Agenesis of the Lung

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September 18, 2008
Outline

- Normal Embryonic Development
- Lung Agenesis
- The Case
  - Patient
  - Examination
  - Diagnosis
  - Prognosis

Right Pulmonary Agenesis
Day 22: Ventral lung buds derived from endodermal foregut begin to form.

As lung bud grows, remains ensheathed in covering of splanchnopleuric mesoderm which will give rise to connective tissue, cartilage, and muscle within bronchi.

Day 26: Bifurcation of the lung buds into primary bronchi.
4 Stages of Bronchial Development

1) Glandular: first 4 months, rest of bronchial tree formed and respiratory-like movements ensue

2) Canalicular: 4th to 6th month, vascularization begins

3) Alveolar: 6th month to term, branching continues until 24 generations of alveoli formed, as alveolar sacs form the tissue separating blood of pulmonary circuit from alveolar air becomes thin enough to allow exchange of $O_2$ and $CO_2$

4) Birth: this stage can occupy first 8 years of life, terminal bronchioles continue to divide, expansion of alveoli, and surfactant is produced (alveolar surface tension)
The lungs contain millions of tiny alveoli.

Oxygen (O₂) from air breathed in, goes into the red blood cells via alveoli. Carbon dioxide (CO₂) goes from the red blood cells into alveoli and breathed out.

Lung showing alveoli

Terminal Bronchiole: Continuous epithelium which gives rise to two...
Respiratory Bronchiole: Discontinuous; interrupted by many alveoli
Alveolar Duct: More interruptions than walls
The lung is a composite of endodermal and mesodermal tissues.

- Endodermal gives rise to mucosal lining of bronchi and epithelial cells of alveoli.

- Mesodermal gives rise to remaining components of lung (muscle and cartilage supporting bronchi and visceral pleura covering lung).
Also known as pulmonary agenesis, is classified into 3 different diagnoses:

1) Bilateral Agenesis: death is imminent

2) Unilateral Agenesis
   a) no bronchi
   b) rudimentary bronchus present
   c) main bronchus poorly developed

3) Lobar Agenesis
Lung Agenesis cont.

- Rare condition that occurs in approximately 1 out of every 20,000 births

- In a study of 200 unilateral pulmonary agenesis cases:
  - 70% was of the left side
  - more males than females
  - 50% had related malformations
Related Malformations

- Esophageal stenosis (narrowing)
- Stenosis of trachea
- Absence of pleura
- Supernumerary bronchi of normal lung
- Tracheoesophageal atresia
- Congenital diaphragmatic hernia

Esophageal stenosis
Causes of Lung Agenesis

1) Genetics
   a) Autosomal Recessive

2) Poor fetal nutrition

3) Mechanics
   a) FGF10 and SHH are growth factors, genetic deletion of FGF10 or SHH leads to inhibition of development
1) Cyanosis (blue coloration of skin due to lack of oxygen)

2) Dyspnea (shortness of breath)

3) Feeding difficulties

4) Wheezing
The Patient

Sex: male

Age: 8 months

Symptoms: wheezing, dyspnea, but no cyanosis

Prior to hospital admission treated for pneumonia
The Examination

◆ Chest was asymmetric (left side much smaller)
◆ Dullness on percussion
◆ No breathing sounds on left side
◆ X-ray and flouroscopy (imaging technique to obtain real-time moving images of internal structures) showed left lung opaque, right lung larger than usual
◆ Mediastinum shifted left
◆ No left bronchus on a bronchogram
◆ Bronchoscope showed an obstruction of left primary bronchus just below carina
Diagnosis

- Agenesis of the left lung with no other malformations
Since no other malformations were found, the child can live normally.

Therefore, he was discharged under pediatric care for regular checkups for respiratory infections.

Next 4 years, shortness of breath subsided and development was normal.
Questions?

Thank you for listening!