Cardiopulmonary Syndromes: Conditions With Concomitant Cardiac and Pulmonary Abnormalities

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Cardiopulmonary Syndromes

- Heterotaxia syndrome
- Primary ciliary diskinesia
- Ring-sling complex
- TAPVR
- PAPVR (Scimitar syndrome (Hypogenetic lung))
- Interrupted pulmonary artery
- Pulmonary sequestration
- Congenital lobar emphysema
- Tetralogy of Fallot
- Pentalogy of Cantrell
- Pulmonary agenesis / hypoplasia
- VACTERL association
- Down syndrome (Pulmonary hypoplasia and lung cysts) (NEJM Cooney 1982)
PAPVR / Scimitar Syndrome

Four features

• Hypoplasia of right lung with dextroposition of the heart
• Hypoplastic right pulmonary artery
• Anomalous arterial supply to right lower lobe from aorta
• Anomalous pulmonary venous connection to inferior vena cava
Scimitar Syndrome

• >75% have congenital heart disease
  - ASD (secundum type) in 65%
  - VSD in 16%
  - PDA in 9%

• 28% complex congenital heart disease
  - TOF
  - Hypoplastic left heart syndrome
  - TAPVR

PAPVR associated with diaphragmatic hernia. Contrast enhanced CT shows the anomalous vein (small arrow) and the liver herniated into the right hemithorax (large arrow).
Scimitar syndrome associated with pulmonary sequestration. Chest x-ray shows a round mass in the right upper quadrant (black arrows). Venous phase catheter angiography shows the anomalous vein (white arrow).
PAPVR + Sinus Venosus ASD + PLSVC
PAPVR Extracardiac Associated Anomalies

- 25% are associated with other congenital abnormalities:
  - Aortic coarctation (20%)  
  - ASD  
  - VSD  
  - Tetralogy of Fallot  
  - Diaphragmatic hernia  
  - Diaphragmatic duplication
PAPVR associated with diaphragmatic hernia. Contrast enhanced CT shows the anomalous vein (small arrow) and the liver herniated into the right hemithorax (large arrow).
Primary Ciliary Diskinesia

- Recessive genetic disorder characterized by sinopulmonary disease secondary to abnormal ciliary structure and function
- Prevalence of congenital heart disease in PCD with heterotaxy is 200-fold higher
- Situs inversus totalis occur in 48% (Kartagener’s syndrome)
- Situs solitus in 46%
- Situs ambiguus in 6% (Heterotaxy)
  - most have CV abnormalities

Kennedy MP et al. Circulation 2007;115: 2814
Kartagener’s Syndrome

Primary Ciliary Dyskinesia (PCD) was previously referred to as immotile cilia syndrome until it was discovered that most of the cilia moved but are dysfunctional.

PCD is also referred to as Kartagener’s Syndrome if situs inversus is present.

About half of patients with respiratory ciliary disease have situs inversus.

PCD follows an autosomal recessive mode of inheritance.
Heterotaxia Syndrome
(situs ambiguus)

Right isomerism
- Ivemark syndrome
- Asplenia syndrome
- Both lungs have 3 lobes
- Eparterial bronchi
- CHD in 100%
- M > F

Left isomerism
- Polysplenia syndrome
- Both lungs have 2 lobes
- Hyparterial bronchi
- CHD in 70%
- Azygos continuation 80%
- F > M
Bronchial Isomerism

Right isomerism

Left isomerism
Right bronchial isomerism

Three lobes in the bilateral lungs
Congenital interruption of the PA

- Congenital absence of the PA
- Term interruption is preferred
- Only the mediastinal segment of the right or left PA is absent
- Interrupted artery is in the opposite side of the aortic arch
- May be seen associated with CHD (ASD, VSD, TA and TOF)

Congenital interruption of the PA

- Right pulmonary artery more commonly affected than the left
- May be associated with hypogenetic lung
- Systemic collateral circulation from bronchial arteries
- Women are affected more than men
- Cystic changes in the ipsilateral lung demonstrated in HRCT

Fem 22 y/o.
Multiple small cystic dilatations of the alveoli aligned along the subpleural surface or diffuse in advanced cases.

Pathophysiology is not clear.

Frequently associated with congenital heart disease.

Might contribute to the development of pulmonary hypertension.

2 y/o Male
Down syndrome
Aberrant Left Pulmonary Artery

• Pulmonary sling
• Failure of development / obliteration of left 6th aortic arch.
• Development of a collateral branch of right pulmonary artery to supply the left lung.
• LPA passes above right mainstem bronchus and between trachea and esophagus.
Aberrant Left Pulmonary Artery

• Associated with
• Napkin-ring trachea (absent pars membranacea) 50%
• “carrot-shaped” trachea (long segment tracheal narrowing)
• Obstructive emphysema
• PDA (most common)
• ASD, persistent LSVC
Branch PA Stenosis

- Commonly associated with CHD - TOF, TGA, ASD, VSD
- Also associate with Noonan syndrome and Williams syndrome
- Maternal rubella syndrome
- Ehler-Danlos syndrome
- Cutis laxa syndrome
- Alagille’s syndrome
Branch PA Stenosis (TOF)