapping.

DIAGNOSIS

Most are well seen on CT

Bronchoscopy & bronchography may allow direct visualization.

Tracheal bronchus

- Most patients can be treated conservatively.
- In symptomatic patients surgical excision of the involved segment is necessary.
- Diagnosis should be considered early in the clinical course of intubated patients with right-upper-lobe complications.
- Tumoral lesions developing in a tracheal bronchus are infrequent (5 case reports – Small cell Ca/ Squamous cell Ca)

Tracheomalacia

- Tracheal wall softening

  -Cartilaginous ring abnormality &

  -Hypotonia of the myoelastic elements.

  - Dynamic expiratory tracheal collapse → Airway obstruction.

- Primary tracheomalacia- Congenital immaturity of the tracheal cartilage
- Secondary tracheomalacia – Previously normal cartilage degenerates

Tracheomalacia

- Symptoms - Wheeze, cough, stridor, dyspnea, tachypnea, cyanosis, and recurrent respiratory tract infections.
- Congenital diffuse malacia improves by age 6–12 months.
- Diagnosis - Lateral fluoroscopy and esophagography
- Cine CT - Dynamic cross-sectional evaluation of tracheal compliance and anatomy.

Tracheomalacia

Rare anomaly

No definite prevalence data

Congenital variety may be associated with other anomalies like vascular rings and tracheoesophageal fistula

Tracheal Stenosis

- Focal or diffuse complete tracheal cartilage rings → fixed tracheal narrowing.
- Isolated or with other anomalies.
- ~50% focal, 30% generalized & 20% funnel-shaped.
- 90% present during the 1st year of life, often with biphasic stridor.
- Infants diagnosed early in life - worse prognosis.
- Degree of stenosis more critical than length.

Tracheal Stenosis

- Diagnosis - Frontal and lateral high-kilovoltage filtered radiographs combined with barium esophagography.
- CT only in selected cases.
- Helical CT or EB- CT - useful for evaluating dynamic changes in the airway.
- Pulmonary artery sling – Left PA originates from the Right PA , encircles the right mainstem bronchus and distal trachea, causing compression of each.

Be aware of pulmonary artery sling when examining an infant with apparent segmental distal tracheal stenosis.

Pulmonary underdevelopment

- Classification (Schneider and Schwalbe)
- Group 1 - Bronchus and lung are absent (agenesis)
- Group 2 - Rudimentary bronchus is present and limited to a blind-end pouch without lung tissue (aplasia)
- Group 3 - Bronchial hypoplasia with variable reduction of lung tissue (hypoplasia)
- Agenesis occurs during the embryogenic period (approximately 4 weeks gestation)
- >50% of children with pulmonary agenesis have associated congenital anomalies
- Cardiovascular (more frequent patent ductus arteriosus and patent foramen ovale), gastrointestinal, skeletal, and genitourinary systems.

Lung agenesis

- Patients with right-lung agenesis have a shorter life expentancy than those with left-lung agenesis.
- Chest radiography - Small, completely opaque hemithorax with displacement of the mediastinal structures and diaphragm
- Ocasionally, confined to one lobe, most frequently the left upper lobe.
- CT angiography and MR angiography are currently the imaging modalities of choice in the diagnosis of this entity, with angiography used only in selective cases
- Bilateral pulmonary agenesis is extremely rare and uniformly fatal.

Pulmonary hypoplasia

- Presence of both bronchi and alveoli in an underdeveloped lobe.
- Caused by factors directly or indirectly compromising the thoracic space available for lung growth
- Most common manifestation - Early respiratory distress after birth, cyanosis, tachypnea, hypoxia, hypercapnia, and acidosis.
- Pneumothorax and pulmonary hypertension are common serious complications.
- Chest X Ray - Decreased aeration of the affected hemithorax (more frequent in the right lung) and a small thoracic cage.

Congenital diaphragmatic hernia

- Posterolateral Bochdalek hernia (Left posterolateral commonest – 85 %)
- Anterior Morgagni hernia
- Hiatus hernia.

- Left-sided hernias - Herniation of both the small and large bowel and intraabdominal solid organs into the thoracic cavity.
- Right-sided hernias (13%) - Only the liver and a portion of the large bowel tend to herniate.

- Commonly associated with pulmonary hypoplasia.

- Bilateral hernias are uncommon and usually fatal.

- **Symptomatic CDH at birth is considered a surgical emergency**

- **Correction of defect is carried out as early as possible.**

Tracheobronchomegaly (Mounier-Kuhn syndrome)

- Rare disorder , described in 1932.
- Marked dilatation of the trachea and main bronchi, sometimes with tracheal diverticulosis, bronchiectasis, and recurrent lower respiratory tract infection.
- Etiology is uncertain.
- Predominantly occurs in men in their third and fourth decades of life.
- Treatment –
  - Physiotherapy to assist in clearing secretions
  - Appropriate antibiotics during infectious exacerbations

RADIOLOGIC CRITERIA	TRACHEA	RIGHT MAIN BRONCHUS	LEFT MAIN BRONCHUS
STANDARD CXR/ BRONCHOGRAPHY	3.0 cms	2.4 cms	2.3 cms
CT	3.0 cms	2.0 cms	1.8 cms

Esophageal atresia & Tracheoesophageal fistula

Suspected - **Polyhydramnios, inability to swallow saliva or milk, aspiration during early feedings, or failure to successfully pass a catheter into the stomach.**

VACTERL anomalies

Accessory cardiac bronchus

- Origin - Inferior medial wall of the right main or intermediate bronchus.
- May end blindly or be associated with small amounts of abnormal pulmonary parenchyma.
- May serve as a potential reservoir of infectious organisms.
- Symptoms – Asymptomatic

  Cough , hemoptysis , Recurrent pneumonia

Congenital pulmonary venolobar syndrome / Hypogenetic lung syndrome / Scimitar syndrome

Almost always right sided lung.

Scimitar syndrome

- Suprahepatic IVC, hepatic veins, portal veins, azygous vein, coronary sinus, and right atrium (May also receive)
- Cardiovascular defects - Sinus venosus or secundum ASD.
- Others- Vertebral anomalies, abnormal lung lobation, tracheal stenosis and diverticula, & gastrointestinal tract anomalies
- Infants - Present early when they have coexistent congenital heart disease or systemic arterial supply to the right lung. Higher morbidity and mortality
- More likely to develop pulmonary hypertension

Diagnosis & Treatment

- Chest radiograph
- CT & MR imaging - Direct visualization of the anomalous vein.
- Conventional angiographic studies - To delineate the arterial and venous anatomy before surgical repair.

Pulmonary sequestration

- Segment of lung parenchyma ,receives its blood supply from the systemic circulation & does not communicate with the tracheobronchial tree.
- ELPS - Entirely separate segment of lung tissue invested in its own pleural layers (25% of PS).
- Typically found in the costophrenic sulcus on the left side. (90 % left sided).
- May also be located in the mediastinum, pericardium, and within or below the diaphragm.

Pulmonary sequestration

- **ILPS - Shares the visceral pleural covering of the normal adjacent lung tissue(75% of PS).**
- **Usually located in the posterobasal portion of the lower lobes.**

VASCULAR SUPPLY	Arterial Supply	Venous drainage
ELPS	Abdominal/ thoracic aorta (85-90%) (10%–15%)Celiac Axis/subclavian arteries/ intercostal arteries	80% - Systemic Circulation (Azygous System/IVC) 20 % - Pulmonary Circulation
ILPS	Thoracic/ upper abdominal aorta	Pulmonary venous system.

Pulmonary sequestration

- ELPS – Usually diagnosed early (4:1 Male predominance).
- Associated with other congenital malformations in over 50%.
- Cong. Diaph. Hernia, Congenital HD & CCAM type II (hybrid lesions).
- Infants - Respiratory distress/feeding difficulties/incidental prenatal or postnatal imaging finding.
- During pregnancy - Polyhydramnios /nonimmune hydrops. High-output cardiac failure may occur(L→R Shunt).

Pulmonary sequestration

- ILPS – Usually diagnosed in later childhood or even in adults.
- Incidental or Recurrent pneumonia/hemoptysis.
- Prenatal diagnosis increasingly being recognized.
- May be associated with neonatal cardiac failure.
- Associated congenital anomalies in (6% to 12% of cases).
- Main diagnostic feature in either type of PS is the feeding systemic arterial vessels.
- IDENTIFIED BY IMAGING

Imaging in PS

- US with color doppler is particularly useful in the neonate and young infant.
- CT – Useful diagnostic modality.
- Can identify structure and also the arterial and venous supply.
- MR imaging - Identify the feeding arterial vessels & delineate the character of the mass.
- Usually of high signal intensity on both T1- and T2-weighted images

Pulmonary sequestration

- Recently ,antenatal MR imaging being used to confirm the diagnosis.
- Most PS is managed surgically.
- ELPS are resected.
- ILPS generally require a segmentectomy or lobectomy, particularly with history of recurrent infection.
- Embolization of the feeding arterial vessels has recently been described in the treatment.

Hybrid lesions (CCAM with PS)

- ELPS reported to exist with type II CCAM in up to 50% of cases.
- Importance - Search for an aberrant systemic arterial vessel when imaging suspected cases of CCAM.
- Definite diagnosis by pathology.
- Manifestations depend on size and the associated complications.

Pulmonary arteriovenous malformations (AVM)

- Persistence of primitive arteriovenous communications
- Single feeding artery and draining vein
- 50 % with multiple AVMs have HHT
- Most patients develop symptoms by the third or fourth decade
- Symptoms - dyspnea on exertion and central or peripheral cyanosis.
- Clubbing , polycythemia and pulmonary hypertension can develop , Paradoxical CNS embolism may occur
- Platypnea , Orthodeoxia can be observed

Pulmonary arteriovenous malformations (AVM)

- Screening asymptomatic relatives (HHT) - can avert the serious CNS complications.
- Contrast-enhanced echocardiography is the most commonly used imaging technique.
- Also helpful to monitor patients following therapy.
- Transcatheter balloon or coil closure of pulmonary AVMs is now the preferred method of treatment

Interruption (Absence) of a Main Pulmonary Artery

- Unilateral congenital absence - Rare malformation.
- Affected lung is decreased in size, as is the hilum.
- The lung is hyperlucent.
- Interruption of the left pulmonary artery is much less common and may be associated with congenital cardiovascular anomalies
- Recurrent pneumonia, limited exercise tolerance, and hemoptysis may occur
- Usually only the proximal section of the vessel that is absent.

Echocardiography - Recommended to assess the intracardiac anatomy and to look for associated defects

CT or MR angiography - Accurately depicts the hilar anatomy, and
delineates the collateral vascular network

Anatomical variants (Normal parenchyma)

- **Pulmonary isomerism**

  Anomaly of the number of lung lobes.

  Common variety - Right lung has 2 lobes, whereas the left has 3.

  May be associated with situs inversus, asplenia, polysplenia, and/or anomalous pulmonary drainage.

- **Azygous lobe**

  Malformation of the right upper lobe caused by an aberrant azygous vein suspended by a pleural mesentery. Radiographic curiosity without clinical significance

  Occurs in 0.5% of the general population.

- **Superior segment of lower lobe delineated by separate fissure**

- **Medial accessory left lower lobe**

Conclusion

- Most congenital and developmental anomalies have certain characteristic imaging features.
- These features may aid in the differentiation from more sinister abnormalities.