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CASE REPORT

Agenesis of the lung

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ABSTRACT: Agenesis of the lung is extremely rare. Developmental defects of the lung are usually associated with other congenital malformations. Half of all reported patients die either at birth or within the first 5 yrs of life. We report a case of right lung agenesis with absence of the left kidney, and fusion anomaly between ribs 4 and 5 on the left hemithorax.
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Agenesis, affecting both hemithoraces and both sexes almost equally, should be separated for physiological reasons from hypoplasia by the total absence of lung parenchyma, bronchial tree, and supporting vasculature [1]. This extremely rare condition was first described by De Pozze who discovered it accidentally at the autopsy of an adult female in 1673 [2]. Genetic, teratogenic, and mechanical factors may have a bearing on the aetiology. Developmental defects of the lung are usually associated with other congenital malformations, which include the skeletal, cardiovascular, gastrointestinal, and genitourinary systems [1–8].

Case report

A 24 day old, white female infant was admitted to the hospital for evaluation of dyspnoea, cough and cyanosis on crying. After a normal term pregnancy, the labour and delivery were unremarkable. The birthweight was 2600 g. The baby cried spontaneously but soon became dyspnoeic.

Physical examination produced the following findings: body weight 2750 g, length 48 cm, pulse rate 160·min^{-1}, respiratory rate 80·min^{-1}. The patient had tachypnoea, the right hemithorax was flat to percussion, and the left was dull. Heart sounds were loudest to the right of the sternum, without a murmur. With positive pressure oxygen followed by high enviromental oxygen, there was improvement.

X-ray study revealed a homogeneously dense right hemithorax with fusion anomaly between ribs 4 and 5 on the left hemithorax (Fig. 1). Pulmonary scintigraphic examination revealed a complete absence of perfusion in the right lung, and a partial absence of perfusion at the apex of the left lung (Fig. 2). The *i.v.* pyelogram (Fig. 3) and abdominal ultrasonogram disclosed absence of the left kidney as well as hydronephrosis and hydroureter in the right kidney. Electrocardiographic examination and blood chemistry analysis were within normal limits.

The child expired on the forty-fifth day. The parents did not give consent for postmortem examination.

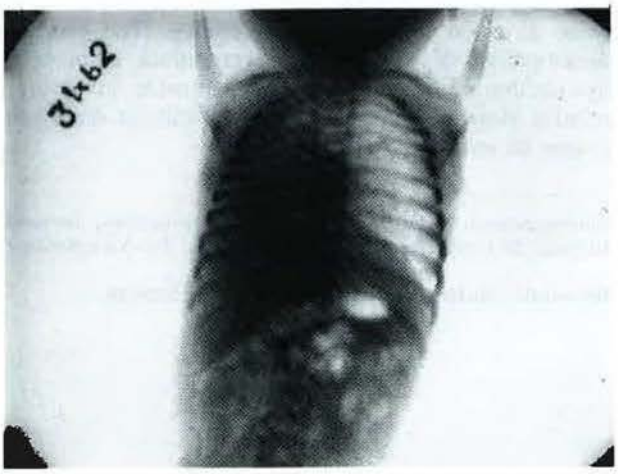

Fig. 1. – Anteroposterior view of chest made on the 24th day of life. There is complete opacity of the right hemithorax. The heart and mediastinal contents are displaced entirely into the right side. The left lung is somewhat emphysematous, the ribs on that side are more widely spread, and the diaphragm is flattened. Also there is a fusion anomaly between left ribs 4 and 5.

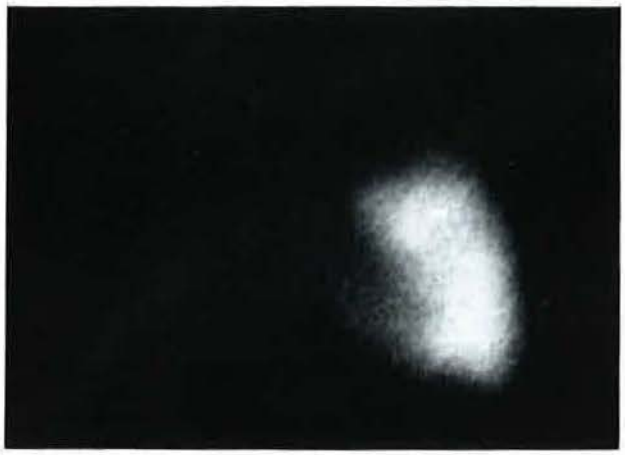

Fig. 2. – Pulmonary scintigraphy.

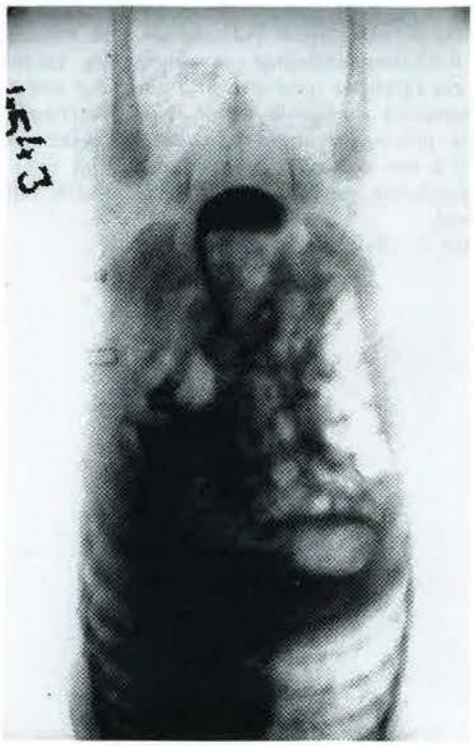

Fig. 3. – Intravenous pyelogram.

Discussion

Agenesis of the lung may be defined as total absence of bronchi beyond the bifurcation, pulmonary parenchyma and pulmonary and bronchial vasculature [1]. All gradations of underdevelopment of the lung are encountered, ranging from hypoplasia of one segment or lobe to complete absence of both lungs. Unilateral pulmonary agenesis is extremely rare and in most of the less than 200 examples recorded there were additional congenital anomalies [3].

In 1953, OYAMADA *et al.* [9] found, in a survey of the world literature, 109 reported cases of all varieties. Fifty of these were right-sided defects, 59 left-sided.

Agenesis of the lung itself does not give rise to symptoms unless complicated by bronchopulmonary disease. In most cases the patients are investigated because of repeated chest infections [8]. The time of onset of symptoms is remarkably variable. Some infants are in great difficulty at birth and survive only one or two gasping respirations. The presenting symptom is, at times, tachypnoea without cyanosis; tachypnoea occurs with cyanosis only upon undue exertion. More often, the presenting complaint has been repeated attacks of lower respiratory tract infection, described as bronchitis, asthmatic bronchitis, or pneumonia [7].

Physical examination reveals characteristic signs. The affected hemithorax may be slightly flattened, but usually little difference can be found in the size of the two sides. One side, however, moves less well than the other. On this side, the percussion note is at first flat from top to bottom, front and back, but later some resonance appears over the upper part of the chest in front, where the emphysematous normal lung has herniated across the midline. Breath sounds follow the same pattern, completely absent over one lung at first, later present but diminished over the upper lobe in front. The heart and mediastinum are found, by percussion and auscultation and by observation and palpation of the apex beat, to have shifted far to the affected side. The note on the good side is resonant or hyperresonant, the breath sounds are stronger, and no adventitious sounds are heard [7].

Chest roentgenograms suggest the diagnosis, but careful bronchographic, angiocardiographic, and echocardiographic studies, as well as computed tomography in some instances, are necessary for confirmation [1, 3, 4, 10]. Other X-rays studies (*e.g. i.v.* pyelogram) must also be conducted for other possible malformations. In our case, in addition to absence of the left kidney, a fusion anomaly between ribs 4 and 5 on the left hemithorax was observed.

The aetiology of these malformations remains unknown. The high incidence (greater than 50%) of associated cardiac, gastrointestinal, genitourinary, and skeletal malformations, as well as frequent abnormalities in the bronchopulmonary vasculature, lends support to generalized teratogenic factors [1, 7, 8]. On the other hand, SCHOBER *et al.* [11] reported a case with pulmonary agenesis in partial trisomy 2p and 21q.

Half of all the reported patients die either at birth or within the first 5 yrs of life [1, 9, 12]. Mortality and morbidity are related to complications and associated cardiac anomalies as well as severity of the lung anomaly. Death is due to progressive respiratory failure following a complicated clinical course. Severe respiratory infections are common in infancy and may lead to pneumonia and death [1, 12].

Individuals with agenesis of the left lung have a longer life expectancy than do those with agenesis of the right lung. This is probably related to a more severe mediastinal and cardiac displacement. The excessive mediastinal shift and malrotation of the carina, which is greater in right than in left lung agenesis, hinders proper drainage of the functioning lung and

reduces the resistance to respiratory infection. This may account for the higher mortality and morbidity [1, 7, 8, 12].

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Agénésie pulmonaire. Observation clinique. S. Kaya, U. Dilmen.
RÉSUMÉ: L'agénésie pulmonaire est extrêmement rare. Des troubles du développement pulmonaires sont habituellement associés à d'autres malformations congénitales. La moitié de tous les cas rapportés meurent soit à nasisance soit au cours des 5 premières années de la vie. Nous décrivons un cas d'agénésie pulmonaire droite associée à l'absence du rein gauche et à des anomalies de fusion entre les cotes 4 et 5 dans l'hémithorax gauche. La littérature actuelle fait l'objet d'une revue.
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