Pulmonary Lymphangioleiomyomatosis (LAM)

Key Points:
- LAM is a rare lung disease characterized by atypical smooth muscle proliferation and cystic airspace dilatation affecting women of childbearing age.
- Will present with dyspnea or cough but can have pneumothorax, chylothorax, renal angiomyolipomas, or meningiomas
- HRCT shows diffuse, homogeneous thin-walled cysts
- Treatment is lung transplant or anti-estrogen therapy can be tried.

Definition
- A rare lung disease affecting young women of childbearing age

Epidemiology
- Almost exclusively in white, premenopausal women (70% between the ages of 20 and 40)
- Seen in 5% of cases of documented tuberous sclerosis

Pathology/Pathophysiology
- Proliferation of atypical smooth muscle cells around bronchovascular structures and interstitium
- Also, diffuse cystic dilatation of the terminal airspaces
- Can have tortuous and dilated lymphatics and veins with enlarged lymph nodes
- Pathogenesis probably a mutation in the TSC2 gene on chromosome 16 (tuberous sclerosis)

Clinical Features
- Nearly all patients have dyspnea at presentation; also cough and chest pain
- There are many intra- and extrathoracic complications
  - Spontaneous pneumothorax in 50% of cases
  - Chylothorax, chyloperitoneum, chyluria, or chylopericardium can occur
  - Renal angiomyolipomas in 50% - usually asymptomatic
  - Some increased risk of meningioma

Diagnosis
- Pulmonary function tests will show “obstructive” or mixed pattern and low DLCO
- Chest xray can show interstitial opacities or honeycombing
- HRCT will show diffuse, homogenous, small thin-walled cysts
  - Ddx of cystic dz includes: emphysema (+/- α1-anti-trypsin), histiocytosis X, cystic sarcoidosis, chronic hypersensitivity pneumonitis, emphysema from IVDU

Prognosis
- Median survival 8 – 10 years from diagnosis
- Pregnancy and supplemental estrogen accelerate the disease

Treatment
- Hormone regulation is the key
  - Oophorectomy, progestin tx, tamoxifen, and LHRH analogs have been used
  - No randomized trials of therapy
- Can treat with lung transplant

References: