Unilateral Lung Agenesis, Aplasia or Hypoplasia – which one is it?

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Abbreviations Used:
CT - Computed Tomography
FVC – Forced Vital Capacity
FEV1 – Forced Expired Volume in 1 second
TLC – Total Lung Capacity
RV – Residual Volume

Congenital lung malformations can lead to symptoms in the immediate newborn period or early childhood, but may also be diagnosed incidentally on routine imaging or autopsy, especially if the individual has remained asymptomatic. We report a case where incidental detection of abnormal intrathoracic structures led to a different diagnosis while being evaluated for scoliosis.

A 14-year-old female with scoliosis underwent thoracolumbar spine imaging for assessment of the angle of curvature of her scoliosis. Her radiographs showed dextroconvex scoliosis with complete shift of the mediastinum and the heart to the right side of the chest (Figure 1A). A
chest computed tomography (CT) with intravenous contrast revealed severe right lung aplasia with presence of right bronchus leading to small volume of residual lung parenchyma and complete shift of mediastinal structure in to right hemithorax (Figure 1B and 1C). Due to the mediastinal shift and inability to assess her cardiovascular anatomy, she also underwent cardiac magnetic resonance imaging which demonstrated absence of right pulmonary artery and veins, dilated left upper and lower pulmonary veins returning normally in to left atrium, bilateral superior vena cava, a rudimentary blind-ending right bronchus with nearly complete absence of the right lung parenchyma and entire cardiac mass in right chest with otherwise normal cardiac systolic function (images not shown here). Evaluation in the pediatric pulmonology clinic revealed a well-appearing, well-developed adolescent with heart sounds that were able to be auscultated only in the right chest and absent lung sounds in the right lung fields. On further questioning, patient did report a history of dyspnea with mild exertion, and was referred for complete pulmonary function testing and a 6-minute walk test.

The spirometry was suggestive of a very mild restrictive defect (decreased Forced Vital Capacity [FVC - 78%, 2.19L] and Forced Expired Volume in 1 second [FEV1 - 76%] with preserved FEV1/FVC ratio [0.88] and lung volumes measured by body plethysmography showed normal Total Lung Capacity [TLC - 3.93L, 114%], with air trapping (increased RV/TLC). The patient's 6-minute walk distance was moderately reduced (372.86 meters, 57.2% of the predicted distance: 652m), with no desaturations. Additional airway reconstruction from chest CT images showed a rudimentary right main bronchus which confirmed the diagnosis of right lung aplasia (Figure 1D).

A simplified classification scheme was recently proposed (Seear et al., 2017) (Online Supplement Table1) that classifies lung malformations into 3 broad categories. Unilateral absence or reduction of pulmonary parenchyma may be classified as a Group 3 malformation, and further differentiation between pulmonary agenesis, aplasia and hypoplasia depends on the

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extent of development of the airway and lung parenchyma on the involved side. Lung agenesis is generally associated with a complete absence of the pulmonary parenchyma, airways and vasculature of one side, while aplasia is associated with a blind ending mainstem bronchus and some lung tissue (Online Supplement Table 2).

Lung aplasia and agenesis are extremely rare disorders that originate in the embryonic stage of fetal lung development that may represent extremes of a spectrum of impaired lung development (Langston, 2003). It may be bilateral or unilateral with estimated frequency between 1/10,000 to 1/15,000 autopsies (Kayemba-Kay's et al., 2014). Right sided lung aplasia is generally less common and has been associated with worse prognosis, as it can be associated with congenital anomalies of the skeletal, cardiovascular, gastrointestinal and genitourinary systems (Booth & Berry, 1967, Maltz & Nadas, 1968). In addition, right lung aplasia may result in frequent respiratory infections secondary to greater mediastinal shift with resultant distortion of vascular structures and airways, with the resultant impairment of the clearance of airway secretions (Kushwaha et al., 2012). Cases of lung aplasia may present at any age with varying clinical presentations which include incidental diagnosis (Muthusami et al., 2010), mild chest pain (Barison et al., 2012), dyspnea on exertion, recurrent respiratory infections, and asthma-like symptoms (Kushwaha, Ranganath, Garg & Anand, 2012). There is limited data on long term outcomes in patients with lung aplasia, but prognosis generally remains poor when associated with congenital cardiac lesions. Further assessment of the etiology of lung aplasia or hypoplasia (Online Supplement Table 3) can be based on a combination of clinical and radiologic features. Management remains supportive, with emphasis on prevention of repeated chest infections to keep the remaining lung healthy.

Disclosures: None.

References:

**Figure Legend:**

Figure 1: Radiologic images of index case. Figure 1A shows the initial radiograph obtained during scoliosis evaluation which showed right lower zone opacity with complete mediastinal shift. Figure 1B shows the coronal images from mediastinal windows of the Chest Computed Tomography (CT) scan with heart in right hemithorax, and the left lung hyperexpanded to fill the upper part of the right chest. Figure 1C shows the same image as 1B in lung window demonstrating the lung parenchyma being normal. Figure 1D shows the 3-D construction of the airway from the CT images, with the arrow pointing out the blind ending right main stem bronchus.