

Pulmonary Agenesis in a Newborn

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There are all transitions from hypoplasia of the lung to agenesis or aplasia of a lung lobe and to complete absence of the entire lung system as classifies such malformations as follows: a) unilateral complete absence of lung and bronchus, b) absence of one half of the lung and a short blind course of the associated bronchus, c) trunk bronchus formed, lung as a hazelnut- to fist-sized, unflap fist- sized, unlobed, fleshy structure in the mediastinum [1-31]. The malformations mentioned in a) and b) occur very rarely, somewhat more frequently the others. Concomitant malformations are often, but by no means always, observed in pulmonary agenesis: Tracheal anomalies, diaphragmatic defects, intestinal malformations, absence of a kidney, facial malformations, auricular appendages, congenital heart defects and oesophageal atresia [6,7,8-22]. Most of the cases are left-sided concomitant malformations [1,14,17,22]. A familial occurrence has been reported, but the family history was not very helpful in this regard [1-31].

The admission of the newborn took place during the U3 examination (German standard). The parents reported the grandmother had already complained on the boy's second day of life about the "special breathing" in the sense of asymmetrical thorax. The parents noticed the "special breathing" in the sense of asymmetric thorax on the second day of life. The patient had intermittent retractions at night, coughing or a cough had also occurred. The drinking behavior was good, reports show weight gained. In terms of nutrition, the patient received breast milk. The micturition and defecation behavior was inconspicuous. The birth weight was 3370 grams. U2 revealed a bent foot left, an icteric and hypotonic child. Clinical examination findings were as follows: Alert child, pale pinkish skin color. Fontan pervious, oral mucosa moist, clavicles intact. No efflore and rhythmic, no heart murmur, heart rate 120/min. Tachypnea AF 48/min, puerile breath sound left-sided AG attenuated from normal volume, no accessory sounds, SpO₂ 100% below high abdomen in level, ubiquitous bowel sounds, softly depressible inguinal pulses palpable bilaterally, genitals unremarkable, testes bilaterally regular, side-to-side spontaneous motor activity in supine and lateral positions. On admission, the temperature was 36.6 °C, pulse: 139/min, SaO₂: 100 % below 7L/min highflow and FiO₂ 0,21, Blood pressure: 118 mmHg /66 mmHg, Somatogram: Weight: 4500 grams. 56 cm, KOF: 0.26 m2, BMI: 14.35 k percentiles <4 months. Cor pure with abutting respiratory pattern, right side nearly ubiquitous low (5L/min, FiO₂ 0.21). The right apical lung portions, described as consolidations on chest radiograph, presented on bronchoscopy of

right upper and middle lobes. To exclude further malformations, an ultrasound of the abdomen was performed. The examiner recognized no dilated retrovesical ureters, hepatosplenomegaly but found a small gallbladder. A request for co-evaluation and tethering was performed. In echocardiography a small ASD II appeared. In addition, significant hypoplasia of the right pulmonary flattening is from a slightly increased right ventricular pressure severe PAH. Follow up is scheduled in 3 months. During the course of the inpatient stay, respiratory support was necessary and the newborn was stable even overnight. Oxygen saturation was always above 97%, pCO₂ was also high-flow support capillary always below 55 mmHg, respiratory support was discharged at home.

Pulmonary agenesis is a congenital malformation of the lung in which sections of the lung or the entire lung are not created on one or both sides [1-31]. If only rudimentary, non-functional lung tissue is present, the condition is also referred to as pulmonary aplasia [26-28]. Pulmonary agenesis is very rare, with approximately 45 cases described to date. The exact cause of pulmonary agenesis (and aplasia) is currently not definitively understood. A major cause is thought to be perfusion defects in the fetal period. Pulmonary agenesis is associated with agenesis of the pulmonary vessels. Concomitant cardiovascular, renal, and spinal malformations as well as esophagotracheal and rib abnormalities are common. Occurrence with VACTERL syndrome has also been described [6,7,8-22]. Three types can be distinguished in pulmonary agenesis [23]. Type I shows a very rare unilateral absence of the main bronchus (may also be present as a rudimentary stump), pulmonary vessels, and lung parenchyma. Left-sided pulmonary genesis is sometimes associated with tetralogy of Fallot. In type II, lung tissue and vessels are absent, and a bronchial bulge is seen, in type III a rudimentary main bronchus with bifurcation is present. The surrounding area consists of undifferentiated lung tissue, and the vessels are absent. Pulmonary agenesis or aplasia more commonly affects only one lobe of the lung (primarily the upper and middle lobes) than the entire lung. In some neonates, pulmonary agenesis is relatively asymptomatic; in severe cases, severe respiratory failure develops in the first hours of life. Complete bilateral pulmonary agenesis is incompatible with life. On chest x-ray, unilateral pulmonary agenesis or aplasia appears as a relatively dense, homogeneous shadowing with ipsilateral mediastinal displacement. The healthy lung is usually compensatory hyperinflated and herniates over the anterior superior mediastinum to the affected side. Furthermore, the healthy lung shows compensatory increased

pulmonary vascularity. The hemithorax of the affected side may be reduced in size. Differentiation between agenesis and aplasia is possible only by computed tomography or magnetic resonance imaging. Lung malformations also occur as part of syndromes. This is the case in Ellis-Yale-Winter syndrome, a very rare autosomal recessive disease, an incidence of 1:1000000, with a combination of microcephaly, heart defect and lung malformation. It has been described in three female sibs (including a fetus). Dysmorphic features were not characteristic. The condition seems to be hereditary, and transmitted as an autosomal recessive trait. Prognosis of Ellis-Yale-Winter syndrome is poor and all infants have died in infancy till date [32-34].

Declaration

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Conflict of Interest

None

Informed Consent

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Author Contributions

SB wrote the article, LB made suggestions and corrections of the references, EL worked on manuscript, EMA checked the references in detail. The data supporting the findings of this study are available from the corresponding author upon reasonable request. Any inquiries regarding supporting data availability of this study should be directed to the corresponding author.

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