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CASE REPORT

F. Linke · W. Kraemer · M. Ansorge · R. Brzezinska
S. Berger

Right esophageal lung in a preterm child with VACTERL association and Mayer-Rokitansky-Küster-Hauser syndrome

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Abstract We report on a preterm girl (birth weight 1,200 g) with a right esophageal lung in esophageal atresia type VIag (according to the extended classification of Kluth). Additionally, the child suffered from an atrioseptal defect, a dextrocardia with a left descending aorta, a duodenal atresia, a high type of anal atresia (VACTERL association), agenesis of the left kidney, and agenesis of the vagina, uterus, and ovarian tubes (Mayer-Rokitansky-Küster-Hauser syndrome or incomplete MURCS association). The child was treated with an emergency gastrostomy because of increasing abdominal dilatation. Thereafter, the parents refused further surgical treatment, and the child was maintained on basic therapy. After an uneventful period of 4 weeks, the child died of an acute massive aspiration. This case shows that sufficient spontaneous ventilation is possible in esophageal lung as long as a gastrostomy is kept on suction to prevent overinflation of the affected lung and the stomach. Ethical aspects have to be considered when treatment is planned in cases of prematurity and associated malformations when a chance of good survival is rather limited. The stepwise approach as proposed in the present case appears to be the only possible therapeutic regimen that can be offered in this complicated condition.

Keywords Esophageal atresia · Esophageal lung · VACTERL association · MURCS association · Mayer-Rokitansky-Küster-Hauser syndrome

F. Linke (✉) · S. Berger
Department of Pediatric Surgery,
Johannes Gutenberg University,
Langenbeckstrasse 1, 55131 Mainz, Germany
E-mail: frank.linke@chiru.med.uni-giessen.de
Tel.: +49-641-9944705
Fax: +49-641-9944709

W. Kraemer
Neonatal Intensive Care Unit,
Department of Pediatrics,
Johannes Gutenberg University, Mainz, Germany

M. Ansorge
Department of Pediatric Pathology, Institute of Pathology,
Johannes Gutenberg University, Mainz, Germany

R. Brzezinska
Pediatric Radiology Unit, Johannes Gutenberg University,
Mainz, Germany

Introduction

Communicating foregut malformations have been reported as rare variants of esophageal atresia. Esophageal lung belongs to this group of communicating bronchopulmonary foregut malformations, representing an extremely rare abnormality. Treatment of this condition, if recognized at all preoperatively, has mainly been unsuccessful. The right lung is predominantly affected by this malformation. A proper preoperative diagnosis is difficult to make because only an esophageal and tracheobronchial contrast study is able to reveal the exact pathologic anatomy. Once the diagnosis is established, individual treatment can be planned and usually has to include resection of the affected esophageal lung. Long-term survival has been reported for some children, but for the majority, survival was not possible.

Case report

A 32-week, 1,200-g preterm baby girl was delivered by cesarean section because of preterm labor, the first child of healthy, nonconsanguinous parents. Prenatally, a dilated stomach was diagnosed by ultrasound, and a duodenal atresia was suspected. The child was stable with an oxygen flow after birth and spontaneous ventilation. The initial chest x-ray demonstrated a hyperinflated right lung, a normal left lung, and dextrocardia (Fig. 1). The stomach was air-filled; the rest of the abdomen was airless. Long-gap esophageal atresia (EA) with lower tracheoesophageal fistula (TEF) and duodenal atresia were diagnosed. After intubation, an emergency gastrostomy was performed

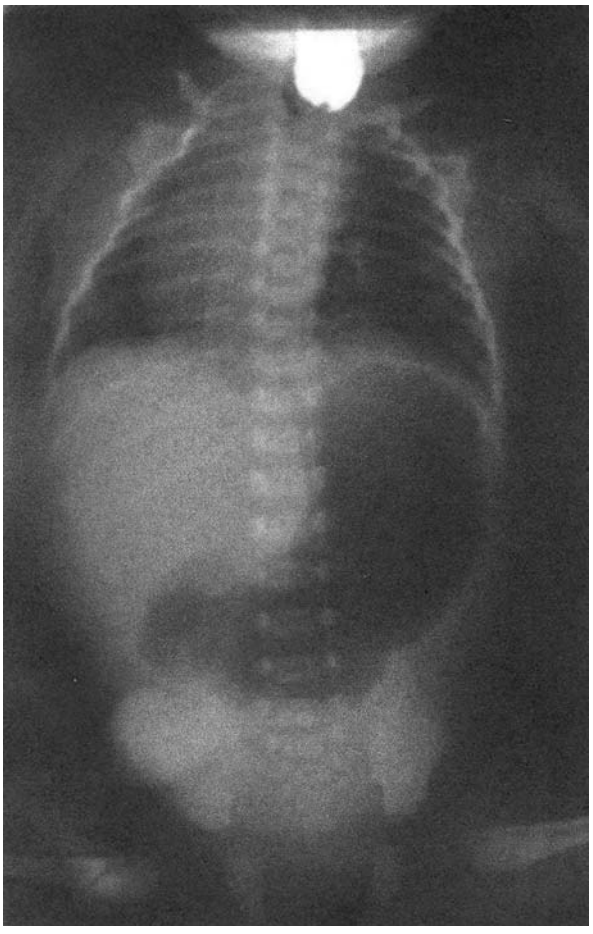

Fig. 1 Initial radiogram showing the dilated proximal esophageal pouch filled with contrast fluid and the dilated, air-filled stomach together with an otherwise gasless abdomen

in the neonatal intensive care unit to prevent gastric rupture. Thereafter the infant's general condition stabilized.

When a contrast study was performed the next day via the gastrostomy to evaluate for the lower esophageal pouch, contrast fluid could be seen in the trachea and right main bronchus (Fig. 2). Therefore, contrast fluid was also administered via the tracheal tube, and the diagnosis of a communicating bronchopulmonary foregut malformation was made. A tracheal stenosis, as first suspected from the contrast study, was not confirmed by the postmortem analysis (Fig. 3a). By ultrasound, a left kidney could not be identified, and dextrocardia with a left descending aorta and atrioseptal defect (ASD) were confirmed. High anal atresia without a visible fistula was also present. Vertebral and limb anomalies were not found. The diagnosis of VACTERL association was made. Treatment was proposed as a multistep approach including (1) a catheter jejunostomy and colostomy to allow for enteral feeding and growth of the child, (2) resection of the right lung with esophageal end-to-end anastomosis and duodenoduodenostomy, and (3) correction of the ARM. At this point, the parents (both medical professionals) rejected any further surgical or intensive care treatment. A do-not-resuscitate order was written, and the child was extubated and maintained on continuous suction to the gastrostomy, sequential suctioning of the trachea, and total parenteral nutrition in the neonatal intensive care unit.

For 4 weeks the infant's clinical course was uneventful, and she gained 300 g of weight. Several episodes of bradycardia were treated by stimulation, tracheal suctioning, and oxygen flow administration. Unexpectedly, the child succumbed after 4 weeks from a massive aspiration. Chromosomal analysis was performed and showed normal findings. Upon autopsy, the diagnosis concerning the intrathoracic and gastrointestinal anatomy was confirmed (Fig. 4). Additionally, a MURCS-like association (Mullerian duct aplasia, renal agenesis, anomalies of the cervicothoracic somites) was found that included aplasia of the vagina, uterus, and both ovarian tubes. Left renal

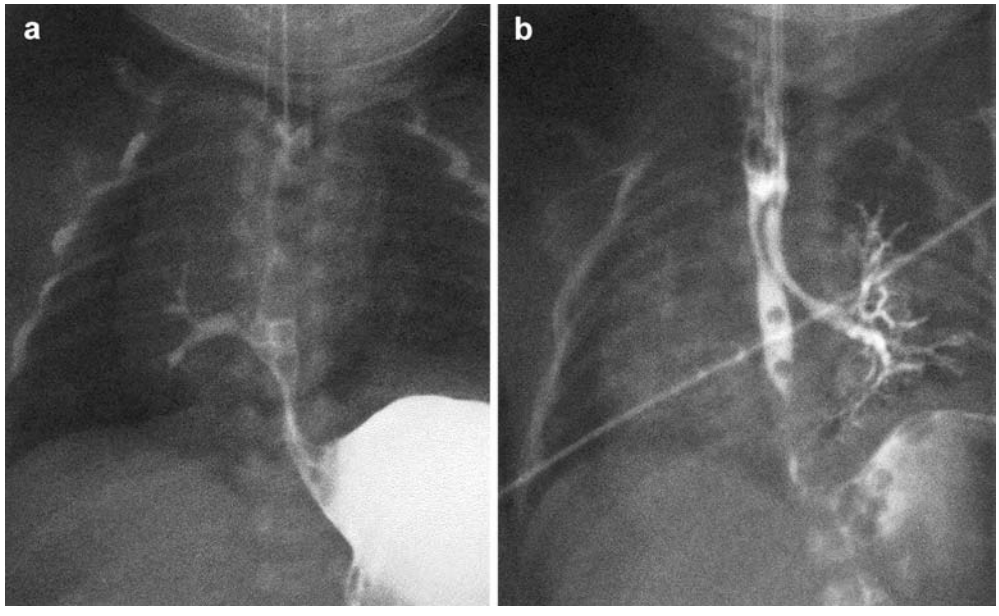

Fig. 2 Contrast study via the gastrostomy (**a**) demonstrating the origination of the right main bronchus from the lower esophageal pouch. When contrast fluid was subsequently administered via the intubated trachea (**b**), a high carina with a missing right main bronchus and a lengthy left main bronchus were found. Instead of a right main bronchus, the lower esophageal pouch was connected to the carina. The right lung was hyperinflated because air could pass through the lower esophageal pouch during inspiration but was trapped in the right lung when the upper part of this esophageal segment collapsed during expiration. Gas was expired from the right lung in part through the gastrostomy

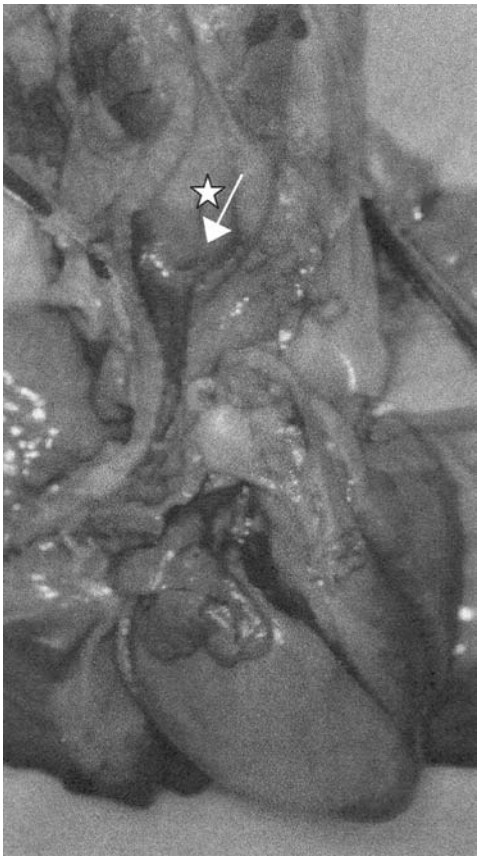

Fig. 3 Postmortem findings: dorsal view of the larynx-lung-esophagus block (**b**). The photograph shows the trachea at the level of the carina. A broad opening into the tracheoesophageal fistula is visible (*arrow*), and a small bud (*asterisk*) instead of the (aplastic) right main bronchus (**b**). The right lung was unilobar, and the left lung was bilobar. The right main bronchus originated from the distal esophageal segment (see Fig. 4). Tracheal stenosis was not present

aplasia was confirmed. Instead of a left renal artery, only a fibrous band was found, and a left ureter was not seen. Cervicothoracic vertebral defects, however, were not present.

Discussion

Classification of the combined malformations

The current case is the 10th case of an esophageal lung in combination with an esophageal atresia [1, 3, 8, 10, 11, 14, 15, 17, 19]. In eight of these previous cases, the esophageal lung was right-sided; only recently was the first case of a left-sided esophageal lung reported [14]. In the group of children with additional major malformations [1, 14, 17], none survived. Among those cases with additional tracheal stenosis, only one has survived [11]. Besides tracheal stenosis (four cases), the following additional malformations have been reported so far: anorectal malformation (two cases [1, 17]), cardiac malformation (one case with ASD [17] and one case with tetralogy of Fallot [14]), and esophageal varices (one case [1]). Although some characteristic malformations of VACTERL syndrome are included in the above, an association with VATER or VACTERL has been reported in esophageal lung only in a few cases [1, 5, 10]. An association of esophageal lung with Mullerian duct aplasia has not been reported yet. The MURCS association [6] has, however, been described in children with VATER/VACTERL-like associations [4, 13]. Abnormal blastogenesis during the 3rd–4th week of embryogenesis has been accused of causing MURCS as a developmental field defect [2]. A differentiation between VACTERL and MURCS is not clearly possible in the present case; rather, a combination of both malforma-

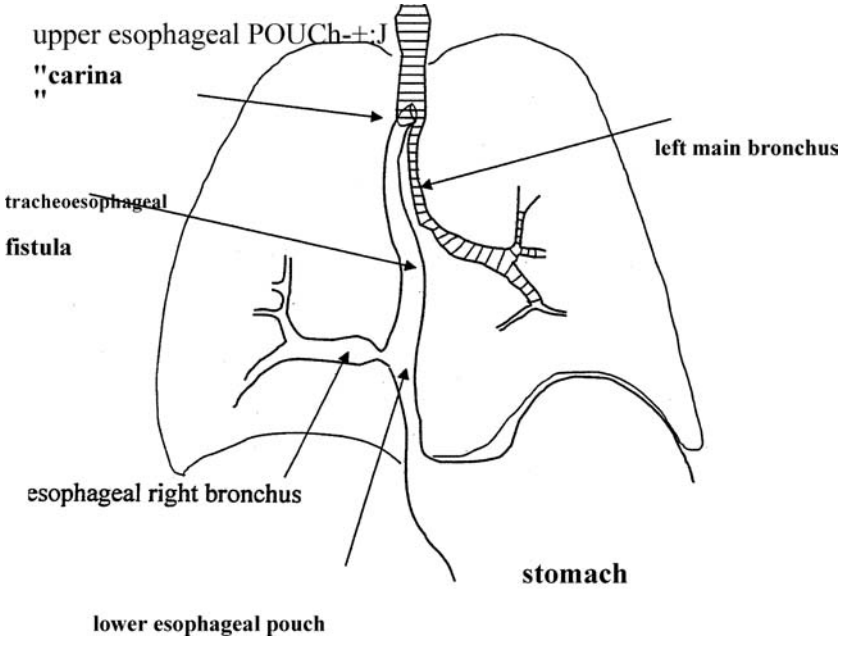

Fig. 4 Schematic drawing of the pathologic anatomy

tion patterns has to be assumed here. Alternatively, a combination of VACTERL association and Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome might be suggested to name the present findings correctly. A similar combination of malformations was previously described in a girl with laryngeal stenosis, EA with TEF, imperforate anus, duodenal atresia, and MRKH syndrome [16]. In summary, the current case represents the first description of associated esophageal lung, VACTERL, and MRKH syndrome. Simultaneously, it is the second case of VACTERL and MRKH, and the 10th case of esophageal lung in esophageal atresia reported in the literature.

Prognosis and ethical considerations

According to the currently used prognostic classifications for esophageal atresia, the present case would be classified in the highest risk group of both the Waterston and Montreal classifications [18]. Thus, even without considering the esophageal lung, a mortality of 40–50% was to be expected. A preterm infant with a number of major anomalies in addition to the right esophageal lung and EA may therefore be seen as a worst-case scenario of this condition. All children who survived an esophageal lung for more than 10 days (a total of four survivors, three long-term survivors) were born at term and received a right pneumonectomy. This surgical option was considered unsuitable for a 1,200-g baby of 32 weeks' gestation. Furthermore, operative access to the dorsal mediastinum would have been complicated due to dextrocardia and a left descending aorta, a condition comparable to a right descending aorta in cardiac situs solitus [7]. The same applied for a trial of bronchotracheal reconstruction as reported in esophageal bronchus without EA [12]. Therefore, the only possible treatment was maintaining the gastrostomy and performing a jejunostomy and a colostomy to allow for enteral feeding until the child had grown to a weight that would make a right pulmonectomy and subsequent correction of the esophageal and duodenal anatomy feasible. An ethical evaluation of the situation was made when the parents refused to give consent to any further surgical treatment.

Duodenal atresia, unilateral renal agenesis, MRKH syndrome, and dextrocardia had not yet been reported as additional malformations in combination with esophageal lung. Because none of the previous children with additional major malformations survived and because the child was affected by prematurity plus several additional malformations, the chance of survival was considered poor even with maximum therapy.

It is concluded that a stepwise treatment including gastrostomy followed by resection of the esophageal lung and esophageal anastomosis may offer a chance for survival in esophageal lung and EA–TEF when diagnosis is made preoperatively and the child is not affected by severe additional malformations or significant prematurity. Conversely, when a number of major malformations accompany this pathology, as is the case in VACTERL associations, ethical considerations have to be made according to the child's prognosis and expected quality of life [20].

References

1. Bleicher M, Melmed A, Bogaerts X (1983) Vater association and unrecognized bronchopulmonary foregut malformation complicating anesthesia. Mt Sinai J Med 50:435–438
2. Braun-Quentin C, Billes C, Bowing B, Kotzot D (1996) MURCS association: case report and review. J Med Genet 33:618–620
3. Chiba T, Ohi R, Hayashi Y, Uchida T (1989) Bronchopulmonary foregut malformation in 3 infants—with special references to cases in childhood. Z Kinderchir 44:105–108
4. David A, Mercier J, Verloes A (1996) Child with manifestations of Nager acrofacial dysostosis, and the MURCS, V ACTERL, and pulmonary agenesis associations: complex defect of blastogenesis? Am J Med Genet 62:1–5
5. Davies R, Kozlowski K, Wood B (1989) Right-upper-lobe esophageal bronchus (with VATER anomalies). Am J Dis Child 143:251–252
6. Duncan P, Shapiro L, Stangel J, Klein R, Addonizio J (1979) The MURCS association: Mullerian duct aplasia, renal aplasia, and cervicothoracic somite dysplasia. J Pediatr 95:399–402
7. Harrison M, Hanson B, Mahour G, Takahashi M, Weitzman J (1977) The significance of right aortic arch in repair of esophageal atresia and tracheoesophageal fistula. J Pediatr Surg 12:861–868
8. Keeley J, Schairer A (1960) The anomalous origin of the right main bronchus from the esophagus. Ann Surg 152:871–874
9. Kluth D (1976) Atlas of esophageal atresia. J Pediatr Surg 11:901–919
10. Leithiser R, Capitanio M, Macpherson R,Wood B (1986) "Communicating" bronchopulmonary foregut malformation. Am J Roentgenol 146:227–231
11. Matsumoto Y, Ohi R, Hayashi Y (1995) Right pneumonectomy syndrome. Surg Today 25:278–280
12. Michel J, Revillon Y, Salakos C, Blic JD, Jan D, Beringer A, Scheinmann P (1997) Successful bronchotracheal reconstruction in esophageal bronchus: two case reports. J Pediatr Surg 32:739–742
13. Orstavik K, Steen-Johnsen J, Foerster A, Fjeld J, Skullerud K, Lie S (1992) V ACTERL or MURCS association in a girl with neurenteric cyst and identical thoracic malformations in the father: a case of gonosomal mosaicism? Am J Med Genet 43:1035–1038
14. Saydam T, Mychaliska G, Harrison M (1999) Esophageal lung with multiple congenital anomalies: conundrums in diagnosis and management. J Pediatr Surg 34:615–618
15. Schwartz D, So H, Anban T, Creedon J (1983) Total lung sequestration in association with oesophageal atresia and multiple tracheo-oesophageal fistulae. Z Kinderchir 38:410–411
16. Scorpio R, Beals D, Powell R (1998) Vaginal agenesis discovered during repair of high imperforate anus: operative management. Am Surg 64:1066–1069
17. Takahashi A, Matsuyama S, Suzuki N (1989) Esophageal atresia associated with pulmonary agenesis: report of two cases and review of the literature. J Jpn Pediatr Soc 25:282–289
18. Teich S, Barton D, Ginn-Pease M, King D (1997) Prognostic classification for esophageal atresia and tracheoesophageal fistula: Waterston versus Montreal. J Pediatr Surg 32:1075–1080
19. Toyama W (1972) Esophageal atresia and tracheoesophageal fistula in association with bronchial and pulmonary anomalies. J Pediatr Surg 7:302–307
20. Ward C, Myers N (1999) Babies born with major disabilities: the medical-legal interface. Pediatr Surg Int 15:310–319