JOURNAL OF THE ANATOMICAL SOCIETY OF INDIA 63 (2014) S43–S45

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Case Report

Pulmonary underdevelopment malformation with cleft lip and palate – A case report

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ARTICLE INFO

Article history:
Received 2 April 2014
Accepted 27 August 2014
Available online 20 September 2014

Keywords:
Absent pulmonary artery
Computed tomogram chest
Lung aplasia
Lung collapse
Rudimentary bronchus

ABSTRACT

Pulmonary underdevelopment malformations are rare. Ipsilateral facial defects and lung aplasia especially on right side are reported in literature. We present an adolescent with right cleft lip and palate incidentally detected to have aplasia of left lung with absent pulmonary artery after two general anaesthesia procedures. Computed tomogram is a good non invasive technique for a definitive diagnosis.

1. Introduction

Pulmonary underdevelopment malformations are rare and include the spectrum of lung agenesis, aplasia and hypoplasia. They occur in isolation or in association with other major anomalies.[1,2] They may be non-syndromic or part of syndromes like Goldenhar, VACTERL and CHARGE associations.[2] We report on an adolescent with cleft lip and palate incidentally detected to have aplasia of lung with absent pulmonary artery for its rarity. Etiopathogenesis and the role of imaging in diagnosis are discussed.

2. Case report

A 14 year old boy was referred for routine medical evaluation prior general anaesthesia (GA) for cheiloplasty revision. He

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http://dx.doi.org/10.1016/j.jasi.2014.08.006

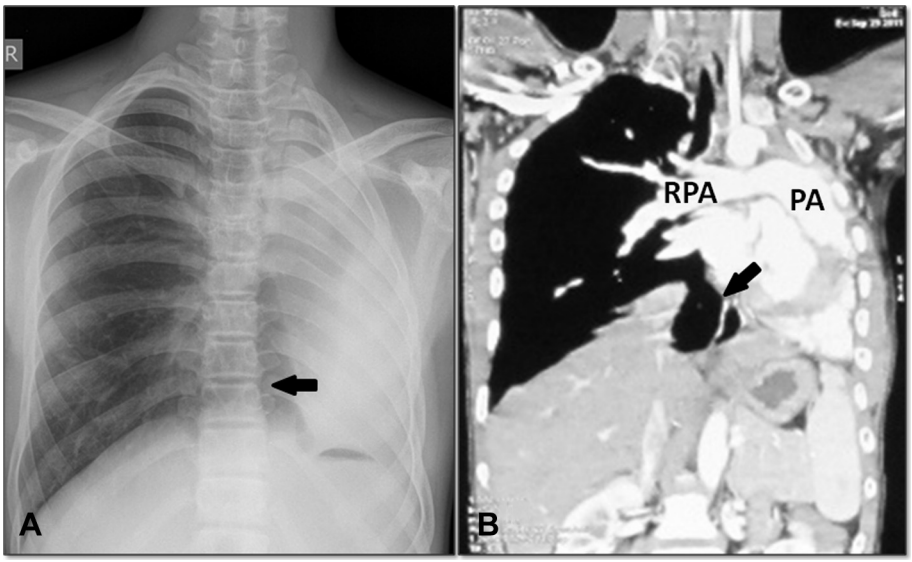

Fig. 1 – A. Radiograph of the chest showing opacified left hemithorax with crowding of ribs, ipsilateral tracheal and mediastinal shift. B. CECT chest coronal view demonstrating absent left lung parenchyma and dilated main (PA) and right pulmonary artery (RPA). Herniation of the right lung is indicated (Solid black arrow).

had undergone cheilo and palatoplasty under GA at six and 10 years of age elsewhere with no reported post operative events. There was no other significant medical history. On examination his growth centiles and vitals were normal. Healed surgical scars were seen over the right lip and hard palate. There were no other dysmorphisms to suggest a syndrome. There was no anaemia, clubbing and lymphadenopathy. Examination of respiratory system suggested left lung collapse with tracheal and mediastinal shift to left, decreased movement, dull percussion note and absent breath sounds on the left. Other systems were normal. Xray chest (Fig. 1A) confirmed left lung collapse. Contrast enhanced computed tomography (CECT) of the chest showed complete absence of left lung parenchyma with dilated main and right pulmonary arteries (Fig. 1B). The left pulmonary artery was absent (Fig. 2A). There was a rudimentary blind ending left bronchus (Fig. 2B). Echocardiogram was normal. With a final diagnosis of left lung aplasia, GA was deferred and the patient was advised to get trivial respiratory infections treated promptly.

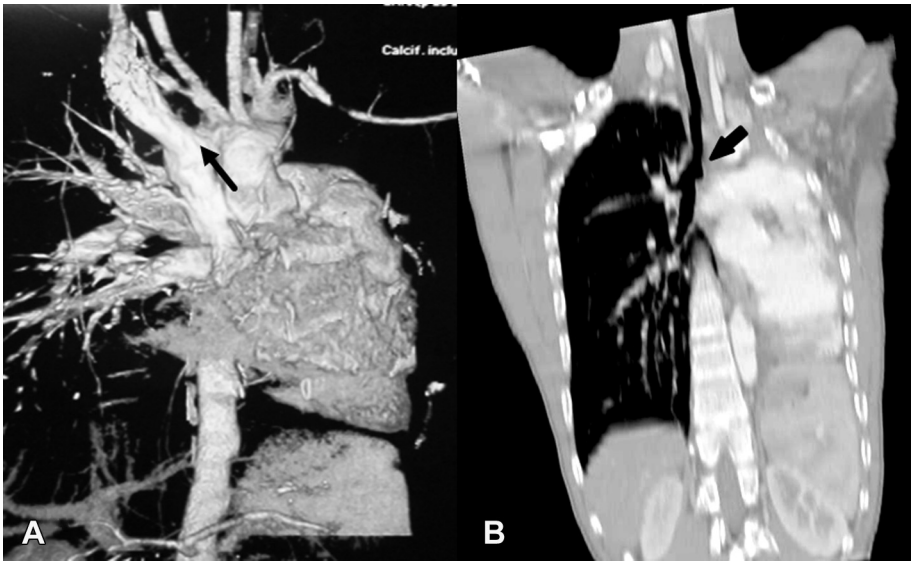

Fig. 2 – A. Reconstructed 3D volume-rendered image of the vascular structures shows the main pulmonary artery with the right branch (Black arrow). Left pulmonary artery is absent. B. CECT chest coronal view shows left rudimentary bronchus (Black arrow).

3. Discussion

Pulmonary underdevelopment malformations spectrum is morphologically divided into three broad groups based on the widely accepted classification by Schneider and Schwalbe.[1,3] Group I consists of lung agenesis with absence of any lung parenchyma, ipsilateral main bronchus and pulmonary artery. Lung aplasia which is group II is similar except that there is a rudimentary bronchus which ends in a blind pouch as in our case. Group III is lung hypoplasia with fully formed or hypoplastic bronchus and pulmonary artery with variable poorly developed lung tissue. Review of case reports suggest that agenesis and aplasia may be considered developmentally and clinically as one entity.[1] Approximately half of the cases are associated with cardiovascular, gastrointestinal (tracheo esophageal fistula, congenital diaphragmatic hernia), skeletal (vertebral, ipsilateral radial ray) and ipsilateral facial anomalies especially when the right side is affected.[1,2] In our case the cleft lip and palate and lung aplasia were on contra-lateral sides.

The critical time in the development of lung agenesis/aplasia appears to be between four and seven weeks of gestation during the embryonic phase of lung development when the laryngo-tracheal bud arises as a diverticulum of the primitive foregut and bifurcates into two lung buds. The etiopathogenesis of lung malformations are still poorly understood and genetic, teratogenic, mechanical and environmental factors have all been postulated.[3,4] An interruption of blood flow in the sixth aortic arch which develops into the pulmonary artery or dorsal root of aorta in the fourth week of gestation has been hypothesised when pulmonary agenesis occurs in isolation or in association with ipsilateral facial/limb defects.[2] The role of retinoic acid in the pathways critical for the initiation of lung morphogenesis and the molecular basis of failure to form lung buds in vitamin A deficiency has been established in animal studies.[4] Pulmonary agenesis in association with oral clefting is rare. In a French cohort of 238,942 births, 460 neonates had facial clefts of which only one had pulmonary agenesis.[5] Both lung and oro-facial developments are dynamic processes progressing through various phases and controlled by groups of homoeobox genes, transcription and growth factors. With no other features to suggest a syndrome in the propositus, the co-occurrence of the two lesions is probably due to a vascular disruption during development of branchial arches even though on contra-lateral sides.

Asymptomatic late presentation is rare and prognosis is good in the absence of recurrent respiratory tract infections. The primary diagnosis in our case was lung collapse having undergone two GA procedures earlier on. Prior to the availability of cross sectional imaging, differentiation of groups I and II depended on high exposure chest radiography, bronchogram, angiogram or autopsy.[1,6] Now multidetector computerized tomography which can employ CT angiography protocol with high resolution multiplanar and three-dimensional reconstruction has replaced the invasive procedures. It can demonstrate the vascular anatomy as well as the lung abnormalities. It can exclude other differentials like collapse due to foreign body, lymph node or aberrant artery. Though magnetic resonance imaging has the advantage of delineating vascular anomalies without any radiation, imaging evaluation of lung parenchyma and airways is limited.

To conclude, lung agenesis/aplasia needs to be considered in the differential diagnosis of opaque hemithorax on chest radiograph especially in the presence of other congenital anomalies. Computed tomogram is a good non invasive technique for a definitive diagnosis.

Conflicts of interest

All authors have none to declare.

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