ISSN: 2755-0141

Japan Journal of Clinical & Medical Research

SCIENTIFIC Research and Community

Letter to Editor

Open Access

Pulmonary Agenesis in a Newborn

Stefan Bittmann*, Elisabeth Luchter, Lara Bittmann, Elena Moschüring-Alieva and Gloria Villalon

Department of Pediatrics, Ped Mind Institute (PMI), Gronau, Germany

***Corresponding author**
Stefan Bittmann, Department of Pediatrics, Ped Mind Institute (PMI), Hindenburgring 4, D-48599 Gronau, Germany.

Received: April 20, 2023; **Accepted:** April 24, 2023; **Published:** April 30, 2023

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, ubuiquitous 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, Sa02: 100 % below 7L/min highflow and Fi02 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 ubuiquitous low (5L/min, Fi02 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 agenesia 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
Acknowledgments
No

Financial Disclosure or Funding
No

Conflict of Interest
None

Informed Consent
Obtained

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.

References

1. Bonacci W, Lazzaroni Fossati F, Bruschettini PL, Magliano P, Campone F, et al. (1983) Agenesia polmonare [Pulmonary agenesis]. Pathologica 75: 133-138.
2. Haas RJ, Schäfer H, Sigmund E, Tosberg P (1972) Unilaterale Lungenagenesie [Unilateral pulmonary agenesia]. Klin Padiatr 184: 135-139.
3. Esipova IK, Vladimirtseva AL (1996) K probleme nekotorykh vrozhdennykh porokov razvitiia legkogo [Congenital malformations of the lungs]. Arkh Patol 58: 49-54.
4. Delgado-Peña YP, Torrent-Vernetta A, Sacoto G, de Mir-Messa I, Rovira- Amigo S, et al. (2016) Hipoplasia pulmonar: análisis de la casuística durante 20 años [Pulmonary hypoplasia: An analysis of cases over a 20-year period]. An Pediatr 85: 70-76.
5. Jend HH, Schmidt M (1980) Sonographische Klärung einer Lungenagenesie beim Neugeborenen [Sonographic clarification of pulmonary agenesia in newborn (author's transl)]. Rontgenblatter 33: 395-398.
6. Strunge P (1972) Infantile lobar emphysema with lobar agenesia and congenital heart disease. Acta Paediatr Scand 61: 209-212.
7. Secouard M, Warnet JF, Assir S, Hervé J (1979) Un cas d'agénésie pulmonaire avec asymétrie faciale chez un nouveau-né [Pulmonary agenesia with facial asymmetry]. Ann Pediatr (Paris) 26: 122-124.
8. De Rosario JL, Braaten V, Dawkins W (1966) Segmental pulmonary alveolar agenesia of congenital origin. Int Surg 46: 147-151.
9. Gibon Y, Borde J, Mitrofanoff P, Lefort J (1978) A propos d'une rare association malformative Agénésie du poumon et de la coupole diaphragmatique gauches associée à une atrésie de l'oesophage [Association of left diaphragmatic hernia, lung agenesia and esophageal atresia (author's transl)]. Chir Pediatr 19: 261-267.
10. Bretagne MC, Hazeaux M, Deschamps JP, Pernot C, Werner J, et al. (1972) Diagnostic radiologique des agénésies et hypoplasies pulmonaires. A propos de 8 observations du C.H.U. de Nancy [Radiological diagnosis of pulmonary agenesia and hypoplasia. Apropos of 8 cases of the Nancy University Hospital Center]. J Radiol Electrol Med Nucl 53: 125-132.
11. Castejón Casado J, Jiménez Alvarez C (1989) Atresia de esófago y fístula traqueoesofágica asociada a agenesia pulmonar derecha [Esophageal atresia and transesophageal fistula associated with right pulmonary agenesis]. An Esp Pediatr 56: 813.
12. Viñuales-Aranda MD, Rodriguez-Sanz J, Gomez-Miranda RI (2022) A Silent Pulmonary Agenesis. Arch Bronconeumol 30: S0300-2896.
13. Cotten CM (2017) Pulmonary hypoplasia. Semin Fetal Neonatal Med 22: 250-255.
14. Greenough A, Ahmed T, Broughton S (2006) Unilateral pulmonary agenesis. J Perinat Med 34: 80-81.
15. de Benedictis FM, Pozzi M (2007) Pulmonary agenesis. Pediatr Pulmonol 42: 480-481.
16. Pimenta DA, Aguiar FL, Fernandes BC, Rolo R (2021) Late diagnosis of pulmonary agenesis. BMJ Case Rep 14: e245233.
17. Fukuoka S, Yamamura K, Nagata H, Toyomura D, Nagatomo Y, et al. (2022) Clinical outcomes of pulmonary agenesis: A systematic review of the literature. Pediatr Pulmonol 57: 3060-3068.
18. Rosenberg DM (1962) Pulmonary agenesis. Dis Chest 42: 68-73.
19. Jones HM, Howells CH (1961) Pulmonary agenesis. Br Med J 2: 1187-1189.
20. Katz S (1954) Pulmonary agenesis. GP 10: 47.
21. Vyas S, Mathew T, Advani M, Meena D (2018) Pulmonary agenesis: A rare entity. Lung India 35: 275-276.
22. Jentzsch NS (2014) Unilateral pulmonary agenesis. J Bras Pneumol 40: 322-324.
23. Malcon MC, Malcon CM, Cavada MN, Caruso PE, Real LF (2012) Unilateral pulmonary agenesis. J Bras Pneumol 38: 526-529.
24. Russell BC, Whitecar P, Nitsche JF (2014) Isolated unilateral pulmonary agenesis and other fetal thoracic anomalies. Obstet Gynecol Surv 69: 335-345.
25. Cunningham ML, Mann N (1997) Pulmonary agenesis: a predictor of ipsilateral malformations. Am J Med Genet 70: 391-398.
26. JB, Berry CL (1967) Unilateral pulmonary agenesis. Arch Dis Child 42: 361-374.
27. Claireaux AE, Ferreira HP (1958) Bilateral pulmonary agenesis. Arch Dis Child 33: 364-366.
28. Kim JY, Kim WS, Lee KS, Je BK, Park JE, et al. (2021) Posterior Lung Herniation in Pulmonary Agenesis and Aplasia: Chest Radiograph and Cross-Sectional Imaging Correlation. Korean J Radiol 22: 1690-1696.
29. Kayemba-Kay's S, Couvrat-Carcauzon V, Goua V, Podevin G, Marteau M, et al. (2014) Unilateral pulmonary agenesis: a report of four cases, two diagnosed antenatally and literature review. Pediatr Pulmonol 49: E96-102.
30. Dembinski J, Kroll M, Lewin M, Winkler P (2009) Unilaterale pulmonale Agenesie, Aplasie und Dysplasie [Unilateral pulmonary agenesis, aplasia and dysplasia]. Z Geburtshilfe Neonatol 213: 56-61.

31. Ordóñez Dios IM, Rienda Moreno MÁ, Lázaro Polo J (2020) Right Lung Agenesis Associated with Dextrocardia and Pulmonary Hypertension. Arch Bronconeumol (Engl Ed) 56: 390.
32. Nazir Z, Qazi SH, Ahmed N, Atiq M, Billoo AG (2006) Pulmonary agenesis-- vascular airway compression and gastroesophageal reflux influence outcome. J Pediatr Surg41: 1165-1159.
33. Field CE (1946) Pulmonary Agenesis and Hypoplasia. Arch Dis Child 21: 61-75.
34. Ellis IH, Yale C, Thomas R, Garrett C, Winter RM (1996) Three sibs with microcephaly, congenital heart disease, lung segmentation defects and unilateral absent kidney: a new recessive multiple congenital anomaly (MCA) syndrome? Clin Dysmorphol 5: 129-134.