November 2015 Imaging Case of the Month

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Clinical History: A 48-year-old non-smoking woman with a history of hysterectomy and right oophorectomy, and cholecystectomy, otherwise previously healthy, presented with right-sided chest pain. A frontal chest radiograph (Figure 1) was performed.

Figure 1. Frontal chest radiograph.

Which of the following statements regarding the chest radiograph is **most accurate**?

1. The frontal chest radiograph shows a right-sided pneumothorax
2. The frontal chest radiograph shows abnormal mediastinal contours
3. The frontal chest radiograph shows bilateral pleural thickening
4. The frontal chest radiograph shows no abnormal findings
5. The frontal chest radiograph shows small lung volumes with multiple small nodules

Correct!

1. The frontal chest radiograph shows a right-sided pneumothorax

The chest radiograph shows a right-sided visceral pleural line, representing a right-sided pneumothorax (Figure 2).

Figure 2. Frontal chest radiography shows symmetric basal reticulation with a visceral pleural line (arrows) visible in the right lung base, representing a pneumothorax. Faint linear and reticular opacities are present in the bases bilaterally.

Additionally, the frontal chest radiograph shows basal linear and reticular opacities. While some of this finding, in the right medial base, could be attributed to passive atelectasis in the presence of the pneumothorax, similar findings are present in the medial left base in the left retrocardiac region. There is no evidence of small nodules, and lung volumes are not abnormally decreased. The mediastinal contours appear normal. No evidence of pleural thickening is seen

The patient was treated with right-sided thoracostomy tube drainage. She denied history of recent trauma or severe coughing. Focused discussion with the patient disclosed a previous right-sided pneumothorax several years earlier.

Which of the following entities is **associated with spontaneous pneumothorax?**

1. Endometriosis
2. Lymphangioleiomyomatosis
3. Malignant pleural mesothelioma
4. Metastatic sarcoma to the lung
5. All of the above

Correct!
5. All of the above

All of the entities listed can cause spontaneous pneumothorax. Lymphangioliomyomatosis is a cystic lung disorder that is well-known to predispose to, and may even present with, spontaneous pneumothorax. Endometriosis may rarely result in pleural implants that can cause spontaneous pneumothorax, and may be a major etiologic mechanism for catamenial pneumothorax, although endometriosis-related spontaneous pneumothorax has been known to present in the intermenstrual period as well. Both malignant pleural mesothelioma and sarcomas metastatic to the lung have rarely been reported to present with spontaneous pneumothorax.

The patient subsequently underwent thoracic CT for evaluation of the potential cause of her pneumothorax (Figure 3).

Figure 3. Axial unenhanced thoracic CT scan

Which of the following is **correct** regarding this thoracic CT examination?

1. The thoracic CT shows numerous, bilateral, thin-walled pulmonary cysts
2. The thoracic CT shows central thromboembolic disease
3. The thoracic CT shows randomly disseminated small pulmonary nodules consistent with miliary disease
4. The thoracic CT shows subtle inhomogeneous attenuation involving the lung parenchyma
5. The thoracic CT shows peribronchial and mediastinal lymph node enlargement

Correct!

1. The thoracic CT shows numerous, bilateral, thin-walled pulmonary cysts

The thoracic CT shows numerous, thin-walled, bilateral, randomly distributed pulmonary cysts (Figure 4).

Figure 4. Axial unenhanced thoracic CT shows numerous, bilateral, randomly distributed thin-walled pulmonary cysts (arrowheads), some of which are subpleural (arrowheads in B, D, E), others of which are more centrally located. A linear focus with hyperattenuation, consistent with pleurodesis, is present in the right anterior thorax inferiorly.

Central thromboembolic disease is not evident, but would be exceedingly difficult to demonstrate on unenhanced thoracic CT displayed only in lung windows. No lung nodules are present; there is no evidence of miliary disease. Aside from the pulmonary cysts, the pulmonary parenchymal attenuation appears relatively homogeneous. The visible hilar and mediastinal contours appear normal; no evidence of lymph node enlargement is seen.

Which of the following represents the **most likely** cause for this patient's spontaneous pneumothorax?

1. Catamenial pneumothorax / endometriosis
2. Esophageal tear
3. Lymphangioleiomyomatosis
4. Malignant pleural mesothelioma
5. Subpleural blebs

Correct!

3. Lymphangioleiomyomatosis

Based on the clinical history and the appearance of numerous, bilateral thin-walled pulmonary cysts, lymphangioleiomyomatosis (LAM) is the most likely cause of the patient's spontaneous pneumothorax among the entities listed above. Subpleural blebs can cause spontaneous pneumothorax, but usually such blebs are found in the extreme subapical region and are unassociated with lung cysts elsewhere. There is no evidence of pleural thickening or subpleural nodularity to suggest the presence of malignant pleural mesothelioma, and the patient is rather young for this diagnosis as well. An esophageal tear would present with pneumomediastinum in addition to pneumothorax, and the patient's history often will include an episode of retching prior to presentation; both are lacking in this case. Catamenial pneumothorax is a possibility, but no historical evidence connecting the pneumothorax development to menses is present, nor is there historical evidence suggesting endometriosis, and the patient is status-post hysterectomy.

The patient was diagnosed presumptive with LAM. She was noted to have several skin lesions that were thought to reflect the facial angiofibromas / angioleiomyomata of tuberous sclerosis complex (TSC) – LAM (Figure 5).

Figure 5. Detail photograph of the upper scalp skin lesions showing multiple, small, 2-4 mm white and skin-colored papules.

Which of the following is **atypical for** the presumptive diagnosis of tuberous sclerosis complex-LAM in this patient?

1. The absence of lung nodules is atypical for TSC-LAM
2. The appearance of the skin lesions is atypical for the angiofibromas / angioleiomyomata of TSC-LAM
3. The lung cyst distribution is atypical for TSC-LAM
4. The lung cyst morphology is atypical for TSC-LAM
5. 1 and 2

Correct!
5. 1 and 2

The angiofibromas / angioleiomyomata seen with tuberous sclerosis complex often are found on the face, in the nasolabial folds, cheeks, and skin, unlike the upper scalp in this patient. Angiofibromas / angioleiomyomata are typically pink or skin-colored telangiectatic papules, most commonly red, rather than the flesh color in this patient. The lung cyst distribution (random) is entirely consistent with lymphangioleiomyomatosis, as is the cyst's thin-walled morphology; therefore, answers 3 and 4 are not correct. However, small lung nodules are commonly present in patients with lymphangioleiomyomatosis, multifocal micronodular pneumocyte hyperplasia, but no such nodules are seen in this patient. The lack of these small nodules does not entirely mitigate against the diagnosis of TSC-LAM, but it does raise some suspicion for the accuracy of that diagnosis; therefore, answer 5 (both the absence of lung nodules and appearance of the skin lesions are atypical for the diagnosis of TSC-LAM) is the best answer.

Which of the following represents the **next most appropriate step** for the evaluation / management of this patient?

1. Biopsy of the scalp lesions
2. Bronchoscopy with transbronchial biopsy
3. Renal ultrasound
4. Repeat high-resolution thoracic CT
5. Thoracoscopic lung biopsy

Correct!

1. Biopsy of the scalp lesions

Biopsy of the scalp lesions represents the easiest, most direct route to obtain potentially useful information regarding the accuracy of the diagnosis of TSC-LAM in this patient. Bronchoscopy with transbronchial biopsy could be potentially useful in this regard, and thoracoscopic lung biopsy could establish the correct diagnosis, but both are needlessly invasive in light of a more accessible alternative. In fact, material from an outside thoracoscopic lung biopsy, from the right middle lobe, was subsequently obtained and showed only "emphysematous changes with bullae and blebs, and fibrinous pleuritis", the latter consistent with the history of previous pneumothorax. Repeat thoracic CT using high-resolution technique is not required- the imaging obtained thus far is adequate. Renal ultrasound could be of benefit should angiomyolipomas- the characteristic fat-containing lesion seen in the kidneys of patients with TSC and TSC-LAM- be detected, but angiomyolipomas may occur sporadically (unassociated with TSC-LAM), and the lack of their demonstration would not exclude the diagnosis of TSC-LAM.

The patient subsequently underwent biopsy of the scalp lesions, which showed trichodiscoma (aka fibrofolliculoma).

Which of the following entities are **associated with trichodiscomas**?

1. Alport syndrome
2. Birt-Hogg-Dubé syndrome
3. Carney complex
4. DiGeorge syndrome
5. Tuberous sclerosis

Correct!

2. Birt-Hogg-Dubé syndrome

Trichodyscoma, or fibrofolliculoma, is a hamartoma of the hair disc, and is a relatively specific finding for Birt-Hogg-Dubé syndrome, and does not occur in association with the other disorders listed. A number of skin lesions may be seen in patients with tuberous sclerosis, including facial angiofibromas, periungual fibromas, hypomelanotic macules, Shagreen patches (connective tissue nevi), and Confetti lesions. Dermatologic manifestations of Carney complex include myxomas and lentigines (hyperpigmentation). Alport syndrome is a hereditary nephritis characterized by glomerulonephritis and end-stage renal disease and hearing loss; skin disorders are not a primary manifestation of Alport syndrome. DiGeorge syndrome results from a chromosomal abnormality and is characterized by various combinations of congenital cardiac defects, palatal defects, recurrent infections resulting from T-cell abnormalities that may be associated with thymic hypoplasia, endocrine abnormalities, and facial dysmorphism, among other disturbances.

Subsequent intensive family history showed that at least 4 immediate family members had similar skin lesions. This finding, in concert with the other findings calling into question the accuracy of the diagnosis of TSC-LAM, prompted testing for the folliculin gene mutation that is often present in patients with Birt-Hogg-Dubé syndrome, and the testing was positive.

Diagnosis: Birt-Hogg-Dubé syndrome

References

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