Congenital Airway Anomalies

Lakshmi Uppaluri, MD
Faculty Pediatric Pulmonary medicine
UMDNJ/RWJMS
Choanal Atresia

• Most common congenital anomaly of nose with incidence of 1:7000 live births

• Etiology:
  ❖ Failed oronasal membrane rupture
  ❖ Abnormal migration of neural crest cells into nasal vault

• Clinical manifestations vary depending on involvement (unilateral vs bilateral)
Choanal Atresia-Clinical manifestations

• **Bilateral:**
  - Respiratory distress at birth
  - Presents with cyanosis that worsens during feeds and improves with crying
  - Could be suspected by inability to pass 6 French catheter through the nose

• **Unilateral:**
  - Presents later in life
  - Persistent nasal discharge and recurrent URI
Choanal Atresia-Diagnosis

- CT scan
  - Confirms the diagnosis
  - Shows narrowing of posterior nasal cavity at level of pterygoid plate
- Endoscopy:
  - Can be performed by flexible or rigid
  - Shows obliteration of “plate” or aperture
Choanal Atresia

• Associated anomalies:
  - Treacher Collins
  - CHARGE
  - Kallman
  - VATER
  - Craniofacial anomalies

• Differential diagnosis:
  - Nasal septum deformity
  - Nasal Foreign body
  - Choanal polyp
Choanal Atresia-Management

• Bilateral, symptomatic in delivery room. Place oral airway and intubate

• Support growth with gavage feeds

• Repair:
  ❖ Transnasal puncture and stenting
  ❖ Endoscopic resection of posterior nasal septum
  ❖ High risk of recurrent stenosis even after successful surgery
Normal larynx
Laryngomalacia

• Most common cause of congenital stridor in children
• Male : female 2:1
• Inspiratory stridor worse when the child is crying, supine or with upper respiratory tract infection
• Cause: Intrinsic defect or delayed maturation of supporting structures of larynx
  ❖ Airway obstructs with inspiratory prolapse of flaccid epiglottis, arytenoids and aryepiglottic folds
Endoscopic view

<table>
<thead>
<tr>
<th>Inspiration</th>
<th>Expiration</th>
</tr>
</thead>
</table>

- Inspiration image with arrows indicating specific features.
- Expiration image with arrows indicating specific features.
Laryngomalacia-Diagnosis

- Fiberoptic laryngoscopy while awake, with omega shaped epiglottis and taught aryepiglottic folds
- Direct laryngoscopy with bronchoscopy
Laryngomalacia-Management:

- Worsens in the first 8 months, plateaus at 9-12 months and resolves spontaneously by 18-24 months of age in most patients
- Symptomatic relief of stridor can be sometimes achieving by lying the infant on side
- Severe cases require surgical treatment with supraglottoplasty where excessive arytenoid folds, lateral epiglottis and supra-arytenoid folds are resected
- Rarely tracheotomy or Bipap may be required in very severe cases
Tracheomalacia

• Abnormal tracheal collapse secondary to inadequate cartilaginous and myoelastic components supporting the trachea
• Tracheal narrowing occurs during expiration and causes stridor

• Clinical manifestations:
  ❖ Intra thoracic: barky croup like cough
  ❖ Extra thoracic: inspiratory stridor
  ❖ Both are associated with respiratory distress
Tracheomalacia-Diagnosis

- Barium swallow: extrinsic intrathoracic compression of trachea secondary to vascular ring
- Bronchoscopy during spontaneous respiration
- CT/ MRI can further define the location
Tracheomalacia-Management

• Spontaneous resolution at 6-12 months

• With severe airway compromise, recurrent infection, respiratory failure
  ❖ Tracheotomy or PPV
  ❖ Tracheal reconstruction
  ❖ Tracheopexy or Aortopexy
  ❖ Tracheal stents, rarely due to high failure rate
Vignette

• A 3kg female infant is born to a 23 yr old G1P0 at 36 weeks GA. Her APGARS are 5 and 5. She was noted to be apneic in the delivery room and cyanotic. Her PE was significant for decreased breath sounds in chest and a scaphoid abdomen.
Congenital diaphragmatic hernia

• Definition: It is a defect of the diaphragm which allows herniation of abdominal contents into the thorax
• Incidence: 1 in 2000 to 4000 live births
• Etiology:
  ❖ It is believed to result from incomplete fusion of the pleuroperitoneal membrane and passage of abdominal contents into the chest
  ❖ The small bowel, stomach, spleen, and colon are the most frequently herniated organs. Also seen infrequently are pancreas, liver, adrenal glands, and kidneys
Congenital diaphragmatic hernia

- Clinical features:
  - At birth abdomen is scaphoid, chest is funnel shaped
  - Trachea and mediastinum are pushed to contralateral side
  - Clinical presentation can be of frank respiratory failure to apnea and episodes of choking
  - 90% are left sided
  - 50% are associated with other congenital malformations (esp. heart, kidneys, gastrointestinal tract, abdominal wall and central nervous system)
  - Associated with pulmonary hypoplasia and pulmonary hypertension
  - Can be associated with chromosomal disorders
Congenital diaphragmatic hernia

- Diagnosis:
- Prenatal ultrasound:
  - Shows polyhydramnios as well as fluid filled bowel with peristalsis at the level of heart
  - Right sided hernias are usually missed due to echogenicity of liver and lung
  - Differentials: Congenital cystic adenomatoid malformation, bronchogenic cysts, mediastinal cystic teratoma and neurogenic tumors
Chest X-ray

Postnatal chest radiograph in an infant who has a large left CDH


Copyright ©1999 American Academy of Pediatrics
Congenital diaphragmatic hernia

- Management:
- Prenatal: FETO
- Respiratory stabilization in the delivery room
- AVOID BAGGGING THE INFANT IN DELIVERY ROOM
- May need Positive pressure ventilation, inhaled nitric oxide, surfactant therapy, ECMO
- Surgical correction after respiratory stabilization
Congenital diaphragmatic hernia

- Prognosis:
  - Has high mortality rate
  - Is poor if diagnosed in utero
  - Inability to tolerate enteral nutrition, GERD
  - Depends also on pulmonary hypoplasia, pulmonary hypertension
  - Can develop scoliosis eventually
Vignette

- A 3kg male child born to G3 P2L2 @ 40 wks GA. In the delivery room he had increased oral secretions which were removed with bulb suctioning. Prenatal history is significant for polyhydramnios. Was admitted to the nursery with vital signs of Temp: 37C, RR: 40, O2sat: 98% on RA. He was attempted to be fed with significant non-bilious emesis.
Tracheoesophageal Fistula

• Incidence is 1 in 3000 to 5000 live births
• Embryology: It is due to incomplete mesoderm separation of the primitive foregut, resulting in a fistula between esophagus and trachea
• Aspiration is the primary cause of lung damage
• Very good prognosis
Tracheo-esophageal Fistula

- Types:
  - Esophageal atresia with associated distal tracheoesophageal fistula - 80-90%
  - Esophageal atresia without a tracheoesophageal fistula - 10%
  - H-type tracheoesophageal fistula - 3%
TEF-EA

Blind pouch
Distal TEF
Tracheo-esophageal fistula with Esophageal atresia

- Is the most common of TE fistulas constituting 85-90%
- Incidence is 1 in 4000 live births
- Frequent component of VATER
- Embryology: Interruption of events responsible for elongation of esophageal and tracheal tubes during 4th week of development
- Prenatal Ultrasound is characterized by polyhydramnios
Tracheo-esophageal fistula with Esophageal atresia

• Presentation:
  ❖ Excessive amount of saliva
  ❖ Intolerance of oral feeds
  ❖ Non- bilious emesis
  ❖ Gastric distension, not scaphoid abdomen
  ❖ Tachypnea
  ❖ Cyanosis
Tracheo-esophageal fistula with Esophageal Atresia

• Diagnosis:
  ❖ Difficulty to pass a NG tube which ends in the blind pouch
  ❖ KUB/ baby gram shows dilated esophageal pouch or a catheter ending in blind pouch and air through out the GI tract

• Management:
  ❖ Sump catheter into upper pouch
  ❖ Elevate head end to 45 degrees
  ❖ IV fluids and antibiotics
TEF-EA CXR
Tracheo-esophageal fistula with Esophageal atresia

• Management:
  - Surgical repair of fistula can be undertaken in infant as little as 1200 g
  - Determined by associated anomalies and clinical status
• Esophageal Atresia:
  - Double lumen orogastric tube to suction
  - Gastrostomy tube
  - Progressive esophageal dilatation
  - Esophageal substitution procedures like colonic interposition, ileal interposition or reverse gastric tube
Esophageal Atresia without a TE fistula

- Presents with increased oral secretions and drooling
- FLAT AND GASLESS ABDOMEN
- Inability to tolerate enteral feeding
- Esophageal pouches have large gap thus making anastomosis difficult
- Diagnosis: By KUB or passing a catheter that ends in the blind pouch
- Management:
  - Orogastric tube to suction
  - G-tube insertion
  - Progressive dilation of pouches and subsequent insertion of gastric tube, colon or piece of small intestine
H-Type Tracheoesophageal Fistula

• Present with increased choking in the newborn period or chronic cough and recurrent pneumonia in a older child

• Diagnosis:
  ❖ Could be subtle so difficult to diagnose
  ❖ Upper GI with esophagogram
  ❖ Flexible bronchoscopy with esophagoscopy

• Management: Surgical division by the cervical approach
Tracheo-esophageal fistula with Esophageal atresia

- Prognosis:
  - Excellent and depends upon associated anomalies
  - Narrowing or stricture at anastomotic site is very common, requiring progressive dilatation
  - Esophageal dysmotility at the stricture site, esp. in colonic interposition or illeal interposition
Congenital Cystic adenomatoid malformation

- Most common pulmonary anomaly
- It is dysplastic development of terminal bronchiolar structures resulting in hamartomous lung
- Embryologic insult is around 7th week of gestation
- Communicates with tracheobronchial tree
- Symptomatic in new born period or early infancy
- Divided into 4 types depending upon its constituents
Types of CCAM

• Type 1: Cysts are multiloculated and larger than 2 cm. Rare transformation to bronchoalveolar carcinoma

• Type 2: Multiple small cysts. Associated with renal agenesis, CV anomalies, diaphragmatic hernia and Syringomelia

• Type 3: Uncommon. Involve whole lobe. Macroscopically-solid appearing and microscopically-resemble late fetal lung

• Type 4: Very rare. Multiloculated thin walled cysts. Can have malignant transformation to pleuropulmonary blastoma
Congenital Cystic adenomatoid malformation

- Diagnosis:
  - Prenatal Ultrasound
  - Chest X-ray
  - CT scan

- Management: By resection

- Complications:
  - Respiratory compromise
  - Recurrent infection
  - Could be premalignant
Congenital Cystic adenomatoid malformation
Vignette

A 10 yr old male presents to ER for sore throat and cough, low grade fever. Physical Exam in ER, Temp: 98.6, RR: 28/ minute, O2sat: 98% on RA. General: Alert, no tachypnea, HEENT: Within normal limits, Lungs: Decreased breath sounds in right anterior lung fields, left lung is clear to auscultation, no wheezing, no crackles. Chest: PMI is displaced to the right chest, RRR, S1S2: normal, no murmur, Abdomen: Soft, non tender, non distended, no hepatosplenomegaly, bowel sounds: normal, Extremities: No clubbing, no cyanosis. Work up included: rapid streptococcal antigen test: negative. Chest X-ray which lead to an incidental diagnosis
Congenital lobar emphysema

- It is progressive over inflation / over distension of the lung

- Etiology:
  - Bronchial atresia/stenosis
  - Absent or dysplastic bronchial cartilage (bronchomalacia)
  - External compression by vessels

- Clinical findings:
  - Before 6 months: mild tachypnea, wheeze to severe dyspnea and cyanosis
  - Poor feeding and failure to thrive
  - Older child: Incidental finding on chest X-ray or recurrent pneumonia
Congenital lobar emphysema

- Pathology: Decreased alveoli and bronchial wall cartilage
- Affected lobes remain the same after first year of age and therefore become smaller in relation to normal lobes
- CXR findings:
  - Hyperlucency with scant pulmonary markings
  - Hyper inflated lobe with collapse of the contralateral lobe
  - Mediastinal shift with flattened diaphragm
Congenital lobar emphysema

• Differential diagnosis:
  - Foreign body
  - Bronchogenic cyst
  - CCAM

• Diagnosis:
  - Chest X-ray
  - Flexible bronchoscopy
  - CT scan is diagnostic
Congenital lobar emphysema

• Complications:
  ❖ Pneumothorax with positive pressure ventilation
  ❖ Compromise respiratory reserve by compressing uninvolved lobes

• Therapy:
  ❖ Surgical excision, indications: respiratory reserve is compromised or in the setting of recurrent infections
  ❖ Rule out congenital heart disease before surgery
Vignette

• 13 month old male presents to the ED with cough, high fever and increased work of breathing. Vitals in the ER: Temp: 39 C, RR: 32, O2sat: 96% RA. PE: General: Sub coastal retractions, HEENT: Rhinnorhea, Lungs: decreased breath sounds at left base, no crackles, no wheezing and good air entry, rest of physical exam is unremarkable. Chest X-ray showed left lower lobe infiltrate

• Past Medical history: Born at 39 wks GA by NSVD. Was found to be tachypneic in nursery. CXR in nursery, showed left lower lobe infiltrate suspected to be congenital pneumonia and was treated with IV antibiotics. Was seen by pediatrician 5-6 times with recurrent upper respiratory tract infections and persistent tachypnea
X-ray in nursery   X-ray on admission
Pulmonary Sequestration

• Pulmonary tissue that is isolated from normal functioning lung and is nourished by systemic arteries. It is dysplastic and non-functioning. Has no connection to tracheobronchial tree

• Embryology: Interruption of orderly lung development and persistence of perfusion of sequestered lung tissue from systemic circulation

• Rarest of all congenital lung malformations
CXR of Pulmonary Sequestration
Pulmonary Sequestration

• Divided into intra lobar and extra lobar types depending upon blood supply and if it is contiguous with the lung
• Intralobar sequestration:
  ❖ 3-6 times more common
  ❖ Pleural covering is contiguous with the lung
  ❖ Usually left sided
  ❖ Arterial supply is by systemic artery, venous drainage is by pulmonary veins
  ❖ Rarely associated with anomalies
  ❖ Presents in childhood with recurrent pneumonia
Pulmonary Sequestration

- Extra lobar sequestration:
  - Less common
  - Has its own visceral pleura and separate from rest of lung
  - Usually left sided
  - Associated with anomalies
  - Arterial supply is systemic, venous drainage is to systemic veins
  - Presents in infancy with respiratory distress
  - Can be diagnosed by prenatal ultrasound
Pulmonary Sequestration

• Diagnosis:
  ❖ Chest X-ray
  ❖ CT scan with contrast
  ❖ MRI/ MRA

• Management:
  ❖ IV antibiotics
  ❖ Resection when primary infection is cleared
Arteriogram of sequestration
## Congenital anomalies by age of Presentation

<table>
<thead>
<tr>
<th>At Birth</th>
<th>Children</th>
</tr>
</thead>
<tbody>
<tr>
<td>Congenital diaphragmatic hernia</td>
<td>H-type TE fistula</td>
</tr>
<tr>
<td>Tracheo esophageal fistula with esophageal Atresia</td>
<td>Laryngomalacia</td>
</tr>
<tr>
<td>Choanal Atresia- bilateral</td>
<td>Tracheomalacia</td>
</tr>
<tr>
<td>Extra lobar sequestration</td>
<td>Intralobar sequestration</td>
</tr>
<tr>
<td>Congenital cystic adenomatoid malformation</td>
<td>Congenital lobar emphysema</td>
</tr>
<tr>
<td>Congenital lobar Emphysema</td>
<td></td>
</tr>
</tbody>
</table>
# Congenital anomalies by presentation

<table>
<thead>
<tr>
<th>Respiratory distress / noisy breathing</th>
<th>Recurrent Pneumonia</th>
</tr>
</thead>
<tbody>
<tr>
<td>Congenital diaphragmatic hernia</td>
<td>H-type TE fistula</td>
</tr>
<tr>
<td>Choanal Atresia- bilateral</td>
<td>Congenital lobar emphysema</td>
</tr>
<tr>
<td>Laryngomalacia</td>
<td>Congenital cystic adenomatoid malformation</td>
</tr>
<tr>
<td>Tracheoesophageal fistula and esophageal Atresia</td>
<td>Intralobar sequestration</td>
</tr>
<tr>
<td>Congenital lobar Emphysema</td>
<td>Congenital lobar Emphysema</td>
</tr>
<tr>
<td>Tracheomalacia</td>
<td></td>
</tr>
<tr>
<td>Congenital cystic adenomatoid malformation</td>
<td></td>
</tr>
</tbody>
</table>