BSTI case presentation

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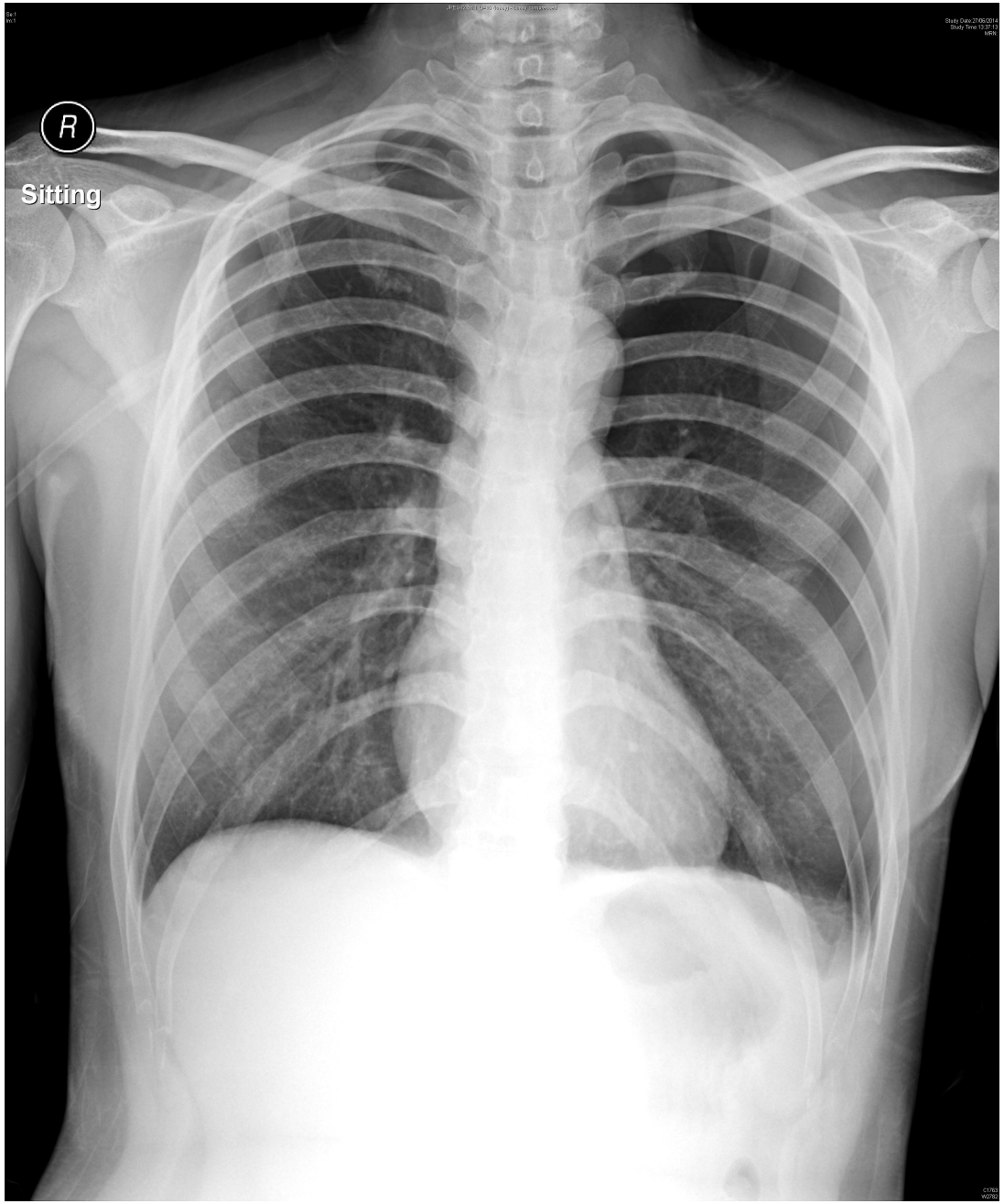
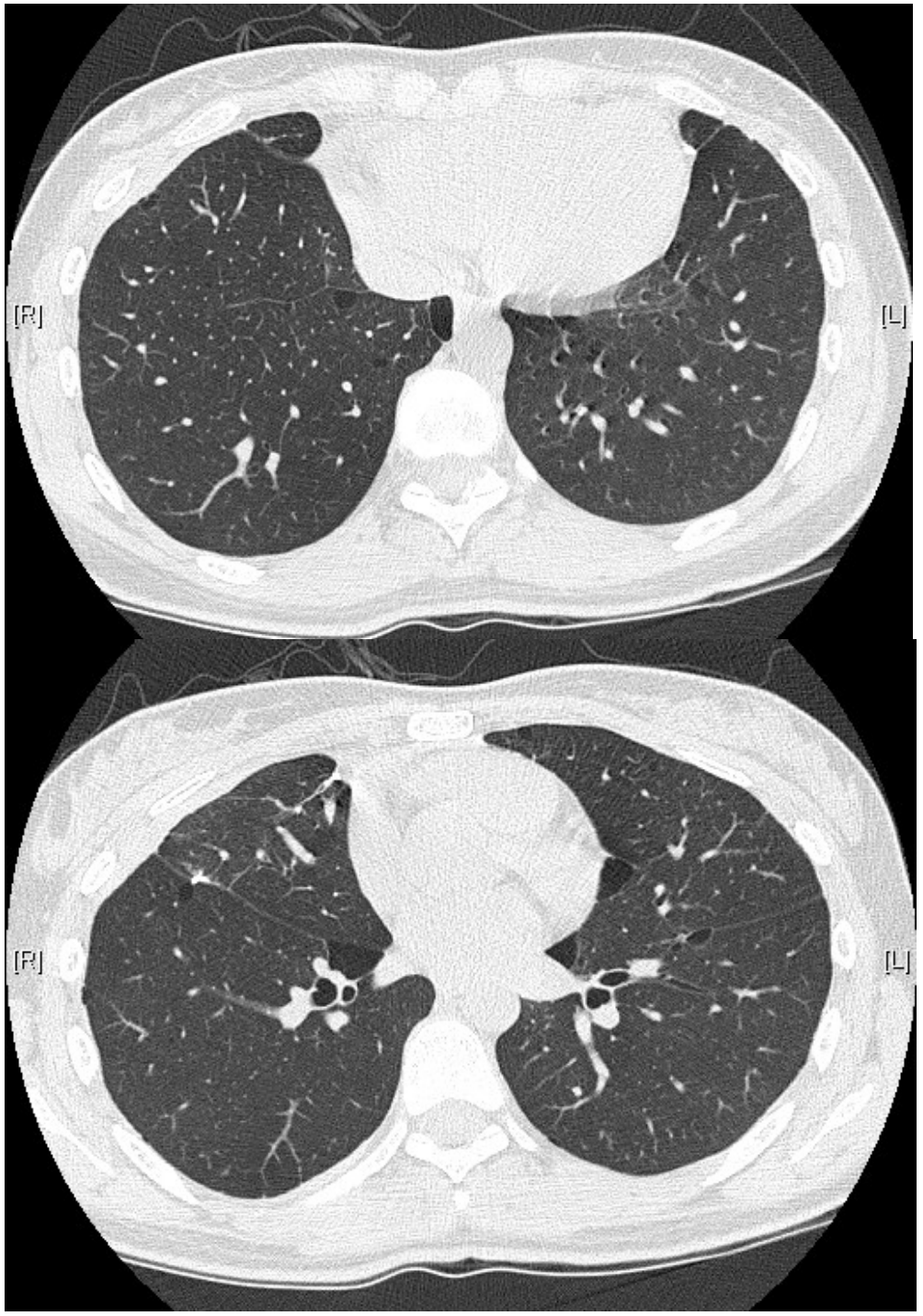
Clinical History

- A 34-year-old Chinese female with good past health, presented spontaneous bilateral pneumothoraces.
- The episode of pneumothorax is not related to menstruation nor oral contraceptive pills.



Clinical History

- HRCT showed scattered scant blebs <2cm over bilateral pulmonary sub-pleural regions in upper and lower lobes.



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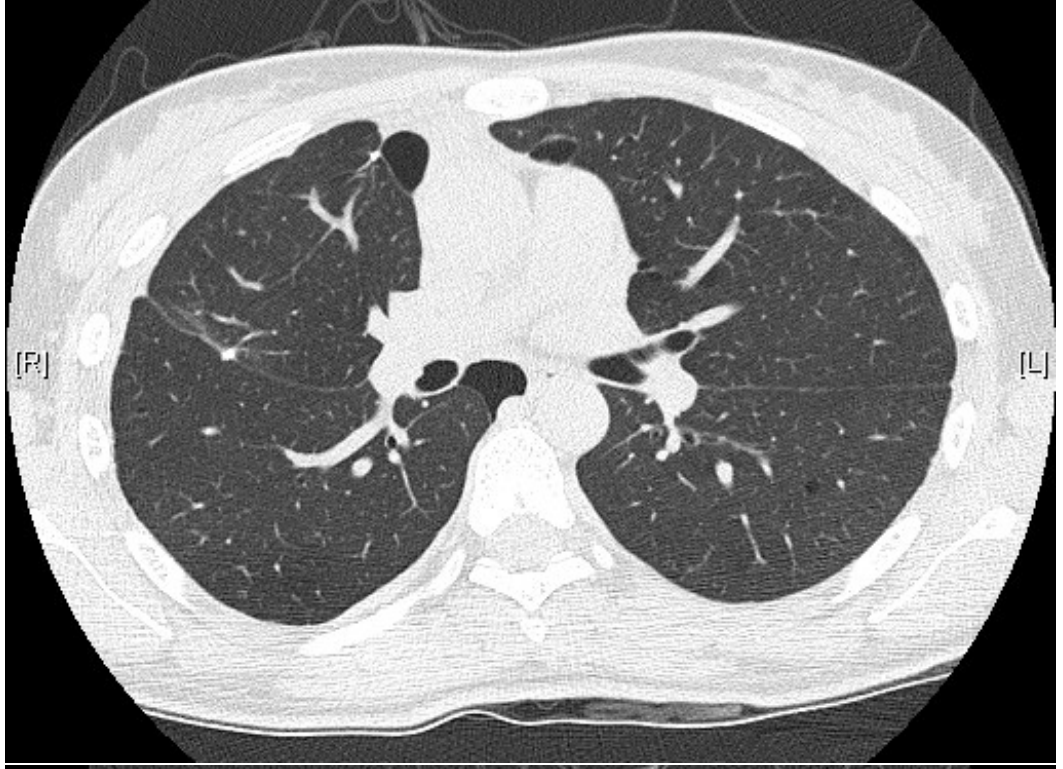

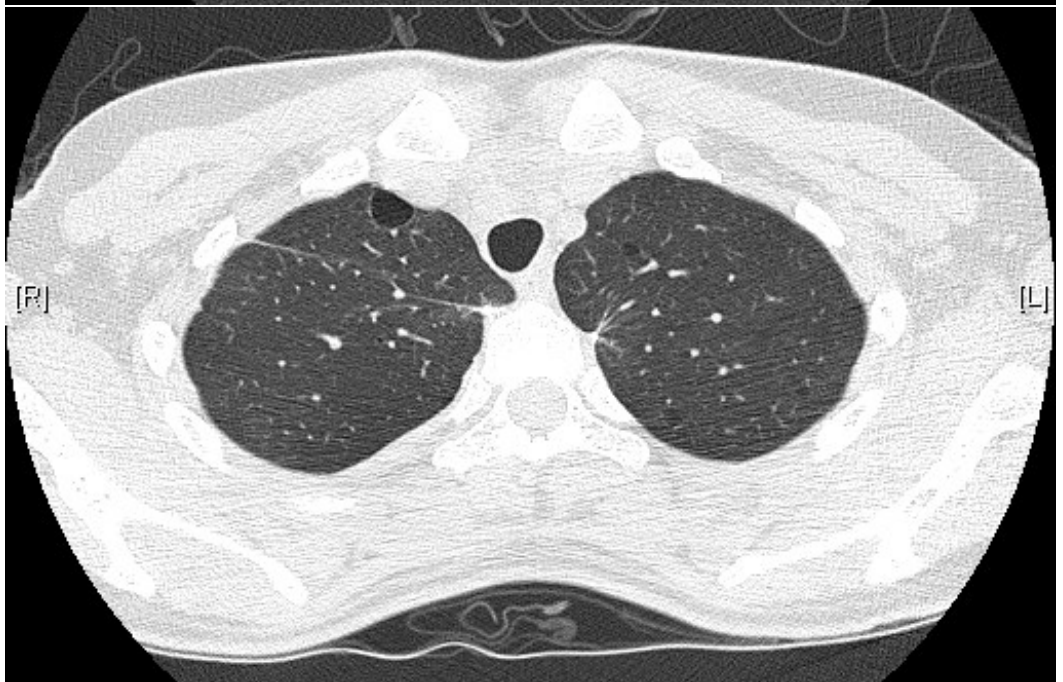

Clinical progress

- Bilateral pleurodesis was undergone
- Histology
  - para-septal emphysema
  - formation of subpleural blebs right upper and middle lobes, left upper and lower lobes.
  - No evidence of malignancy.

Investigation

- Her alpha anti-trypsin level is normal.
- Pulmonary function showed no airflow obstruction.

Clinical progress

- On the meanwhile, patient's elder sister (36 years old) also developed an episode of left spontaneous pneumothorax at later date, which was resolved with conservative treatment.
- The patient and the patient's elder sister did not present any skin lesions.

Diagnosis

- Their genetic test showed heterogeneous for a 1 BP duplication variant, suggestive of the heterogeneous status of Brit-Hogg-Dubé syndrome (BHDS).

Further investigation

- USG kidney for younger sister
  - absence of renal tumor.
- MRI kidney for elder sister
  - no renal tumor was detected.

Management Plan

- They have regular respiratory and urology follow up for BHDS

Discussion

- Birt-Hogg-Dubé syndrome (BHDS) is an autosomal dominant multi-systemic disorder.
- It is characterized by clinical manifestation including
  - skin hamartoma
  - renal tumors
  - pulmonary cysts with spontaneous pneumothorax

Birt Hogg Dube syndrome – Skin manifestation

- Skin hamartomas
  - Fibrofolliculomas
  - Trchodiscomas
  - Arochodons
- Location
  - Face
  - Neck
  - Upper trunk
- Morphology
  - smooth dome-shaped and white to fleshed color papules
- Age of presentation
  - 3rd to 4th decade

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Birt Hogg Dube syndrome – Pulmonary manifestation

- Lung cysts are a hallmark of lung involvement, causing an increased risk of pneumothorax
- BHDS related lung cysts
  - multiple, irregular shaped
  - variable in size and number
  - tend to be located at the basilar, subpleural, and mediastinal region of the lungs
    - in contrast to the typical apical location in the primary spontaneous pneumothorax and emphysema
- In our case series, both Chinese young adult female siblings presented with pneumothorax as a first presenting symptom.

Birt Hogg Dube syndrome – Renal manifestation

- Increased risk for developing different types of renal tumors
  - ranging from benign oncocytomas to malignant renal carcinomas
  - Approximately 30% of the patients will develop renal tumors which correspond to a seven-fold increased risk
- The most threatening complication of BHDS is renal cell carcinoma
  - About 15% of patients with BHDS by the age of 70
  - Regular abdominal imaging surveillance is important.

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