

# Uncommon Coexistence of Isolated Left Lung Aplasia and Thalassemia Minor: Case Report

## Izole Sol Akciğer Aplazisi ile Talasemi Minörün Nadir Birlikteliği

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**ABSTRACT** Pulmonary aplasia is an uncommon anomaly that has been reported in isolation and in association with other congenital defects. Such defects include oesophageal atresia, cardiac malformation, horseshoe kidney, and anal atresia. A 16 year-old girl was referred for an opaque left hemithorax for which she had received antibiotherapy for pneumonia. A detailed evaluation including contrast computed tomography of the thorax, fiberoptic bronchoscopy, and ventilation-perfusion scintigraphy led to diagnosis of left lung aplasia. Additionally in further investigations we found thalassemia minor both in the patient and her mother. We discussed pulmonary aplasia and the likely association between thalassemia minor and development of the lungs during pregnancy. To the best of our knowledge, this is the first reported case of unilateral lung aplasia associated with thalassemia minor.

**Key Words:** Lung; abnormalities; beta-thalassemia

**ÖZET** Pulmoner aplazi nadir bir anomali olup diğer konjenital defektlerle ile olan ilişkisi daha önceden tanımlanmıştır. Bu konjenital defektler özefagus atrezisi, kardiyak malformasyon, atrnal böbrek ve anal atreziyi içermektedir. Kliniğimize akciğer grafisinde sol hemitoraksta opasite olup, pnömoni nedeniyle antibiyotik tedavisi gören 16 yaşında bir kız çocuğu refere edildi. Kontrastlı toraks tomografisi, fiberoptik bronkoskopi, ventilasyon-perfüzyon sintigrafisini içeren detaylı incelemede, hastaya sol akciğer aplazisi tanısı kondu. İleri araştırmamızda hastamızın hem kendisinde hem de annesinde talasemi minör tespit ettik. Bu olgu sunumunda pulmoner aplazi ve talasemi minörün hamilelik esnasında akciğerlerin gelişimi ile olan ilişkisini tartıştık. Elimizdeki bilgilere göre talasemi minör ile ilişkili tek taraflı akciğer aplazisi olgusu rapor edilen ilk olgudur.

**Anahtar Kelimeler:** Akciğer; aplazisi; talasemi minör

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**C**ongenital malformations of the lung are rare disorders occurring with variable degree of severity. These are the result of insult to the developing embryo during the 4<sup>th</sup> and 24<sup>th</sup> weeks of intrauterine life.<sup>1</sup>

Radiographically, agenesis of a lung simulates pneumonectomy. The remaining lung is overinflated with accompanying shift of the mediastinum. Boyden clearly categorised these congenital anomalies as pulmonary agenesis, aplasia and hypoplasia. In pulmonary agenesis, carina, main bronchi, lung tissue, and vascular structures are not involved sites. Pulmonary

aplasia is different from pulmonary agenesis in that a pouch-like, blind-ending main bronchus and carina are present. Pulmonary parenchyma and vessels are absent.<sup>1</sup>

Hemoglobinopathies that lead to decreased production of globin chains ( $\alpha$  or  $\beta$ ) produce a clinical syndrome characterized by anemia of variable severity with hypochromic and microcytic red cells. The  $\alpha$  and  $\beta$  thalassemias result, respectively, from deletions or mutations of the  $\alpha$  and  $\beta$ -globin genes that lead to decreased transcription or translation of the gene product.<sup>2</sup>

$\beta$ -Thalassemias usually result from mutations that affect transcription, translation, or RNA stability. When a single  $\beta$ -globin gene is affected,  $\beta$ -thalassemia minor results. Erythrocytosis and a mild hypochromic microcytic anemia characterize this heterozygous condition. High pressure liquid chromatography (HPLC) analysis shows an increase in Hb A2 and, in some cases, Hb F.<sup>3</sup>

A few studies including small numbers of patients suggested a favorable perinatal outcome of patients with  $\beta$ -thalassemia minor.<sup>3</sup> There are few reported adverse outcome caused by hypercoagulability.<sup>4</sup> Oligohydramnios is associated with intrauterine growth restriction (IUGR) and might be part of the relative hypoxemic state.<sup>5</sup>

We report a 16 year old girl with complaints of cough and sputum who had isolated left lung aplasia as confirmed by contrast computed tomography (CT) of the thorax, fiberoptic bronchoscopy (FOB), and ventilation-perfusion scintigraphy and, in addition, as a further diagnosis, thalassaemia minor. To our knowledge, it is the first report of the co-occurrence of these two congenital abnormalities.

## CASE REPORT

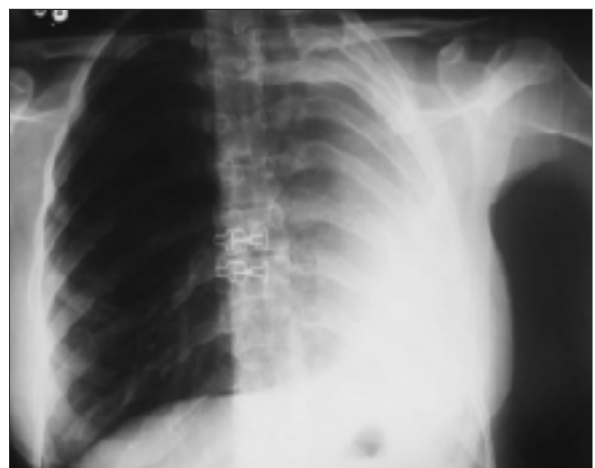
A 16-year-old girl with the symptoms of cough and sputum who was treated several times as pneumonia in a cottage hospital was admitted to our hospital.

Physical examination revealed a girl in no acute distress. There was no clubbing or cyanosis. Normal vesicular breath sounds were audible on the

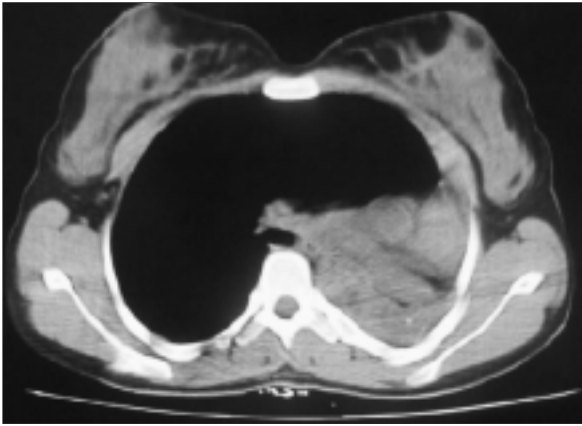
right side along with polyphonic rhonchi, but the intensity of breath sounds on the left side was vastly reduced. Heart sounds were normal in character and intensity.

The posterior-anterior chest roentgenogram revealed no lung tissue at the left hemithorax. The trachea and mediastinal structures were found to be herniated to the left (Figure 1A). CT revealed no left pulmonary artery and preaortic and precardiac herniation of the normal lung with left and posterior displacement of the heart was also seen (Figure 1B). On ventilation perfusion scintigraphy, no radioactivity uptake was seen at the left lung. FOB visualised a short left main bronchus, which ended in a blind pouch. The tracheobronchial tree on the right side was normal. Electrocardiogram showed low voltage normal waves with clockwise rotation of the heart. An echocardiogram demonstrate a normal pulmonary artery pressure of 20 mmHg with normal lumen; however, the bifurcation of pulmonary artery could not be seen. Atrial and ventricular functions were essentially normal. A skeletal survey and an ultrasonographic examination of the abdomen were normal.

The laboratory tests revealed mild hypochromic microcytic anemia. Further investigation like HPLC analysis show an increase in Hb A2. A fam-



**FIGURE 1A:** On the posterior-anterior chest roentgenogram, there is no visible lung tissue in the left hemithorax. The right lung shows compensatory hyperinflation passing through to the left from the midline. The trachea deviates to the left.



**FIGURE 1B:** The anterior part of the right lung passes through the anterior mediastinum to the left hemithorax. The ascending and descending parts of the aorta and the main pulmonary artery are located in the left hemithorax.

HPLC: High pressure liquid chromatography, CT: Computed tomography, FOB: Fiberoptic bronchoscopy, IUGR: Intrauterine growth restriction.

ily history of thalassemia minor was elicited from her mother too.

Written informed consent was obtained from the patient.

Finally a diagnosis of isolated left lung aplasia with thalassemia minor was made.

## DISCUSSION

Unilateral pulmonary aplasia is a rare condition and associated with some other abnormalities in more than 50% of the patients mainly involving the cardiovascular, gastro-intestinal, musculoskeletal, urogenital systems. Pulmonary aplasia is usually seen in infancy or early childhood. It is not common in adults. Patients who have no or mild associated anomalies may survive into adulthood.<sup>6</sup> In our patient we have found thalassemia minor and to our best knowledge this is the first case of the co-occurrence of these two congenital abnormalities.

Its frequency is estimated at 1:15,000 autopsies. The development anomalies of the lung at the 4<sup>th</sup> and 24<sup>th</sup> gestational weeks may cause bronchopulmonary foregut abnormalities. Etiology is not completely known but vitamin A deficiency, viral

agents, or genetic factors are discussed.<sup>6,7</sup> Both of our patient and her mother have thalassemia minor. Chronic maternal anemia during gestation might lead to fetal hypoxia, predisposing the fetus to IUGR and oligohydramnios.<sup>8,9</sup> Maternal hypoproteinemia and anemia might be associated with hydrops fetalis and lung hypoplasia.<sup>10</sup> So fetal hypoxia caused by thalassemia minor may play a negative role in the intrauterine development of the lung.

Radiographic findings are generally diagnostic and include mediastinal displacement, the hyperinflation of a normal site, and decreased volume in a hypoplastic lung.<sup>11</sup> In our patient, thorax CT showed compensatory hypertrophy in the contralateral lung and the displacement of mediastinal structures to the left side.

Differential diagnosis includes total atelectasis, pneumonectomy, pulmonary hypoplasia, diaphragmatic evantration, pneumonia and pleural effusion.<sup>11</sup> Our case was diagnosed with contrast CT, ventilation-perfusion scintigraphy and FOB.

Dyspnea, recurrent pneumonia due to blind-ending bronchi, and a susceptibility to infection affect the prognosis. Limited surgical intervention with the resection of blind-ending bronchi can be performed in such patients.<sup>11</sup> In our patient after a medical treatment she had no complaint and she was good for one and a half year.

In the current report the patient had pulmonary aplasia with thalassemia minor and her mother had also thalassemia minor too. We argued the probable means for the formation of the pulmonary aplasia, which might be well related to thalassemia minor. Thalassemia minor during pregnancy could make hypoxia and this could affect the development of the lungs.

In conclusion, to our knowledge this is the first reported case of isolated left lung aplasia associated with thalassemia minor. During the pregnancy of patients who have thalassemia minor effort should be achieved to detect if there is a congenital anomalies or not.

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